Investigating the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta

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Abstract

Osteogenesis imperfecta (OI) is a complex condition which requires specialist care. This care is readily available to children but is not available for adults. There are also limited transition services for people with OI, leaving them without support during transition. This novel research study was aimed at exploring the lived experience of transition, the resulting self-management, and aimed to deliver a resource, providing information and support.

This thesis comprises three phases: a qualitative phase, a quantitative phase, and a resource development phase. The qualitative phase primarily involved a focus group and two interviews, to explore issues raised in the literature, including the lack of knowledge and the lack of support for adults with OI. This phase revealed adults with OI struggle to access information and frequently experience dismissal or doubt from healthcare professionals (HCPs), due to a lack of knowledge among HCPs. As a result, adults with OI need to engage in self-management and advocacy, despite the lack of necessary information or skills.

The quantitative phase involved an online semi-structured survey aimed at extending our understanding of the qualitative findings but for the wider OI population. The findings were consistent and suggested a connection between missed fractures and poor HCP knowledge. Finally, we iteratively developed a resource that reflected the needs of the OI community, garnering feedback at key points.

Despite the small sample sizes, this research has implications for practice and policy, providing a resource which can fill the knowledge gap, support adults with OI and educate HCPs. Future research should explore the efficacy of the developed resource for OI in specific settings. Finally, as a result of findings, a new theoretical model of self-management was developed, future research should further test the model and consider its applicability in the context of other chronic conditions.

Dedication

For mam, you have supported me and encouraged me from day one. Without you I would not have been able to get to this point. You have pushed me to be the best I can be and allowed me to exceed every expectation.

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I also want to thank the Brittle Bone Society who have supported me throughout this journey and have never wavered in their efforts to help me achieve my goals.

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Declarations

This work has not previously been accepted in substance for any degree and is not being concurrently submitted in candidature for any degree.

Signed..... .....  .....

Date.....15/09/2023.....

This thesis is the result of my own investigations, except where otherwise stated. Other sources are acknowledged by footnotes giving explicit references. A bibliography is appended.

Signed.....  .....

Date.....15/09/2023.....

I hereby give consent for my thesis, if accepted, to be available for photocopying and for inter-library loan, and for the title and summary to be made available to outside organisations.

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Date.....15/09/2023.....

The University's ethical procedures have been followed and, where appropriate, that ethical approval has been granted.

Signed.....  .....

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Chapter 1 – Osteogenesis Imperfecta

1.1 Introduction

The primary objective of this thesis is to investigate the transition from paediatric to adult services of people with osteogenesis imperfecta, specifically how this is managed and the effect of the process on people with osteogenesis imperfecta. Transition to adult services is an important issue for people with chronic conditions including osteogenesis imperfecta given the complex and systemic nature of osteogenesis imperfecta. In addition to investigating the transition to adult services, this thesis will also investigate the ongoing care of adults with osteogenesis imperfecta. This will include exploring the principles of self-management and expert patients.

This chapter will explore the complex nature of osteogenesis imperfecta and will provide the context needed for the thesis and the discussions of osteogenesis imperfecta. This will include discussing the various types of OI, diagnosis of the condition, existing treatments and current services available for people with OI. This will be important for the review of the existing literature and the discussions of the needs for people with osteogenesis imperfecta during their transition to adult services. An understanding of OI and the wide range of effects is vital for this thesis as there will be extensive discussion on the management of OI during the transition to adult services and in order to understand the needs during this time, an understanding of the condition as a whole is necessary.

1.1.1 What is Osteogenesis imperfecta?

Osteogenesis imperfecta (OI), also known as brittle bone disease is a metabolic bone disorder which affects the manufacturing of collagen in the body (Rauch & Glorieux, 2004). Collagen is an abundant protein in the body and when it is not formed correctly or is in insufficient quantities, the impacts on the functionality of the body is significantly affected (Shoulders & Raines, 2009). OI affects approximately one in every 15,000 people in the UK, this means that there are around 5000 people in the UK with OI (Brittle Bone Society, 2022).

OI occurs in people who have a genetic mutation in one of the genes which control the manufacture of collagen. There are many different types of OI, and they differ in presentation

and severity. There have been 21 types identified through genetic testing (Genetics Home Reference, 2020). However, without testing being readily available, most people diagnosed with OI will identify with either types I, II, III or IV, they are the four original types which were first documented by Sillence et al. (1979). Severity of OI has also been described using the terms mild, moderate and severe in lieu of the types as the presentation within the type groups can vary greatly. The use of these terms can be seen in as early as 1975 (Bauze et al., 1975), however the types described by Sillence et al. (1979) have been dominant in the literature and many still use those terms today. There is greater flexibility with these terms as individuals can be more descriptive and distinguish how their OI affects them without having to go into immense detail, through combination of terms such as 'mild-moderate' or 'very severe'. This would be beneficial both when describing their condition to lay people but also in appointments with healthcare professionals' (HCPs) who may not be familiar with the classification system laid out by Sillence in 1979.

The OI type classification system involves dividing individuals based on their symptoms and general presentation. Type I is the mildest type of OI and is the most common, it accounts for roughly half of all cases (OIF, 2020). Type II is the only lethal form of OI, the lungs are underdeveloped, and infants often only live for a matter of minutes or hours (Ayadi et al., 2015). Type III is a severe form of OI, those with this type will experience a high number of fractures, will require multiple surgeries, and will likely need to use a wheelchair (Sinikumpu, 2015). Type IV is a moderate form of OI and falls between type I and type III in terms of number of fractures (Rauch & Glorieux, 2004).

The remaining types of OI vary in severity and presentation of cases of OI can vary greatly even among those who have been diagnosed with the same type of OI. Table 1 shows the different types of OI, the genetic mutations associated with them and the severity of disease that is experienced with that type. Some information on severity of rare types remains unclear, as a result, the severity of those types has not been added to the table (Genetics Home Reference, 2020).

90% of OI cases are the result of dominant mutations and many of the newer types are rarer forms of the condition (Cohen, 2014). This is due to the genes causing those types of OI being recessive and therefore two copies of the gene are required in order for the disease to

present (Byers & Pyott, 2012). If only one copy of the mutation is present, the person will be a carrier but will not be affected by the condition (Griffiths, 2012, p. 57). Five of the 21 types are caused by dominant mutations, this means that only one copy of the gene will result in the condition occurring. Dominant mutations can also occur spontaneously, this would be due to mutations occurring during the production of gamete cells (Griffiths, 2012, p. 582). These mutations were not present in the genes of either parent.

Table 1: OI types, pattern of inheritance, affected genes and severity of the condition

Type	Inheritance	Gene	Severity	Reference
I	Dominant	COL1A1/2	Mild	(Pollitt et al., 2006)
II	Dominant	COL1A1/2	Lethal	
III	Dominant	COL1A1/2	Severe	(Augusciak-Duma et al., 2018)
IV	Dominant	COL1A1/2	Moderate	
V	Dominant	IFITM5	Moderate	(Hanagata, 2016)
VI	Recessive	SERPINF1	Moderate	(Homan et al., 2011)
VII	Recessive	CRTAP	Severe	(Tang et al., 2020)
VIII	Recessive	P3H1	Severe	(Barbirato et al., 2015)
IX	Recessive	PPIB	Severe	(van Dijk et al., 2009)
X	Recessive	SERPINH1	/	(Shapiro, 2014)
XI	Recessive	FKBP10	/	(Kelley et al., 2011)
XII	Recessive	SP7	Moderate	(Fiscaletti et al., 2018)
XIII	Recessive	BMP1	Severe	(Xu et al., 2019)
XIV	Recessive	TMEM38B	Variable	(Ramzan et al., 2021)
XV	Recessive	WNT1	Moderate	(Fahiminiya et al., 2013)
XVI	Recessive	CREB3L1	Severe	(Keller et al., 2018)
XVII	Recessive	SPARC	/	(Mendoza-Londono et al., 2015)
XVIII	Recessive	TENT5A	Severe	(Gewartowska et al., 2021)
XIX	X-Linked	MBTPS2	Severe	(Lindert et al., 2016)
XX	Recessive	MESD	/	(Moosa et al., 2019)
XXI	Recessive	KDELR2	/	(van Dijk et al., 2020)

Timing and method of diagnosis of OI often varies depending on the severity of the case and whether the case is the result of a spontaneous mutation or has been inherited from a parent. If one of the parents has OI, health professionals would know to look specifically for signs of OI. More severe types such as types III and IV are commonly diagnosed before birth (National Organization for Rare Disorders, 2020). There are a number of signs which can be identified on ultrasound scans including fractures, bowing of long bones and the presence of a high number of wormian bones in the skull (Thompson, 1993). Wormian bones are small bones which form in the suture lines of the bones of the skull. A high number of these small bones would suggest the presence of OI (Semler et al., 2010). The diagnosis could be confirmed with amniocentesis which can be performed around 20 weeks gestation (GARD, 2018).

In mild cases of OI, there are often no visible signs on ultrasounds and children would not show signs of OI until they begin walking and consequently start experiencing fractures. In these cases, the presence of OI could be confirmed with x-rays, bone density scans and skin or bone biopsies (NIH, 2016). If severe cases were not diagnosed prenatally, postnatal diagnosis could be made via a physical examination and any of the other aforementioned methods.

Symptoms of OI are not limited to fractures as the name Brittle bone disease would suggest. The lack of collagen or weakened collagen affects the whole body, as a result, OI must be seen and treated as a systemic condition (Martin & Shapiro, 2007). The most common signs and symptoms associated with OI are chronic pain, bowed limbs, scoliosis, short stature, bruising and blue sclera (Marini et al., 2017). People with OI can also suffer with dentinogenesis imperfecta, which means their teeth are very weak and prone to breaking in a similar manner to their bones (Glorieux & Rowe, 2012). In addition to these common signs and symptoms, OI also presents an increased risk of respiratory problems including a higher risk of pneumonia, increased risk of kidney stones, increased risk of cardiovascular disease and much higher risk of hearing loss (Thiele et al., 2012). Hearing loss occurs in around 50% of OI cases and is not dependent on the severity of OI (Swinnen et al., 2012).

There is no cure for OI and therefore the goal of health professionals is to manage the condition and prevent fractures and other complications associated with the condition. For

children with the condition, treatment may include surgeries to place intramedullary rods or nails in the long bones such as the tibias and femurs of the legs (Mulpuri & Joseph, 2000). This would allow the bones to be stabilised and prevent displacement in the event of fracturing. Although placing the rods would be difficult without first correcting the bow in the long bones which is common, especially in severe cases. Once placed, intramedullary rods or nails would help to prevent future bowing and would increase mobility (Georgescu et al., 2013).

Pharmaceutical treatment of OI is limited to analgesics to manage acute pain caused by fractures and chronic pain. Bisphosphonates are also a common therapy for children with OI to increase bone mineral density (Antoniazzi, 2010). However, bisphosphonates are given to children more readily than to adults as some research has suggested that these therapies can lead to osteonecrosis of the jaw and increase the risks of infection after operations and other procedures (Glorieux, 2007).

People with severe forms of OI will often rely on the use of a wheelchair as the deformities can be very severe and prevent them from walking (Ralston & Gaston, 2020). People with milder forms of the disease may use a wheelchair when they suffer fractures but when they are not recovering from fractures may be able to walk unaided (Montpetit et al., 2015).

This discussion has made it is clear that OI is a complex condition, and this condition will affect individuals throughout their lives. This shows how important it is that OI is understood by both individuals with OI and the HCPs who are responsible for the treatment plans offered to people with OI, as the condition is systemic, and the effects are not limited to only bones which the name 'Brittle Bone Disease' may suggest.

This section of the chapter has given an introduction to OI as a chronic condition and has provided the foundational information which will be drawn upon throughout this thesis.

1.1.2 Existing Services and Guidance

This section will discuss the current services available for people with OI in the UK and the current guidance which is in place for HCPs regarding the management of OI.

Given the wide range of symptoms and complications that are associated with OI which have been discussed in this chapter, the management of OI relies on a multidisciplinary approach and the ability to identify issues early is imperative. For children it is common for them to be kept under the care of an orthopaedic consultant at all times, as this gives them a point of contact who understands the condition and would be able to intervene and act in a proactive manner. There are six specialist OI centres for children with OI in the UK (Brittle Bone Society, 2021). These are located in London, Bristol, Birmingham, Sheffield, Glasgow and Belfast. These centres offer specialist care which is administered by a multidisciplinary team (MDT). There are no specialist centres for adults with OI in the UK. As a result, once people reach the age of 16, they can be discharged and have to rely on standard adult services. These services often have extremely long waiting lists and people struggle to find a health professional who is familiar with their condition (Wilson et al., 2005). They would also not be able to receive multidisciplinary care and as a result in order to receive the care they need, would have to be under the care of multiple health professionals from a number of specialities (Marr & Seasman, 2020). This would be both time consuming and overwhelming. Given the complexities of their condition, it is essential that people with OI are prepared for the move from paediatric to adult services.

OI is a complex condition and requires a comprehensive approach which is currently lacking in the UK. Comprehensive management has been shown to improve patient outcomes by preventing complications and this can improve quality of life which is vital in a condition which can be extremely debilitating (Ibrahim & Crockard, 2007). Adults with OI will then rely on consultants who may have limited OI knowledge, these consultants would be reliant on guidance when prescribing medications or recommending other treatment options. However, at the time of writing, there is no OI specific guidance available from the NHS Wales website, the National Institute of Health and Care Excellence or from the NHS Health A-Z website. On the NHS England website, guidance for OI in adults is limited to guidance regarding Teriparatide for the treatment of osteogenesis imperfecta (adults) (Specialised Commissioning Team, 2016).

OI is also mentioned in a number of guidance documents from NHS England. Firstly, the guidance for paediatric rheumatology services, it is mentioned that OI is managed by rheumatology teams but does not offer a source of information regarding OI (NHS England,

2013e). Secondly, the adult rheumatology service document also mentions OI as a rare condition to be managed by rheumatology services (NHS England, 2013c). This document offers a source of information for managing OI in the form of a link to a page from the Osteogenesis imperfecta foundation (OIF). However, upon investigation, at the time of writing in March of 2022, it was found that this link does not work. The third piece of guidance from the NHS is specific to treating OI in children and discusses how the specialist centres should conduct their care of children with OI (NHS England, 2013a). This document is comprehensive, fully explaining OI and the possible comorbidities but limits the discussion of treatment to children. It also briefly mentions transition to adult services but does not go so far as to instruct HCPs in how the handover between services should occur, instead it simply states when this should occur.

Reference to OI was not found in either the guidance for metabolic disorder services or the specialised orthopaedic services (NHS England, 2013b, 2013d). Despite OI being a metabolic bone disorder, which is known to lead to recurrent fractures and surgeries. When searching the NHS Wales website for OI information, only one document can be found (NHS Wales, n.d.). This document appears to be geared towards parents of children with OI and gives information about OI generally and explains the benefits of physiotherapy. It is not clear where this document originated, and it is not dated.

This section has shown that information regarding OI can be difficult for HCPs to access. This brings into question the availability of information and guidance for how the transition process should be managed and also brings into question the level of support available for individuals with OI.

1.1.3 OI Charities and Organisations

There are a number of organisations around the world which provide information about OI and offer support to people with OI, this section will describe these organisations and discuss their roles in the management of OI and how they function within the wider OI community.

The Osteogenesis imperfecta foundation (OIF) is the organisation based in the USA, the UK organisation equivalent to this is the Brittle Bone Society (BBS). This charity was established over 50 years ago and offers support, information and is a vital resource for people with OI,

their families and other parties who may benefit from support and knowledge of the condition, such as people who are in positions where they have to care for people with OI. In addition to being a point of contact, the website is a source of information and knowledge. The information is found in the form of factsheets which can be downloaded by anyone, no login is required. There are also links to videos on YouTube, the majority of the videos on their YouTube channel are recordings from conferences. The videos have also been organised into playlists to make it easier for people to find videos specific to their needs. The availability of videos ensures that more people are able to get information on OI, some may struggle to read the vast amount of information provided in the factsheets.

There are 14 factsheets available on the website at the time of writing, eight of which are indicated as being exclusively designed for parents of children with OI or for staff within schools responsible for those children with OI. Four of these factsheets are aimed entirely at providing guidance for schools. One for nurseries or preschools, the second for primary schools, the third for secondary schools and the fourth provides information regarding participation in physical education by children with OI. Of the remaining six factsheets, only one deals exclusively with information designed to aid adults living with OI. None of the factsheets are intended for use by carers or partners of adults with OI.

While the six general factsheets do include information for adults with OI, the majority of the information is still geared towards children, or rather, those caring for children with OI. Whether they be parents/ guardians or healthcare professionals. The factsheets can be found both in the support section of the website, under the information and resources heading and the sections which is indicated as being “For Professionals”. However, the same factsheets are displayed in both areas of the website. There are no differences in the information offered.

The factsheets themselves do offer a great deal of information, however, this information is difficult to read due to the sheer volume of text on each page. Although people with chronic conditions should be regarded as being experts in their own condition, these factsheets would not be easy to understand or process. Between the 14 factsheets, there are 66 pages of information. While it is true that not all of this information would be relevant to everyone with OI, it can be expected that many people with OI or carers / partners / parents would attempt to take in all of this information.

As this is also designed for HCPs, it can be assumed that some HCPs would want to read some of this information in order to ensure they are able to provide appropriate care for their patients. However, it is unclear whether there would be opportunity for HCPs to read the factsheets due to the limited time for appointments and the overall demand on services within the NHS.

This section of the chapter has explored the role that charities and organisations play in the management of OI. In lieu of guidance from governing bodies which was previously discussed, these organisations attempt to fill the gap in information and support for people with OI.

1.1.4 Reflection

My interest in this topic is the result of personal experience of OI, as a result, OI has been the topic of choice in previous academic pieces of work. The psychosocial impacts of OI were the focus of a narrative review for a master's dissertation and this review highlighted issues with both the transition from paediatric to adults' services and the management of OI in adults. One of my key findings from the narrative review, was that an effective transition from paediatric to adult services had a positive psychosocial impact as it lessened the strain on the people with OI during this transition. One of the recommendations of that dissertation was further research into the impacts of OI especially in the adult population due to the paucity of information available regarding OI in adults. The current research study was influenced by these findings and recommendations and will build on the narrative review conducted for the master's degree. Personal experience of OI and the issues related with this will be beneficial for the research as it will allow a deeper understanding of the experiences other people with OI have had. However, the personal experience with OI may lead to unintentional biases and it will be important to counteract these in order to ensure the research and the findings will be of consequence.

1.2 Literature Review

In order to research transition in OI and self-management by adults with OI, it was important to establish what research has been conducted and what information was available to both people with OI and HCPs. In order to understand the current knowledge base and to identify any gaps in the literature, a review of the current literature was undertaken. Identifying any

gaps in the current knowledge of OI management and the transition process to adult services was essential and would guide this research by identifying under-researched areas and topics of interest. There were a number of methods which could have been utilised when conducting the review of the literature, these are a narrative review, a scoping review and a systematic review. These methods were compared in order to decide which would be most suitable for this research. The first method which was examined was a narrative review. This type of review summarises the existing evidence but does not explore the gaps in the current literature (Henry, 2018). Although this method allows for evidence to be discussed, it is not as rigorous as alternative methods and unlike scoping reviews or systematic reviews, it is not as structured and does not involve as critical appraisal of the evidence (Sukhera, 2022).

The second method which was examined was a scoping review. This method is utilised when researchers are attempting to identify gaps in the current literature. When conducting a scoping review, researchers will begin with a research question and is the chosen method for literature review at the beginning of a research study (Mak, 2022). Scoping reviews allow for research questions to be quite broad, unlike for systematic reviews which require more specific research questions (Pollock, 2021).

Systematic reviews were the final method of literature review explored for this research. These are comprehensive reviews of the current literature, using both published and unpublished data. These aim to draw broad theoretical conclusions which are linked to existing theory and evidence (Siddaway, 2019). As previously mentioned, these reviews are also guided by research questions, but for systematic reviews, these questions are rigid and may restrict the research (Dehkordi, 2021).

Following this comparison of the methods for a literature review, it was decided that a scoping review would be the most appropriate type of literature review for this research. This is due to the use of scoping reviews for identifying gaps in literature, this was essential for this research as this research was novel. In addition, the scoping review offers more flexibility as the research question could be relatively broad. This section will discuss the how the literature review was conducted and the findings of this literature review.

1.2.1 Search Strategy

A search of the literature was conducted using multiple databases, which were; CINAHL Plus Full Text, MEDLINE, PsycArticles, PsycInfo, ASSIA, British Nursing Index and Web of Science Core Collection. These databases were selected from the university database collection and were selected as they focused on areas such as social sciences, nursing and social care, health literature, behavioural science and mental health. All of these fields needed to be reviewed in order to obtain a comprehensive understanding of the current body of literature and to identify any gaps in the knowledge of this complex condition.

These databases were searched between the 8th of October 2019 and the 27th of January 2020. Alerts were set up on the databases to ensure that any newly published papers could be included in the literature review. Literature was checked throughout the research to ensure no new research was missed, any publications relating to OI published after the literature review was conducted was not relevant to the research.

Initially the search was limited to publications between 2000 and 2020, however, it was felt that research published prior to 2010 may no longer be relevant so, the search dates were amended to literature published between 2010 and 2020. Earlier than 2010 there were no additional publications relating to transition in OI. The exploration of this issue on people with OI is limited to newer publications hence the requirement that publications must have been published after 2010 in order to be included in this review.

In addition to searching databases for relevant literature, a search for grey literature was also conducted. This included searching the National Institute for Health and Care Excellence website for guidelines on OI, the Cochrane database and the ProQuest dissertations and theses global collection.

1.2.2 Search Terms

Searches of databases included the use of the Boolean functions and therefore the terms “and” in addition to “or” were utilised. This was done to maximise the number of results and ensure relevant publications were not missed. The terms used for all searches to identify publications discussing OI were “osteogenesis imperfecta” OR “OI” OR “brittle bone disease”.

Each database was searched twice, once to identify publications relating to the management of OI and once to identify publications relating to the transition from paediatric services to adult services. In addition to the OI terms, the terms used to find publications relating to the management of OI were “management” OR “managing” OR “manage”. The second wave of database searches, in addition to the OI terms, the search term “transition” was used.

Appendix 1 shows the terms that were used, and the searches conducted on the databases. It also shows the number of results found in the databases and how these results were narrowed down before completing the review.

1.2.3 Inclusion criteria

In order to be included in the review, the publications had to be available in English, had to discuss OI, management of OI or the transition of people with OI from paediatric to adult services. Publications should have a study sample including adults and/ or adolescents with OI. There were no restrictions set on where the publications could be published meaning all countries were included. This was done to maximise the potential results and it was not felt to be inappropriate as some countries may have practices in place which could be implemented in the UK.

1.2.4 Exclusion criteria

Any publications that were not available in full text or English were excluded. Publications were also excluded if they failed to discuss OI at all or only discussed genetics, biomechanics or other similar areas which are not relevant to the focus of this research. Publications were also excluded if they were published prior to 2010 as they may not reflect current practices or latest scientific knowledge. Publications were also excluded if the study sample was exclusively children with OI.

1.2.5 Search process

Figure 1 shows the process which led to the selection of publications to be included in the review.

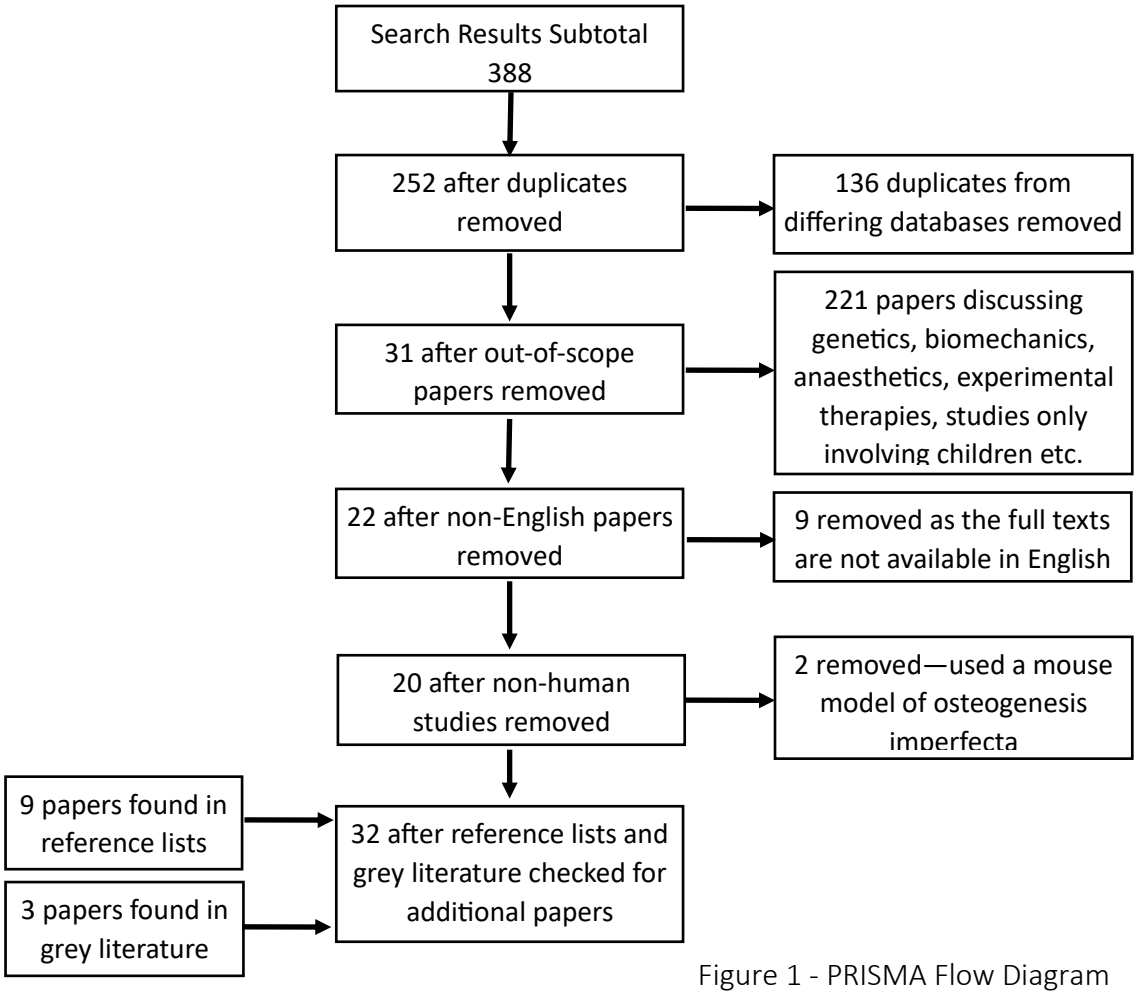

Figure 1 - PRISMA Flow Diagram

1.2.6 Geographic distribution of publications

Of the 32 articles included in the literature review, six were conducted in the USA and six were conducted in the UK. This is followed by Canada with five studies. Two studies were conducted in each of the following countries; Denmark, France, Italy and Sweden. One study was conducted in each of the following countries; Belgium, Greece, Japan, Norway, Spain, Switzerland and the Netherlands. Of the six conducted in the UK, three were guidelines for healthcare. Two of which were published by NHS England and one by the National Rheumatoid Arthritis Society.

1.2.7 Quality Assessment

The quality of the included studies was assessed using the critical appraisal skills programme (CASP) checklists. This method was selected as there are different versions of the CASP checklist suitable for the various types of studies. This allows for appropriate questions to be posed and ensures the different methods are respected and analysed appropriately.

The only publications found in the literature review that were not assessed with CASP checklists were the guidelines. There was no appropriate checklist for these documents and as a result, any identified limitations of these guidelines have been included in lieu of a quality assessment.

1.2.8 Overview of methodological critique

29 studies had their quality assessed with one of the CASP checklists (CASP, 2018a, 2018b, 2018c, 2018d). Of the 29 studies, only two were systematic reviews. There were 15 described as either narrative or literature reviews. Although literature reviews are useful for providing a summary of the existing knowledge in a given field, they do not provide any new data or improve the understanding of a topic. As a result, although the findings of the studies are insightful, their results may not be transferable.

Of the 29 studies, 14 were empirical studies which collected data and did not rely on existing evidence from the literature. There were ten cohort studies, two qualitative studies and two case studies.

1.2.9 Theoretical critique

None of the 29 studies discuss the theoretical frameworks used when designing the study or during the analysis.

1.2.10 Thematic Analysis

The majority of included papers focus on OI in adults or adolescents. Although there is a wealth of research for children with OI, those studies do not fall within the scope of this research as they were not focused on the transition from paediatric care and are instead focused on the treatment of OI in children. After assessing the quality of each of the papers

included in the literature review, a thematic analysis was conducted. This involved reviewing the focus of each of the papers and their key findings. Papers were then grouped according to these findings and this allowed for the identification of the main themes. The themes identified were; transitioning to adult services, management of OI, and living with OI. The focus of the studies within the management of OI theme was varied, as a result, the studies were sub-categorised and this led to the identification of the following sub-themes; general OI management, multidisciplinary team management, audiological management, cardiovascular management, pregnancy management and management guidelines.

1.2.10.1 Transitioning to adult services

This theme relates to the transition from paediatric to adult healthcare services. This is an important step for people with chronic conditions as their care would be handed over to new doctors and other healthcare professionals (Nieboer et al., 2014). There have been a number of studies conducted looking into the transition for other chronic conditions, including a number of studies on the transition of people with diabetes. There has been little research done on this aspect of OI and as a result the only evidence regarding the transition included in this literature review comes from Canada and the USA. While these countries do have different healthcare systems, the findings of this research may still have relevance for people with OI in the UK. Given the paucity of evidence, the findings of studies within this theme may still be beneficial not only during the transition from paediatric to adult services but also for future research in this field.

All of the studies found that knowledge plays an integral role in the transition. Carrier et al. (2018) was described as a knowledge-synthesis study which looked at the results of seven articles. The sample included children with OI (n=1548), adults with OI (n=254), parents of children with OI (n=14) and hospital staff (n=20). Three of the studies did not disclose the sample size or characteristics. This study found that young people with OI want to be educated, not only on their condition but also on healthcare services and self-care management. Allowing young people to take control of their care would allow for the development of expert patients and enable people to become empowered (Cordier, 2014). This would not only allow patients to have a say in their care but would also allow for collaboration between patients and healthcare professionals allowing them to design

treatment plans together that will help patients to achieve their goals and achieve what they want out of life (Wensing, 2000). This would also be beneficial to their quality of life and provide better outcomes (Donaldson, 2003).

Enabling people with OI to take control of their care allows them to take responsibility for their health and allow them to identify potential barriers (Shapiro & Germain-Lee, 2012). The article by Shapiro & Germain-Lee (2012) is an observational report and has used existing literature to identify the needs of people with OI during the transition to adult services. This article found that developing the skills needed to identify barriers will allow them to develop the skills needed to overcome these barriers in adulthood when they would be expected to be more independent. The presence of a transition tool was also found to lead to the development of these skills by Carrier et al. (2018). The tool developed and discussed by Carrier et al. (2018) includes 11 sections and provides a summary of the care given to the person with OI during their time in paediatric services and ensures health care professionals (HCPs) understands the needs of the person with OI.

The absence of these skills would be detrimental to people with OI and is highlighted by Dogba et al. (2014). This was a qualitative study drawing on data gathered through case studies, interviews, observation and review of documents. This study was conducted in Canada, as was the review conducted by Carrier et al. (2018). Dogba et al. (2014) found that people who do not have these skills had a tendency to struggle with the responsibility associated with self-management. This study also found that people who did not have a supported transition to adult services would often miss appointments as they were not accustomed to managing this aspect of their care prior to being moved to adult services.

Another key finding in these papers was the need for continuity of care. Both Dogba et al. (2014) and Shapiro & Germain-Lee (2012) found that there was a need for transition services to facilitate continuity between paediatric and adult services. This continuity includes ensuring treatments are not interrupted and identifying HCPs within adult services to take over the provision of care before the patient is discharged from paediatric services. This would require HCPs from both services to communicate and allow for a smooth and coordinated transition. This may be difficult to achieve in the UK as waiting lists in adult services are often very long and therefore achieving this continuity may not be possible. This

continuity and preparation was seen to ease anxiety for people with OI (Carrier et al., 2018). The benefit of continuity is not limited to providing reassurance and easing anxiety to people with OI but it would also be important in ensuring the health and function is maintained (Shapiro & Germain-Lee, 2012). Lapses in any treatment, could threaten the functionality of people with OI and compromise their independence.

While the studies by Carrier et al. (2018), Dogba et al. (2014) and Shapiro & Germain-Lee (2012) all have overlapping findings and support the need for a transition programme, Jeong et al. (2019) presents a very different method for enabling the transition from paediatric to adult services. Jeong et al. (2019) presents a tool to facilitate care transitions. This study was also conducted in Canada, was a knowledge-synthesis study and included 12 studies of the "Good2Go My Health Passport" and other similar tools. The "Good2Go My Health Passport" is a resource for people with chronic conditions and is not limited to OI. It allows people to enter their details and a brief medical history, and this can then be given to new HCPs and provide them with an overview of their condition. This would be beneficial in emergencies and provides HCPs with only the crucial information, the need for preparation for emergencies was discussed by (Shapiro & Germain-Lee (2012). Consideration of emergencies is an essential part of OI management as fractures can be a common occurrence.

In summary, a supported transition to adult services is essential for people with OI. This support is needed to ensure people with OI have the necessary skills to manage their care as adults and to ensure that there is continuity of care between paediatric and adult services.

1.2.10.2 Management of OI

The second theme identified in the literature review is the management of OI. As previously stated, due to the complex nature of OI as a chronic condition, there are several sub-themes within this theme. The first sub-theme, general OI management, includes the studies which discuss the management of OI as a whole. The second sub-theme, multidisciplinary team management, focuses on the benefits and drawbacks of collaboration in the management of OI. The third sub-theme, audiological management, includes studies which focus solely on the management of hearing loss in people with OI. The fourth sub-theme is cardiovascular management which includes a study focused on the abnormalities of the heart which are

associated with OI. The fifth sub-theme is pregnancy management, which includes studies describing the outcomes of pregnancy in women who have OI. The sixth sub-theme is management guidelines, which includes documents aimed at improving management of OI and other conditions which have been published by either the NHS or charities.

1.2.10.2.1 General OI management

There were nine studies identified in the literature search which explored the management of OI as a whole.

One of the main points seen in this sub-theme is the need for care to be tailored to the needs of each person. Gil et al. (2017) argue in a narrative review that people with OI must be assessed and a treatment plan must be designed to meet their needs as cases, as even in those with mutations of the same gene, can have dramatically different presentations. Gil et al. (2017) also state that this need for custom-made treatment plans supports the need for MDTs. This links back to the multidisciplinary teams' theme and the benefits that this method of treatment can have. The need for individually tailored care is also supported by the findings of Tournis & Dede (2018), who discuss the use of physical therapy in people with OI. The use of physical therapy is vital for OI as exercise can prevent osteoporosis which would further weaken the bone. The narrative review by Tournis & Dede (2018) argue that exercise regimes developed by physiotherapists have to meet the needs and abilities of the person with OI.

A common treatment for OI is bisphosphonates, which aim to increase bone density and reduce the fracture rate. These are commonly administered to children but less so to adults (Dwan et al., 2016). It is argued by Bishop & Walsh (2014), that this is due to a lack of research regarding the efficacy of bisphosphonates in adults. The lack of evidence regarding bisphosphonates for adults with OI was also discussed by Lindahl et al. (2014) in their literature review. Tournis & Dede (2018) concluded that although bisphosphonates may be indicated following a fracture, given the lack of evidence regarding efficacy in preventing fractures, they are contraindicated for adults with OI. There have been some reports of osteonecrosis of the jaw (ONJ) occurring as a result of prolonged use, however, Tournis & Dede (2018) stated that in the studies they reviewed, there were no cases of ONJ. However,

these studies were on children and adolescents and therefore that may not be the case for adults with OI.

Both Forlino et al. (2011) and Marini et al. (2017) included lengthy discussions on the genetics involved with OI and the subsequent issues with the manufacture of collagen in the body. Following this, Forlino et al. (2011) discussed the neurological complications which have been associated with OI in their literature review. These include; macrocephaly, hydrocephalus and basilar invagination. Macrocephaly refers to an abnormally large head (Williams et al., 2008). Hydrocephalus is a disorder of the cerebrospinal fluid which causes an increase of intercranial pressure which can cause brain damage if untreated (Kahle et al., 2016). Basilar invagination can also result in brain damage and is the result of the spine prolapsing into the skull (Smith et al., 2010). These are aspects of the condition which have not been mentioned in any of the other studies included in this review. This may suggest that they are exceedingly rare occurrences in OI but the lack of information about the risks suggests a need for further research.

Following the discussion on genetics, Marini et al. (2017) goes on to discuss both the need for a smooth transition from paediatric to adult services and the affect that OI has on quality of life. This links in two of the previous themes discussed and highlights the importance of comprehensive management of OI as issues such as transition and quality of life are intrinsic to the successful management of OI. Marini et al. (2017) also states that screening for complications associated with OI is crucial for adults with OI as it would allow for early intervention. This screening for complications should include dentinogenesis imperfecta, which is a weakening of the teeth and can lead to teeth breaking more easily. This complication is common in OI and is mentioned in several of the studies in this sub-theme, including; Forlino et al. (2011), Marini et al. (2017), Gil et al. (2017) and Hald et al. (2018). The study by Hald et al. (2018) found that dentinogenesis imperfecta occurs in 25% of OI cases and is more likely to occur in severe cases of OI.

Finally, a narrative review by Stewart (2015) reports on the common treatments, the genetic factors and comorbidities associated with OI. There are three central points made by this study. Firstly, the need for MDTs to manage the care of adults with OI. Secondly, the need for a smooth transition from paediatric to adult services. Finally, the lack of specialist services in

the UK for people with OI. This paper highlights that there are only four specialist services in England and that they are key for facilitating collaboration and successful management of OI.

The key finding of this sub-theme is the need for individually tailored care as the needs of people with OI can vary greatly and in order for the condition to be successfully managed, there cannot be a one-size-fits-all approach to management of OI.

1.2.10.2.2 Multidisciplinary team management

Multidisciplinary teams (MDTs) involve an integrated approach between HCPs from various disciplines. The goal of this approach is patient centred care with a focus on the needs of the individual (Goodwin et al., 2013). As OI is a systemic condition, the use of MDTs was discussed in a number of articles. This sub-theme includes two papers, one from the UK and one from Switzerland. In addition to these two papers which were focused solely on the management of OI with MDTs there are some references to papers which are featured in other themes due to their comments on MDTs. They were not included in this theme as their primary focus was not MDTs but their insight into their use is still important in this discussion.

Both of these articles were narrative reviews but the papers differed in their discussion. Bregou et al. (2016) did not have a specific or focused aim but offered an in-depth discussion on the therapies available to people with OI, such as bisphosphonates, surgical corrections and physiotherapy. The review also discussed the research into cures for the condition and the classification of the types of OI. Whereas, Marr et al. (2017) offered less discussion on therapies such as bisphosphonates and surgical correction and focused more on the use of physical therapies, psychology, speech therapy and social work. Although the aspects of MDTs they focused their attention on varied, their conclusions were remarkably similar.

Both Bregou et al. (2016) and Marr et al. (2017) found that the use of MDTs for the management of OI led to improved health outcomes and a better quality of life. The use of MDTs allows for consideration of the wider determinants of health which is vital for managing a chronic condition Marr et al. (2017). These determinants of health include factors such as living conditions, education, social and community networks, work environment and healthcare services (Green et al., 2015, p. 88).

In addition to allowing for the consideration of determinants of health, the use of MDTs also allows for people with OI to get involved with their care. Marr et al. (2017) found that one of the benefits of using MDTs was the mutual respect given to both the HCPs and the patients and this allowed them to get involved and become empowered. This is in a similar manner to the use of transition programmes which also allowed for people with OI to take control of their care.

The main benefit of using MDTs for a condition like OI is the ability for HCPs from different specialities to work collaboratively (Marr et al., 2017). This would ensure that recommendations made by one of them would not contradict that of another as people with OI would usually have to go to multiple appointments, one with each of the HCPs and follow their recommendations. Due to this reason there is widespread support for the involvement of MDTs for people with OI as evidenced in the following studies; Bishop & Walsh (2014) argued that it is especially important in severe cases, Gil et al. (2017) state that MDTs are needed as treatment plans for OI must be designed on a case-by-case basis, Díaz López et al. (2018) highlighted the need for MDTs for people who suffer with spinal abnormalities and Lafage-Proust & Courtois (2019) describe MDTs as crucial for people with OI.

In addition, the benefits of MDTs was also reported by both Carrier et al. (2018) and Dogba et al. (2014) who found that the transition from paediatric to adult services was improved when it was managed by an interprofessional team. This allowed the systemic nature of the condition to be respected and ensure that all aspects of the condition were managed (Monti et al., 2010). It is clear that the need for MDTs for people with OI has been well established and need to be in place for adults with OI as their care is often complex and their needs will be unique.

The need for MDTs is also discussed by Lafage-Proust & Courtois (2019). This narrative review highlights the importance of consulting with different HCPs in order to achieve positive health outcomes. In addition, it discusses the network in place in France. This network is known as OSCAR and allows HCPs from across the country to collaborate in the care of people with rare bone, calcium, and cartilage diseases.

This sub-theme supports the need for MDTs for OI and highlights that the use of MDTs can not only improve care but also improve the quality of life of people with OI.

1.2.10.2.3 Audiological management

Four studies were identified which discuss the hearing loss associated with OI. These papers varied both in quality and in country of origin. Two came from the USA and Denmark. There is one from Belgium and one from France.

All four of the studies state that hearing loss occurs in OI, however, there is disparity over the incidence of hearing loss. The literature review by Carré et al. (2019) states that hearing loss occurs in between 2% and 94% of cases, while this range does not provide any clarity, other information offered in this paper does give insight into who is most at risk of suffering hearing loss within the OI population. It was found that those with type I OI had the highest rates of hearing loss and those with type IV had the lowest rates of hearing loss. This finding of increased hearing loss rates in mildly affected individuals was also observed by Hald et al. (2018). This study also found that not only is hearing loss more common in type I, but the hearing loss is usually severe in type I.

The cohort study by Swinnen et al. (2012) found that hearing loss occurs in 52.2% of OI cases. This study included 182 people with OI, included both males (n=88) and females (n=94) and included the common types of OI. However, type I accounted for 152 of the participants, this means the type was greatly overrepresented in the study. The incidence of hearing loss in this study varies greatly from those reported by Carré et al. (2019). This may be due to differences in the methods utilised by the researchers, specifically, the threshold at which hearing loss is diagnosed is not the same for all studies. These discrepancies were discussed in Machol et al. (2020), who in their study set the threshold at 20db. They compared this with other studies, where there were five studies set at 15db and one study set at 20db. As a result, it would be extremely unlikely that the results would corroborate.

The study by Machol et al. (2020) included a comparison between the types of OI and age of hearing loss onset. This then showed how the rates of hearing loss in the different groups changes with age. Firstly, in both the under ten age group and the 10-29 age group, type III OI saw the highest rates of hearing loss. However, in the 30-49 age group, type I then see the

highest rate of hearing loss. If the studies by Carré et al. (2019) and Hald et al. (2018) conducted the same comparisons, it is conceivable that it would be possible to be more certain about the group most at risk of developing hearing loss. Furthermore, the study by Swinnen et al. (2012) found that the mean age of hearing loss was 21.3 years and if hearing loss has not occurred by age 40, the likelihood of hearing loss as a complication of OI is reduced.

There is some disagreement between the studies regarding the effect of gender on hearing loss in OI. The study by Swinnen et al. (2012) found that there was no difference in the incidence of hearing loss in men and women. Whereas, the study by Machol et al. (2020) found that hearing loss occurred in 24.8% of women and 17.8% of men. This difference may be the result of a larger number of women in the study compared to men, 182 to 130 respectively or this may be the result of ascertainment bias in the study. Ascertainment bias may have occurred as people who were already known to have hearing loss were recruited for the study and therefore the results of the study may not be a true reflection of the target population (Spencer & Brassey, 2017).

Hald et al. (2018) found that although hearing loss is a known complication of OI, 20% of hearing loss requiring treatment goes untreated. This highlights the need for screening, so that hearing loss can be identified early so as not to compromise quality of life. All four of the studies in this sub-theme highlight the need for regular hearing screening in people with OI.

In summary, this sub-theme shows that hearing loss is common in OI and although the incidence rates vary between the studies, all studies agreed that the risk of hearing loss of people with OI justifies the need for frequent audiological screening so hearing loss can be identified and treated early.

1.2.10.2.4 Cardiovascular management

The third sub-theme of management is focused on the cardiovascular management of OI. There was one study identified which focused solely on the complications and abnormalities associated with the cardiovascular system of people with OI (Radunovic et al., 2011). This was a described as a clinical and echocardiographic survey and was conducted in Norway.

This study found that people with type III OI were more susceptible to cardiovascular abnormalities such as increased left ventricular dimensions and mass. This increase was in comparison to both a control group and the other types of OI (types I and IV). Radunovic et al. (2011) suggest that this may be the result of the mutated collagen gene. As a result, those with severe OI have very weak collagen and weakened structures in their cardiovascular system.

This makes cardiovascular screening extremely important, especially in those with severe types of OI. This need for screening was also identified by Matsushita et al. (2020) who emphasized the need to find serious comorbidities early to allow for timely intervention.

This sub-theme shows that cardiovascular problems have been associated with OI and that in order to identify these problems early, screening of people with OI is required.

1.2.10.2.5 Pregnancy management

The fourth sub-theme of management is pregnancy management. There were two studies identified which explored pregnancy in women with OI.

The first paper was a case study of a 28 year old woman with type I OI living in Italy (Cozzolino et al., 2016). The paper states that as the woman had type I OI she had a high-risk pregnancy. As a result, a MDT approach was needed, providing further support for the use of MDTs in OI management. Despite being high risk, the findings emphasised the need for decisions regarding mode of delivery to be made on a case-by-case basis. This would allow for women with OI to have some control over their choice of delivery. This need for tailored care was also reported by Yimgang & Shapiro (2015) in their cross-sectional study of 274 women with OI living in the USA. However, this study did emphasise that the tailored care should involve giving the women as much information as possible so they can make an informed decision.

Yimgang & Shapiro (2015) also explored the rates of caesarean sections in women who had received genetic counselling. Those who had received genetic counselling were 1.3 times more likely to have a caesarean section compared to women who did not have genetic counselling. This might suggest that women who are aware of all the risks are more likely to opt for the safer option or are persuaded by their doctors to take this option. Although this

finding is insightful, it may not be transferable to the UK given that the USA has much higher rates of caesarean sections than the UK (Boerma et al., 2018). The higher rates of caesarean sections may also be the result of a global trend which sees an increase in caesarean sections, Wise (2018) states that in the UK the number of women having caesarean sections has increased from 19.7% in 2000 to 26.2% in 2015. Additional research is needed in order to get an accurate representation of the rates of caesarean sections among women with OI in the UK.

Yimgang & Shapiro (2015) also found that the rates of complication seen were high, with 26% of women reporting to have suffered pre-eclampsia. According to the NHS (2018), preeclampsia occurs in around 6% of pregnancies. This suggests that women with OI have an elevated risk of pre-eclampsia. This was also seen in the study by Cozzolino et al. (2016). Despite the high risks, both studies agreed that having a severe form OI does not necessarily mean that complications will arise. However, both also agreed that although mode of delivery should be decided on a case-by-case basis, caesarean sections are more likely, and women need to be aware of the risks and be prepared for their mode of delivery to change.

The key finding of this sub-theme echoes the findings of general management of OI sub-theme, that is that care and treatment plans for people with OI must be individually tailored to meet their needs. In addition, although there is a higher risk of complications in women with OI during pregnancy, women must be allowed to have their say and be able to choose their method of delivery.

1.2.10.3 Guidelines

The final sub-theme within the management theme is focused on the guidelines which have been published regarding the care and management of OI. Three sets of guidelines were identified during a search of the grey literature.

The first of these guidelines was published by NHS England and is aimed at the management of children with OI under the care of a specialised service (NHS England, 2013a). Although the inclusion criteria of the literature search meant that studies which only discussed the care of children were removed, this set of guidelines has been retained within the review as some of these guidelines pertain to the transition of people with OI from paediatric to adult health

services. In addition, this set of guidelines is only for England and as a result may not apply to all the devolved nations within the UK. Until recently, there were no specialised services for children with OI in Wales and they were instead referred to English centres. These guidelines may be relevant for both England and Wales as they were produced when there were only centres in England. There was an OI service set up in Cardiff recently and now children with OI in Wales can be referred there instead, as this is a relatively new service there is little information available. It was discovered that this service was in place in Cardiff through connections made at an international conference in September 2022.

These guidelines describe the process that should be undertaken when discharging a child from paediatric to adult services. This includes a review in the clinic, this will determine whether the child is discharged at 16 or 18 years of age. Upon being discharged the person with OI would be passed onto a consultant with an interest in osteoporosis. Although osteoporosis is a condition which results in weakened bones, the treatment of OI requires a HCP with knowledge of the condition.

The second set of guidelines was also published by NHS England and is aimed at the management of adult rheumatology services (NHS England, 2013d). OI is often classed as a rheumatological condition and as a result, OI is mentioned in these guidelines. However, the discussion on the management of OI is severely limited. The guidelines specifically for management of OI is limited to a hyperlink from the Osteogenesis Imperfecta Foundation which is based in the USA. In addition, the hyperlink no longer works. Despite this, there are sections of these guidelines, which may be applicable to OI, including but not limited to the continuity of care which states that complications secondary to a condition should not require re-referral and as a result sub-speciality clinics may be needed in order to improve patient outcomes. These guidelines also acknowledge both the need for MDTs to successfully manage chronic conditions and the need for a smooth transition from paediatric to adult services.

The final set of guidelines identified from the grey literature was published by the National Rheumatoid Arthritis Society, as aforementioned, OI is often categorised with rheumatology conditions and therefore this document includes information relating to the management of OI (NRAS, 2016). This document only mentions OI once, but it does discuss the transition of

children with rheumatological conditions from paediatric services to adult services, however, these services are only available in Cardiff and Bangor. For adults with rheumatological conditions, it also states that there is a need for MDTs to treat rheumatic and musculoskeletal disorders and that being seen by an MDT early is the key to successful treatment.

The key findings of these guidelines are that, although there are guidelines for children with OI in England and OI is mentioned in some guidelines for rheumatology services, there is a lack of guidance regarding care of adults with OI for HCPs to utilise.

1.2.10.4 Living with OI

This theme is primarily focused on the impact that OI has on the life of an individual. This includes the effect on quality of life, functionality and experience that results from living with OI. There were seven papers relating to this theme in the literature review. Although the topics in this theme do not directly relate to transition to adult services, the effects of the condition on the person and their life may affect how individuals manage their condition. This is relevant to this research as adult care and self-management were identified as areas of interest from the outset of the research.

Quality of life was the primary focus of three of the studies in this theme. The first of the three, Balkefors et al. (2013) was a cross-sectional study with 29 participants, both male (n=11) and female (n=18). However, types of OI were not disclosed and therefore it is unclear whether results are transferable to the whole OI population. This study found that people with a mild to moderate type of OI experienced a reduced health related quality of life. The findings indicated that this was the result of a lower physical ability, however, despite this reduction in ability, many participants reported that they met or exceeded the recommended level of daily exercise. With this in mind, context of quality of life cannot be ignored. It is possible that the participants were rating their level of exercise highly as they did not compare their activity to the general population but instead to what they consider to be their capabilities. So, although the level of activity is high for them given their condition, for a healthy person it may be very little.

Hald et al. (2017) also explored quality of life in cohort study of 84 adults, all common OI types were included, and the population also included both males (n=38) and females (n=46).

It was found that the physical component score (PCS) of Short Form (36) Health Survey (SF36) was significantly lower in adults with OI than the general population. This study also observed that OI has a considerable effect on the physical health related quality of life and a small effect on the mental aspect of the health related quality of life. This correlates with the findings of Balkefors et al. (2013), which also found that OI does reduce physical abilities. This fits with the description of OI as a debilitating and progressively worsening condition. The reason for the mental aspect not being considerably reduced was stated to be due to a higher level of satisfaction from achievements in education or work which can increase in life satisfaction. This finding was also observed by Balkefors et al. (2013).

A further cross-sectional study by Matsushita et al. (2020) with 40 adults with OI found that quality of life can be improved through treatment such as surgery. The findings did not show that surgery would improve the PCS of the SF-36 but instead it was found to improve the mental component score (MCS) of the SF-36. This is likely due to the fact that surgical intervention can reduce the severity of fractures and thereby reducing pain and recovery time. The PCS was found to be negatively affected by surgery, specifically in the lower extremities. This is likely due to the severity of the OI and the associated fractures rather than the surgeries themselves.

Quality of life was also reported but was specifically in relation to fatigue in OI by Harsevoort et al. (2020). This cross-sectional study used the fatigue severity scale to investigate the prevalence and effects of fatigue in the OI population. It was found that people with OI do suffer with fatigue to a greater degree than the general population. Harsevoort et al. (2020) stated that fatigue can impact quality of life but as people with OI often suffer with chronic pain, it is unclear how much fatigue actually impacts quality of life.

The impact of pain on quality of life is addressed by Nghiem et al. (2018). This integrative review addressed the pain experience faced by adults with OI and found that pain can have a detrimental impact on quality of life in multiple ways. Firstly, the direct effect that pain has on quality of life is the limited physical capabilities of the person. Secondly, the indirect effect that pain has on quality of life is the result of inhibiting social interaction and ability to participate in social activities. Pain is not discriminatory, it is found in all age groups and the effects that pain has on quality of life are profound (Katz, 2002). In a pilot study by Tosi et al.

(2019) it was observed that there are people with OI receiving treatment for pain in all age groups and found that OI does result in pain interference meaning that the pain they experienced interfered with their mood and daily activities and pain behaviours, such as avoidance (Philips & Jahanshahi, 1986).

Finally, a systematic review investigated the psychosocial experience of people with OI (Tsimicalis et al., 2016). This review included both adults with OI (n=175) and children with OI (n=610). This study showed that OI does have an impact on the psychosocial wellbeing of people with OI. Not all of the findings from this study will be applicable to this research as the majority of the sample was children with OI but as this is the only study investigating the psychosocial experience of people with OI identified in this literature review and some of the findings could be still useful to this research. This is due to the fact that chronic conditions can have an impact on the psychosocial wellbeing of an individual and as a result, poor management of chronic conditions can influence the quality of life of individuals. This cannot be overlooked when considering the management of OI and the transition to adult services. The main finding of this study was that people with OI obtain a great deal of satisfaction from education and work-related achievements. This was reported in both the studies by Hald et al. (2017) and Balkefors et al. (2013). This helps to explain why often the mental component score in the SF-36 of people with OI is not significantly different from the general population.

In summary, the key findings of studies within this theme are that the effects of OI are not limited to the health of an individual and it can have an impact on quality of life. The literature also indicates there is a need for individuals to be equipped with the information and knowledge to self-manage their condition due to the conditions complex and systemic impact on individuals. In addition, pain is common in OI and managing it is paramount, as pain has been seen to inhibit quality of life. The literature also shows that there is a lack of understanding of the impact of OI on adults and there is a paucity of evidence regarding the transition from paediatric services for individuals with OI. In addition, the literature which does exist is focused on the transition in the USA and Canada. There has not been prior research into this transition in the UK. This highlights the need for research into this field and supports the aim and objectives which will be discussed later in this chapter.

1.3 Discussion

This chapter has explored OI as a chronic condition, including the various types of OI, their symptoms and treatments. This chapter has also discussed the current literature on OI and lack of literature discussing the transition to adult services. This exploration of the literature also revealed the lack of information for adults with OI. This literature showed there is a paucity of knowledge for the management of OI in adults. This presents a number of challenges both for individuals with the condition and for the HCPs too.

The issue of adjusting to adult services was discussed in the literature and particular attention was paid to the impact of this culture shock to individuals. Dogba et al. (2014) stated that the shift in responsibility from the parent to the child can lead to fewer appointments being attended. This could have a detrimental effect on the health of the individual and contribute to the development of comorbidities. In addition to the worsening of the health status of individuals, this incidence of missing appointments can have a detrimental effect on the NHS. In the UK, missing appointments given by healthcare professionals in the NHS could be very detrimental. Often there are policies in place which allow for people who miss appointments regularly to be discharged. Although this policy is understandable given the cost incurred by missed appointments to the NHS, which was £216 million per year in NHS England alone (NHS England, 2019). This effect of missed appointments extends beyond the economical but also impacts the health of patients. Those people who are discharged then have to go back to the bottom of the waiting list before they could be seen, this delay could lead to complications and worsening health (NHS England, 2020).

The importance of education for individuals was observed repeatedly in the literature and the findings in the literature supported the argument that people with OI would benefit from being educated and empowered which is also reported in the transitioning to adult services theme. The study by Tosi et al. (2019) found that people with OI want to be informed and have as much information about their condition and treatments as possible. The possession of such knowledge would enable self-management which was discussed as an important method of management for OI. Although the need for education would likely be seen across the OI population, the sample in the study by Tosi et al. (2019) was skewed towards females and lacked ethnic diversity. They state that further research is needed and that a more diverse

group would be beneficial to the understanding of the impact of OI, the incidence of comorbidities, the impact on quality of life and the other needs of individuals with OI including education.

The literature also emphasised the need for expertise from HCPs and many of these studies reference the importance of multidisciplinary team management, this was discussed in the multidisciplinary team management sub-theme. However, in order for this to occur, HCPs need to be educated in OI to ensure they have the requisite knowledge to successfully work in an MDT responsible for treating someone with OI. The need for HCP education is not discussed in the literature and the literature also does not discuss how these MDTs should be set up or the manner in which they should operate. The lack of knowledge among HCPs is discussed by Lindahl et al. (2014) and Marini et al. (2017), however neither of these discuss how these issues can be resolved. This lack of knowledge is also reflected in Tosi et al. (2015) who discussed how this can lead to a misunderstanding of what OI is and the implications of the condition on the health of individuals. They state that it is known predominantly as a condition leading to fractures. They acknowledge the limitations of existing literature but do not discuss how to better educate HCPs or how to address these misconceptions of OI.

One of the existing tools which has been used to facilitate the transition to adult services is the 'Good2Go My Health Passport', as discussed earlier in this chapter, this tool might be beneficial if used in conjunction with a more comprehensive transition programme as it does not address the other aspects of the transition and the problems that this transition presents. Carrier et al. (2018) discussed the use of the 'Good2Go My Health Passport' and stated that this tool was limited and did not provide enough information to HCPs to be considered comprehensive. The limitations of this tool were acknowledged by Jeong et al. (2018) in their study of the tool, as the tool had not been tested, there was no evidence to support the efficacy of the tool. There is a lack of other resources other than the 'Good2Go My Health Passport' to support individuals both during and after their transition to adult services. This would suggest that there is a need for new resources or tools to be development to provide this support and to fill this existing gap in service provision.

The discussions in this chapter have led to the development of the following objectives which will be used to achieve the overarching aim of the thesis which is, to investigate the transition

from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta. The objectives of this thesis will be broken up into two groups; objectives relating to transition and objectives relating to adult care and self-management. The transition objectives are as follows;

1. To gain an in depth understanding of the transition from paediatric to adult services for people with osteogenesis imperfecta.
2. To identify any changes in care given between paediatric and adult services from the perspective of adults with OI.

The objectives for adult care and self-management are as follows;

1. To develop an understanding of the ongoing care needs of adults with osteogenesis imperfecta in the UK from their perspective.
2. To identify any gaps in knowledge of people with osteogenesis imperfecta.
3. To develop a guide for adults with osteogenesis imperfecta to assist them in navigating the healthcare system in order to facilitate their access to the necessary services.

The following chapter discusses differences between qualitative and quantitative research, explores the potential benefits and drawbacks of a mixed methodology approach for this research and will discuss the existing models and frameworks which will be used to frame the research and the discussions of the topics.

Chapter 2 – Methodologies

2.1 Introduction

The previous chapter introduced OI as a chronic condition and explained the purpose of this research and the aim and objectives that this research would attempt to address. This chapter will cover the theoretical positioning of the research. Firstly, this chapter will discuss different epistemological approaches and will also include a discussion and evaluation of existing theories and models which could have been used in this research. Secondly, this chapter will explore research methodologies and explore the various methods which could have been undertaken. Thirdly, this chapter will also serve to explain the choices that were made and will explore the benefits and drawbacks of the methods that were selected.

2.2 Epistemological Approach

This section will focus on the epistemological approaches which could have been utilised, as there are a number of different epistemological approaches which could have been taken for this research. Epistemology in research refers to the way in which knowledge is gathered and where is it obtained from, this will alter the perspective of the researcher and will influence the interpretation of the data. The epistemological approaches which can be used are positivism, post-positivism, and constructivism (Tenny et al., 2022). Positivism, as previously discussed, seeks to explain associations or relationships between events (Park et al., 2020). Whereas post-positivism is based on the understanding that certainty is not possible and instead attempts to find majority agreement in opinions and views (Panhwar et al., 2017). Constructivism is the belief that the view and perspectives a person has are influenced by their experiences (Martí, 2022). Each of these approaches offers different values, advantages, and disadvantages. In order to select the appropriate methodology for this research, these epistemological approaches were reviewed and assessed. This section will explore these approaches and present the arguments behind the decision in selecting one of these approaches.

The first approach reviewed was positivism. This approach seeks to explain events and behaviours so that future events and behaviours can be predicted (Bryman, 2012, p. 28). This

is based on the principle that human beings are predictable and relies on the ability of the research to control the environment and other variables (Park et al., 2020). This would not be suitable for behavioural or social science research as it involves exploring issues objectively, and positivism does not allow for experiences to be considered from an individual point of view (Hasan, 2016). Based on this and given that OI is a rare condition, with a wide variety of symptoms and consequences on individuals, the experiences of the individuals with OI may vary greatly, it has therefore been determined that positivism is not appropriate for this research. This also would not have been appropriate as the assumption that past events can be used to predict future events fails to account for changes in external factors, for this research the potential changes over time include changes in treatments available and changes in NHS policy which may alter how OI is managed.

The second approach to be reviewed is interpretivism, this approach recognises that researchers are part of the world they are researching, and that people interpret the world they live in differently (Ryan, 2018). The assumption of interpretivism is that there is an opportunity to understand the perceptions people have of their experiences (Despres, 2011). This approach is well suited to qualitative research as a means of improving knowledge and gaining a deeper understanding of issues but does not allow for findings to be generalised to a wider population (Pham, 2018). As the aim of this research is to explore the experiences of individuals and to investigate whether they are consistent across the OI population, this approach would not be suitable.

The third approach to be reviewed is pragmatism. This approach embraces the use of multiple methods and respects that social science research often cannot rely on one single method (Kaushik & Walsh, 2019). A drawback of this methodology is the need for researchers to be confident in both methods and have the necessary skills to assess the data collected (Cameron, 2009). However, this methodology allows for both qualitative and quantitative methods to be used whereas interpretivism is more suited to only qualitative research. This epistemological approach was found to be useful when the researcher develops and creates an instrument (Tashakkori & Teddlie, 2003). This method is best suited when the study primarily relies on quantitative methods but would first involve qualitative methods.

Based on the review of these epistemological approaches and their individual benefits, it can be concluded that a pragmatic approach with a mixture of methods is most appropriate for this research. This will allow for the research to involve qualitative and quantitative methods in succession. Both of these methods are necessary in order to fully understand the issue. Each of these approaches is important to the research and both offer advantages and disadvantages (Onwuegbuzie & Leech, 2005). Although it can be argued that they are not compatible with each other due to their differing epistemological and ontological values and assumptions, they are both equally valuable in this study (Bryman, 2012). Therefore, both will be used for this research with the provision that they will both be respected and evaluated separately in order to preserve the integrity of each of the methodologies. In order to achieve this, the study will be sequential, this will allow for one methodology to inform the other without the need for synthesis of the data.

In this section the different epistemological approaches have been discussed and the choice of pragmatism for this research has been discussed. The main benefit of using pragmatism is that this approach allows for both quantitative and qualitative methods to be utilised. This would allow for an in depth exploration of the issues and investigation into whether these experiences are consistent across the OI population.

2.3 Research methodologies

In this section of the chapter, the potential research methodologies which could have been employed will be discussed and compared. The decisions taken will also be discussed and the chosen methodology will be described and explained.

As previously discussed, it was decided that a pragmatic approach was most appropriate for this research as it allowed for a mixture of methods and will allow for both qualitative and quantitative data to be collected. This section will explore the different research methods which could have been utilised and will discuss the decisions made regarding the methodology.

2.3.1 Qualitative Research

Qualitative research involves the collection of narrative data, the aim is to develop insights into problems and explore the problems identified (Moser & Korstjens, 2017). When considering the use of qualitative research, the paradigm which will be utilised must be considered. As previously mentioned, there are a number of epistemological approaches which can be used and although it was felt a pragmatic approach was best for this research, a purely qualitative research study would require a different approach.

There is a broad range of methods used in qualitative research. The different methods each have benefits and limitations and the method used will be affected by the chosen epistemological approach. Case studies involve an in-depth contextual analysis of a person/ an event or an organization and although they can yield both qualitative and quantitative data, researchers are able to interpret and examine the events and the outcomes which may make the results less objective (Tracy, 2020).

Interviews are guided conversations in which a person is able to discuss the topic in question and can also provide insights into their opinions, motivation and experiences (Tracy, 2020). Interviews are beneficial as they allow for a topic to be explored more freely than with a set questionnaire as the researcher is able to respond to statements made and ask supplementary questions which will give a better insight into the issues.

In a similar manner to interviews, focus groups allow for more flexibility in discussions than predetermined questionnaires or surveys. Focus groups involve group discussions and give the benefit of conversation between participants who can discuss the topic freely and find common ground or explore disagreements in the topics (Stewart & Shamdasani, 2015).

The analysis for qualitative data involves coding the data and identifying recurring themes and sub-themes within the content. This may be done as a content analysis whereby the content is categorized and examine for patterns in words used and their relationships (Vaismoradi et al., 2013). A thematic analysis can also be performed, this would involve the use of the six steps laid out by Braun & Clarke, (2006). This method offers a reflexive approach which gives greater flexibility and allows for the coding to evolve throughout the analysis (Byrne, 2021).

There are a number of benefits to the use of qualitative research methods. Firstly, it allows for a thorough discussion of a topic and can yield rich data. Secondly, the flexibility allows for the design to be tailored to meet the specific needs of a study. Finally, qualitative research respects that individual experiences differ and allow for these different opinions and experiences to be shared and explored (Rahman, 2016).

There are also limitations to qualitative research methods. Firstly, these methods are time-consuming, both during the data collection stage and after as the data will need to be transcribed and coded (Queirós et al., 2017). Secondly, these studies tend to have smaller sample sizes which limits the generalizability of the findings (Rahman, 2016).

For this research project, due to the flexibility and ability to explore topics in-depth, interviews and/ or focus groups were considered to be the most suitable methods for collecting qualitative data. It was felt that these methods could result in rich data which is needed for the quantitative phase of the research.

2.3.2 Quantitative Research

Quantitative research involves the collection of numerical data, this data which is measurable and objective (Queirós et al., 2017). The main difference between quantitative research methods and qualitative research methods is that quantitative methods are standardized. This makes it possible to have a larger sample population which will result in more data being gathered (Flick, 2020). This data can be analysed using objective statistical tests and results will not be subjective or subject to the same interpretation as qualitative research.

The types of quantitative research can be broken down into two design groups; experimental and survey (Watson, 2015). In experimental research design, the researchers have control over the experiment and how it will be conducted, this includes the ability to randomise participants which can improve the validity of the research (Gray, 2022). Survey research designs will involve surveys or questionnaires and can be cross-sectional, where data is collected at one point in time or longitudinal which involves multiple data collections (Watson, 2015).

Data gathered through these methods has to be analysed using statistical software which can be used to run many statistical tests. The tests which can be used will depend on the type of the data and the distribution of the data. The analysis of the data will involve descriptive statistics to look at the basic features of the data and will summarize the data, and inferential statistics can then be used to test a hypothesis and to draw conclusions from the data (Gray, 2022).

There are several benefits to the use of quantitative data, first is the ability to generalise the findings to a wider population. This is due to the ability to have a high number of participants and this will allow for a more diverse group (Queirós et al., 2017). In addition, these methods are less time consuming and therefore less costly that qualitative research.

The limitations of quantitative research include the inability to further explore issues with participants and cannot delve into the deeper meanings of issues or explain them. In addition to this, there is a risk that this method of research only provides a snapshot of issues and does not reflect the entirety of the issue to participants (Rahman, 2016).

For this research a survey will be used to allow for the findings to be applied to the wider OI population.

2.3.3 Mixed Methods Research

Mixed methods research involves the use of both qualitative research methods and quantitative research methods. This method of research relies on a pragmatic approach as this approach allows for the use of both methods of research and ensures that principles of each method of research will be respected (Flick, 2020).

The data collection in a mixed method study can be done in multiple ways. Firstly, the qualitative phase and the quantitative phase can happen sequentially. The order these are performed in will decide whether the study is exploratory or explanatory, for exploratory the qualitative phase is conducted first and this is followed by the explanatory phase (Creswell, 2014, p. 225).

Alternatively, the methods could also be carried out at the same time, as a parallel study but kept as separate data collections or a nested design can be used where one of the methods is

used as the main method of data collection and the other is used a complementary question within the study (Shorten & Smith, 2017).

A benefit to the use of multiple methodologies is that, the use of a quantitative research method in addition to a qualitative design would allow for a large sample size to be obtained. This would offset any weaknesses in the data and allow for a stronger inference to be made between factors (Doyle et al., 2009). A potential limitation of a mixed methods study is the disparity between the epistemological approaches that would be used for qualitative or quantitative, however this is resolved through the use of pragmatism which embraces the use of multiple methodologies (Doyle et al., 2009).

For this research a mixture of methods is essential as the topic has not been explored in the UK previously. An exploratory sequential study will be conducted to allow for both qualitative and quantitative data to be gathered.

2.4 Models, Frameworks and Theories

In addition to selecting an appropriate epistemological approach, it was also important to consider what theory, model or framework would be used to assess the data gathered and to evaluate the usefulness of the research. Being theoretically driven was important for this research as it helps to guide the research and can assist with making predictions (Fried, 2020). The use of appropriate theories also allows for robust research to be conducted which will allow the knowledge generated to be valuable to the field (Cash, 2018). There are many models and frameworks which could have been used, this section will discuss and evaluate these models and their associated benefits and limitations. When considering what model would be used, it was important to consider the generalisability of the models, some models are specific to certain groups and those may not align with this research. Therefore, ensuring the model would be adaptable and be able to fit with the OI population was essential.

2.4.1 Transition theory

The first theory which was reviewed is the transition theory. This model was designed to assist people during difficult transitions and does this by discussing the needs of individuals more generally. Although the use of this theory for OI has not been documented, this model

has been used in research regarding numerous different transitions, including during discharge from hospital and for older people moving into residential care (Davies, 2005). It has been recognised that the transition from paediatric to adult services was a process and would leave people more vulnerable to changes in their health (Meleis et al., 2000). Ludvigsen (2021) used transition theory in their investigation of the experiences of parents during the transition of their children from paediatric to adult care. They found that collaboration between all stakeholders was essential.

There are four domains to this theory. The first is the nature of transitions; this shows how the model can be used for various different types of transition, not only between different services but also how this allows for the properties of the transition to vary. This element has bi-directional influence on two other domains of the model, these are; transition conditions; facilitators and inhibitors, and nursing therapeutics. The transition conditions domain shows that the transition can be both positively and negatively influenced by the individual, by others involved in the transition and by society on the whole. As previously mentioned, this domain shows bi-directional influence on the nature of transitions, in addition, this domain also shows bi-directional influence on the other two domains of the theory, these are patterns of response and nursing therapeutics. What this shows is that the transition conditions: facilitators and inhibitors domains is central to this theory and can influence all aspects of the transition. The patterns of response domain, refers to the personal development required to go through the transition. Although the theory was not designed specifically for transition from paediatric to adult services, a number of the elements within this domain are relevant to this type of transition. This domain also has bi-directional influence on nursing therapeutics. This shows how the nursing therapeutics domain plays a significant role during transitions and much like the transitions conditions: facilitators and inhibitors domain, as it interconnects with all other domains of this theory (Li & Strachan, 2021).

This model focuses heavily on the person with the chronic condition and does not account for the influence of family members (Li & Strachan, 2021). Consideration of the family aspect is important for this model as families are more likely to be involved in the lives of those with chronic conditions (Lee et al., 2017). Further to this, this model recognises the need for commitment from the individual which is important, but, it is important to note that

commitment would also be required from the HCPs facilitating the transition, which may not always be possible due to limitations within health services (Fegran et al., 2014).

2.4.2 Chronic care model

The second model which was evaluated for this research is the chronic care model. This model by Wagner et al. (1996), was designed to improve the coordination of care. This model argues that in order for positive changes to happen, systems must address the needs of people with chronic conditions (Grover & Joshi, 2014). Disparities between paediatric and adult services has been described as an excellent example of how a lack of coordination can have a detrimental effect on people with chronic conditions (Rapley & Davidson, 2010). Davy et al. (2015) conducted a systematic review exploring the use of this model and found that this model has been used for many different chronic conditions, including diabetes, cardiovascular disease, respiratory disease and depression among others. This systematic review found that the model was not used in a consistent manner across those studies but found that commonly used elements of the model were self-management and system delivery design. There is no evidence of this model being used for OI but as it has been found that this model can lead to improved health outcomes for chronic conditions generally, this model may be appropriate for use in this research (Yeoh, et al., 2018).

The model is split into six elements (Grover & Joshi, 2014). The six elements are 'Health systems or a health organisation', 'Clinical information systems', 'Decision support', 'Delivery system design', 'Self-management support', and 'Community including organisation and resources for patients'. This model has a stronger focus on the delivery of healthcare and a lesser involvement of people with chronic conditions and aims to improve the coordination of healthcare.

A limitation of the chronic care model is that it does not address transition and this restricts the use of the model within this research as exploring the transition does form a significant part of the aim of this research. In order for the model to be used in this research, it would either need to be amended in order to accommodate the issues arising from the transition from paediatric to adult services or multiple models would need to be utilised.

2.4.3 Minimally disruptive medicine care model

The next model to be reviewed is the minimally disruptive medicine (MDM) care model. This is a comprehensive, patient-centred model which seeks to address any factors which could affect the effectiveness of their care (Leppin et al., 2015). This model recognises that the needs of people with chronic conditions will vary and that the goals of treatments and interventions need to align with the needs and goals of the individual (Leppin et al., 2015).

A major part of the model is the recognition and mitigation of treatment burdens on the individual, as a high burden of treatment could affect their capacity to collaborate in their care (May et al., 2009). Shippee (2012) demonstrates how factors such as life demands, and the burden of treatment will interact and affect the health outcomes the individual has. The burden of illness and the burden of treatment both influence the capacity an individual has for dealing with all aspects of their life. As their burden increases, their capacity will decrease and this will then affect their utilisation of care services. This has a direct impact on their health outcomes and this then feeds back into their burden of illness and burden of treatment.

The elements in the toolkit for the MDM care model are broken down into two groups, the 'tools to identify the right care' and the 'tools to make the right care happen' (Leppin et al., 2015). The tools to identify the right care includes aspects such as shared decision making, patient partnerships and goal-elicitation. The aim of these elements is to ensure the treatment plans are designed with the individual in mind and reflects that the goal for care of people with chronic conditions is to provide the best standard of care which is individualised to meet their specific needs. The tools to make the right care happen consists of procedures which should be undertaken by HCPs and could lead to more efficient care of people with chronic conditions. This includes utilising community navigators, developing resource registries and promoting the choose wisely campaign which aims at ensuring care is patient centred and ensuring people utilise the correct services. On the whole, the goal of this model is to aid in the identification of the right treatments for individuals by taking into consideration their wants and needs through shared-decision making between individual and HCP (Abu Dabrh et al., 2015).

A limitation of the MDM care model is that it does not address transitions in healthcare and as with the chronic care model, this would need to be added to the model in order to address issues regarding the transition from paediatric to adult health services or a second model would need to be used in tandem with a model which does address transition.

2.4.4 Movin' on up health care transition model

The final model to be reviewed was the Movin' On Up health care transition model. This model recognises that health care transitions are long and complex processes (Betz et al., 2016). This model shows how the different aspects of the model are interrelated, which is reflective of how these elements would interact in the life of an individual. There are many aspects to the model; firstly there are the domains which are broken down into different stakeholders or influencers, including the environment, the health system, family/ social support and the individual. These four elements are structured as an ecological model, showing that the broader aspects of the model will influence the others. These elements are affected by the interventions arm, this element includes four types of interventions: case management, surveillance, treatment & procedures, and teaching, guidance and counselling. The final two elements of this model are displayed separately from the others and are the integration into adult health care services and adult competencies. These are displayed within the model as arrows which point away from interventions and across the various domains within the ecological structure. This suggests that both integration into adult services and the competency of adults is assumed to be separate from interventions and further to that, that the interventions are not impacts by integration in adult services or the competency of the adults within the service.

This model not only considers the individual and the health care system but also takes the family aspect into consideration. This is especially important for transitions to adult services as families would be closely involved in care until discharge from paediatric services and they would likely still has some influence after the transition. Another benefit of this model is that it is focused on transitions and as a result a transition arm would not need to be added to the model.

2.4.5 Discussion of models

Each of the models reviewed presented potential benefits and drawbacks to their use in this research. There are a number of similarities between the models with some elements appearing consistently, there were some elements that were included in some models but were omitted from others. A similarity across a number of the models was that decision support was included in the CCM, MOU and MDM models. Conversely, self-management only appeared explicitly in the CCM. Furthermore, education was only included in the MOU, this shows that there was not a single model that contained all of the necessary elements for this research.

Although ensuring this research has a strong theoretical basis was vital, it is important to note that the early stages of this research were data driven. As this research study was investigating issues which have not yet been researched in the UK, the appropriate theories or models could not be identified before gathering data and developing an understanding of the issues in the study population. The theories and models which have been discussed in this chapter were utilised after gathering qualitative data and were then used during the quantitative phase of the study. Theory played a significant role in the development of the patient guide and during this phase of the study the theories and models were further developed in order to fill gaps in the current theory as findings did not fully align with existing models or theories.

These aforementioned models will be used to evaluate the results of the research, and this will allow for comparisons to be made between the findings of this research and the findings of other research studies. These comparisons will allow for assessments to be made of these models and to ascertain whether the models do fit with the findings of the research. This section has described the models and theories which could have been used in this research.

2.5 Conclusion

This chapter has discussed the methodologies which could have been utilised for this research. It was concluded that a pragmatic approach was most appropriate for this study as it allowed for a mixed methodology. It was also concluded that the mixture of methods would be done sequentially with the qualitative phase being done first followed by the quantitative

phase making this an exploratory study. Following both of these phases where data will be gathered and analysed, a resource will be developed and feedback on this resource will be gathered from relevant stakeholders. The selected methodologies and the structure selected can be seen in figure 2.

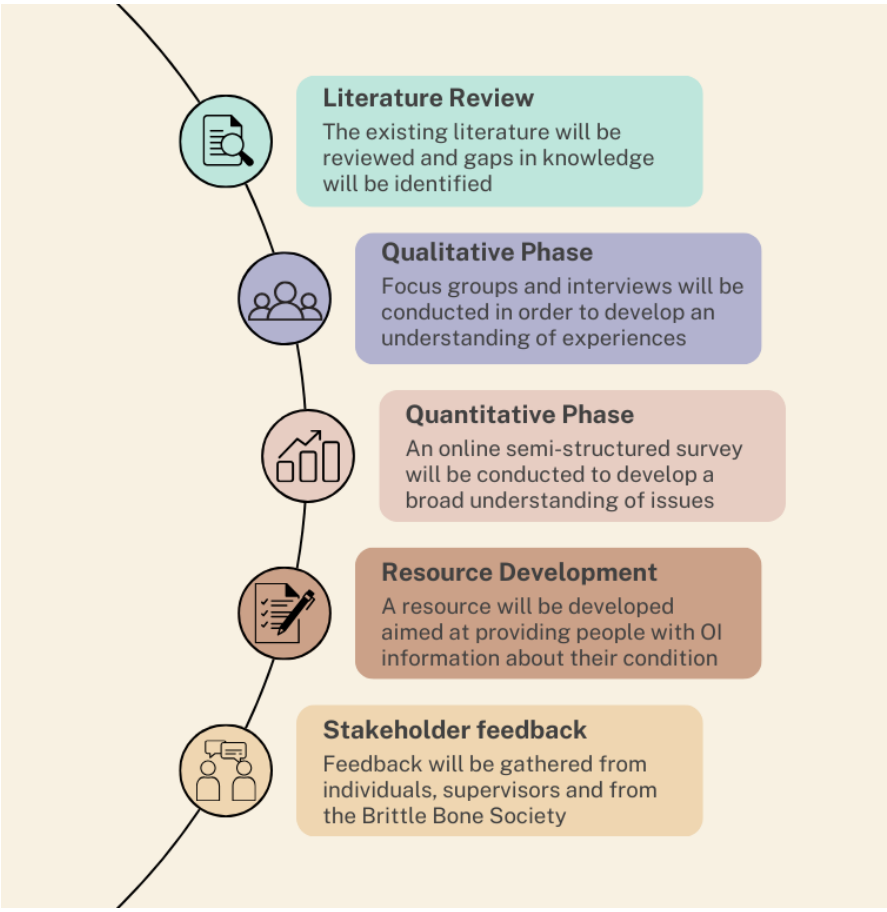

Figure 2: Structure of research

The existing theories and the choice of theory for this research were also discussed. It was concluded that one theory would not adequately cover all of the topics which were to be included in the focus group, the interviews, and the online semi-structured survey and as a result, more than one theory will be used in this research. The following chapter will discuss the qualitative phase of the research.

Chapter 3 – Qualitative Phase

3.1 Introduction

The previous chapter discussed the methods of research which could have been utilised for this research. It was concluded that this research will consist of a mixed-method multiphase study with both qualitative and quantitative methodologies being utilised. This method was deemed the most appropriate for this research as there has been little research conducted in this area. It was felt that the qualitative phase would allow for preliminary exploration of the topic and would allow for the experiences of people with OI to be explored in depth. This is vital for the research as some issues raised in studies from other countries may not be transferable to the UK population due to the differences in health care and other social services. This would then give the basis for a quantitative phase as the data gathered in the qualitative phase could be carried forward and used in the development of an online semi-structured survey. This chapter will focus on the first phase of this research which consisted of a qualitative approach. Initially, qualitative focus groups were the chosen methodology for this phase of the research due to the lack of literature in this area of study. It was felt that this method would allow for exploration of the issues with the sample population which could later be expanded in the form of quantitative research. This method is suitable for this study as the method is flexible and also useful for researchers who are still in the early stages of their career. Despite selecting focus groups as the methodology for this phase of the research, it was necessary to conducted one-to-one interviews in addition to a focus group. This chapter will discuss this change in methodology and explain why that became necessary for this research. Nowell et al. (2017). King (2004) also suggested that thematic analysis is useful as the data generated involves experiences or insights which were not anticipated by the researcher. This is especially beneficial for this study as the area around transition and adult care of osteogenesis imperfecta in the UK has not been explored and as a result, it is unclear what will be discussed by the participants. This chapter will discuss how the qualitative research was conducted, the results of this research and will explore the implications of the findings.

This phase of the research will begin the efforts towards achieving the overall aim ‘to investigate the transition from paediatric to adult services and the management of ongoing

care of adults with osteogenesis imperfecta'. This chapter will begin to address the issues raised by the literature review and will contribute to achieving the objectives laid out in the first chapter of this thesis, specifically first and second objectives; 'to gain an in depth understanding of the transition from paediatric to adult services for people with osteogenesis imperfecta', and 'to identify any changes in care given between paediatric and adult services from the perspective of adults with OI'. Further to this, this chapter will begin to explore the issues relating to the third and fourth objectives of the research; 'to develop an understanding of the ongoing care needs of adults with osteogenesis imperfecta in the UK from their perspective', and 'to identify any gaps in knowledge of people with osteogenesis imperfecta'.

3.2 Method

3.2.1 Participants

The sample was recruited purposively as there were specific criteria that participants had to meet in order of participate in the study. They had to be over the age of 21, had to live in the UK and had to have a diagnosis of osteogenesis imperfecta. The decision for participants to be over the age of 21 was made to ensure that participants would have experienced both child and adult services. As age of discharge can vary, had the minimum age been set at 18, it may have meant that some participants would not have experienced adult services and would not be able to join in the discussion fully. The desired sample for this study was relatively homogenous, as they shared their diagnosis of osteogenesis imperfecta.

The desired sample size for the focus groups was 16 participants, this was chosen as the target sample as this would allow for two focus groups with eight people in. This number of focus groups was selected as Guest et al. (2017) stated that two focus groups should produce at least 80% of all themes. It was decided that a sample size of eight participants within each focus group would be reasonable, more participants within each group would be difficult to manage and Guest et al. (2017) states that fewer than six participants in each group may affect the generalisability of the findings. The recruitment for the focus groups utilised both existing Facebook groups for people with OI and the Brittle Bone Society (BBS). The BBS was approached to assist in the recruitment process and an application was made to their

scientific advisory board which then approved the request. This allowed the recruitment post which was shared in the Facebook groups to also be shared on the BBS social media pages, included in their monthly newsletter to BBS members and it was also included on their website. The Facebook groups selected (Table 2) are specifically for adults with OI. Permission was obtained from the group administrators to place a recruitment advert in the group, including a link to the participant information sheet (which the group administrators were sent prior to giving their approval). The group administrators were viewed as the gatekeepers to this population group, so it was important to obtain their permission prior to posting any information regarding the research on the group page. The recruitment post placed in the groups also included the email addresses for the primary researcher and both supervisors. Prospective participants were asked to email if they had any questions or if they wanted to participate. They were asked to share their age, type of OI and country of residence within the UK to allow for a diverse group of participants to be selected to participate.

Table 2: Osteogenesis imperfecta Facebook Groups

Group Name	Year Created	Number of members	Country of origin	Private or Public Group
Osteogenesis imperfecta (OI) Foundation	Jan-09	10463	USA	Private
Osteogenesis imperfecta UK	Feb-19	139	UK	Private
Osteogenesis imperfecta	May-09	1563	UK	Private
Osteogenesis imperfecta	Dec-10	1335	USA	Private
Osteogenesis imperfecta Research	Oct-06	2410	USA	Private

Following the recruitment, there were a total of seven people who consented to participate in an online focus group. This group was then split into two groups one with four people and one with three. Each was given the zoom information, including the date and time for their focus group. The first focus group was conducted, and all four participants attended and participated. The second focus group could not take place due to withdrawals from the groups. It was decided that a one-to-one interview would be conducted with the one remaining participant from that group. Recruitment was reopened following this one-to-one interview, it was hoped this would allow for a second focus group. However, only one person

agreed to participate in the study at this point and it was decided that a second one-to-one interview be conducted. In total, there were six participants in this phase of the study, following the completion of the second one-to-one interview, the decision was made to cease recruitment and complete the analysis of the data, to allow for the quantitative phase to begin. The demographics of the participants for this phase can be seen below in Table 3.

Table 3: Participant demographics and pseudonyms for the qualitative phase

Group	Gender	Type	Age	Region	Pseudonym
1	M	1	35	England	M, 35yo, T1
1	F	1	66	England	F, 66yo, T1
1	F	1 or 4	32	Wales	F, 32yo, T1/4
1	F	3	33	Wales	F, 33yo, T3
2	F	4	56	Northern Ireland	F, 56yo, T4
3	M	4	26	England	M, 26yo, T4

The small sample size for this phase of the study also brought into question the saturation of the data. Hunter & Gibson (2021) also conducted a qualitative study with a purposive and homogenous sample, their sample size was only 12 participants, the author cited Mason (2010) and stated that the decision to cease recruitment was made after contemplating data saturation. This sample size was justified as Ando et al. (2014) stated that for a study using interviews and thematic analysis, a sample size of 12 is sufficient, as after conducting 12 interviews, all themes had been identified and further interviews provided modified codes, but these did not add to or alter the themes. Although this study did not achieve the sample size of 12 recommended by Ando et al. (2014), there was no minimum number of participants stated and we must also consider the rarity of the osteogenesis imperfecta and the small population from which the participants could be recruited from. As previously mentioned, the total OI population in the UK is around 5000, therefore the population from which a sample could be recruited is limited (Brittle Bone Society, 2022). Data saturation was contemplated after the focus group and the interviews, the codes from the thematic analysis of each transcript were compared and it was observed that there was consistency between the transcripts. The full data analysis was completed after recruitment was stopped, however, preliminary exploration of the transcripts of the focus group and the first interview

showed many similarities and the discussions were echoed in these transcripts. Following the second one-to-one interview, the codes were reviewed again, this showed many of the same codes were found in each transcript and no new codes were identified in the final transcript. Based on this it was concluded that it was unlikely that new topics would be discussed in further focus groups or interviews and the decision was made to cease recruitment. The analysis of the data gathered in the focus group and interviews and the discussion around data saturation is subjective as the stories shared by participants regarding their experiences could be seen to represent new themes if experiences between participants differed slightly. Tracy (2010) argues that when the data gathered is new and can provide a valuable contribution to knowledge, rigor can be achieved with a small amount of data.

Ethical approval was sought from the College and Human Health Sciences Ethical committee. There were some changes required to the application prior to approval being granted, there were a number of comments made by the committee. There were comments regarding the content of the form and some comments on the appendices of the application. The committee asked for clarification on how confidentiality was going to be assured, how participants would be selected, how the conversations were to be recorded and whether peer support would be provided. The comments on the appendices asked for details of the supervisors to be provided in the documents which were to be given to the participants. The committee also asked that proof of permissions from gatekeepers be forwarded to the committee. All of the additions or amendments can be seen highlighted in red in appendix 2. Once these were made the application was approved and the recruitment process was able to begin.

3.2.2 Context of current study

Recruitment for this phase of the study was conducted during the coronavirus pandemic, between April 2020 and July 2020. As the focus group and interviews were conducted online, the social distancing measures and the isolation of vulnerable people did not prevent participants from engaging in the study.

3.2.3 Procedure

The topics and questions to be discussed in the focus group and interviews can be seen in Appendix 3. These questions were based on research conducted in the US and Canada in this field of study. Firstly, the study by Shapiro & Germain-Lee (2012) which investigated the transition from paediatric to adult services for people with OI in the US and highlighted some key issues which would need to be discussed with a sample of the OI population in the UK. Secondly, Carrier et al. (2018) and Dogba et al. (2014) both conducted studies based in Canada which explored the transition of individuals with OI from paediatric services to adult services. Both found there was a lack of evidence in this area and upon investigation the lack of existing transition services had a negative impact on the individuals with OI.

It was decided that the focus group and interviews would be held online, there were several reasons for this. Firstly, it allowed for a wider population to be available for the focus group and interviews as they were not restricted by travel limitations. This improved the chances of meeting the target sample size and gave a better chance of obtaining a diverse sample. The diverse sample required for the research would include both male and female participants, would also require participants from all of the UK nations, a wide range of ages and all of the most common types of OI (I, III and IV). Secondly, it allowed for greater flexibility in terms of dates and times which the focus group and interviews could be held. It was not limited by room availability in the university. Thirdly, it was more accessible for people with disabilities by removing potential physical barriers which may have been encountered in the university such as lift access, parking and any other environmental barriers. Finally, although this method was already chosen prior to the pandemic, the use of online focus group and interviews also meant the focus group and interviews were not limited by covid restrictions and participants were able to take part without putting themselves or others at risk.

The four participants assigned to the first focus group attended the zoom call and the discussion lasted for one hour and 30 minutes. The second focus group which had three people assigned to it was not as well attended with two of the three prospective participants dropping out at the last minute. As the remaining participant did show up and indicated that they were keen to participate in the research, it was felt that it was best to continue and have

a one-to-one interview instead of a focus group. The topic guide was followed, and the conversation lasted for 25 minutes.

Following the focus group and the one-to-one interview, it was decided to re-open recruitment as there was only one male participant and as there were only five participants between the both the focus group and the one-to-one interview, it was not clear whether data saturation had been achieved.

The additional recruitment phase was less successful with only one person consenting to participate. Following the success of the first one-to-one interview and as this additional participant was a male, it was felt that it would be beneficial to the research to conduct a second one-to-one interview. This was a very productive conversation with the zoom call lasting for 50 minutes. The participants were happy to discuss all of the issues within the topic guide and reiterated many of the issues that had been raised in the focus group and the previous one-to-one interview. Given that similar experiences were discussed by all the participants it was felt that following the second one-to-one interview that data saturation had been achieved. This would allow the research to move onto the next phase.

Procedures undertaken to ensure that the participants gave informed consent to participate included allowing the participants to read the participant information sheet. This was available to them through the Facebook group recruitment posts and was also included in the recruitment posts made by the BBS, it was linked in their newsletter, on their social media posts and on their website. Participants were also sent a copy of the participant information sheet along with their consent form to ensure they had plenty of opportunity to read about the study and understand its purpose prior to giving their consent.

Consent had to be given prior to being sent the information to join the zoom call which was password protected to prevent zoom-bombing. The contents of the consent form were read out at the beginning of each zoom call and the participants were asked if they still wished to proceed with the focus group. They were also told they were free to leave the focus group or interview at any time if they wanted to.

The questions to be posed to the participants were mostly open ended to allow the participants to explore the issues, this allowed for other important issues which may not have

been predicted by the literature to arise. This open discussion also allowed for participants to discuss what was important to them and allowed the group to discuss the differences in their experiences.

At the beginning of each focus group or interview, prior to initiating the recording, the participants were reminded that it was voluntary and that if they wanted to cease their participation at any time they could, however their previous contributions to the discussion could not be removed as they may have raised an issue which someone else would discuss further. They were also informed that they could have a rest break during the focus group if they needed to. The consent form was also read back to them, and their consent was confirmed. The reason for recording the focus group and interviews was also discussed with a reminder that this would not be retained following the transcription of the discussion. Finally, they were also told that if they wished to turn off their camera for the discussion they could. All participants were happy with the focus group and interviews being recorded and all elected to keep their cameras on which helped as it allowed for the other participants and the researcher to not only listen to what they were saying but to also read their body language and facial expressions during the discussions.

Following the completion of the focus group and interviews, participants were sent a debrief sheet which included contact information for the researchers and also provided resources on where to go if they wished to get more information regarding OI.

3.2.4 Data analysis

The recordings of the focus group and interviews were transcribed, and these transcripts were used in the analysis. Following transcription, all names of participants, HCPs and hospitals were removed or replaced with pseudonyms to ensure there is no identifiable information included. Thematic analyses were conducted. Coding was done manually on each transcript and was data driven. The identified codes were then used to develop themes and sub-themes. The process for this manual coding can be seen in Appendix 4. Initially the key points were highlighted, organised by colour, with each colour representing a general discussion point and notes were made on the transcripts to assist in the identification of themes. The initial codes were taken from these highlighted statements and from there the

themes and sub-themes were identified. Following the establishment of the themes and codes, these were then added to the transcripts, and these can be seen in Appendix 5.

Thematic analysis is a method of analysis for qualitative data which involves identifying key themes and sub-themes through the use of codes. These codes are used to identify patterns in the data, and these will then be used to extract meaning behind the data. (Byrne, 2021).

There are some limitations to this method of analysis, principally, the questions regarding rigor and the queries in the academic community of how to ensure the analyses have sufficient rigor (Nowell et al., (2017). Holloway & Todres (2003) discussed how this method of analysis can also lead to inconsistency and lack of coherence in the themes which are developed.

The thematic analysis was conducted using the six steps laid out by Braun & Clarke, (2006). The first step of this process is 'familiarisation', this step includes the process of transcription and can allow for initial identification of codes. The second step is 'generating initial codes', this consists of a more thorough review of the data and will require the researcher to begin collating data into codes. The third step is 'searching for themes', here the codes which have been developed and the data which was collated will be reviewed and the identification of emerging themes can begin. The fourth step of this process is 'reviewing themes', at this point the themes which have been developed should be reviewed against the whole data set. The fifth step is 'defining and naming themes', this step requires themes to be refined and defined to ensure themes are consistent and this allows themes to be named. The sixth and final step of the process is 'producing the report', here the final themes and sub-themes should be evaluated against the data set. This can be done using researcher triangulation and this should reveal whether the developed data aligns with the research questions or the existing literature.

This procedure was followed for the analysis of the data. Prior to the change in primary supervisor, a sample of extracts from the transcripts, along with the developed themes and sub-themes were sent to Professor Joy Merrell. She reviewed the themes and sub-themes with the transcript and recommended several changes to the themes and sub-themes.

Previously the coding frame consisted of three themes with 14 sub-themes. Following a discussion regarding these sub-themes a number of changes were made and a further seven sub-themes were developed, and the 21 sub-themes were divided into four themes. The fine tuning of the coding frame was still underway when the primary supervisor of the research became Dr Laura Wilkinson. It was her recommendation that we perform researcher triangulation, whereby around 10% of the quotes from the transcripts are sent to a secondary researcher, along with the coding frame and they then will code those quotes. A comparison is then done by the primary research to see how much agreement there is between them. A percentage match is then calculated, and this will indicate how clear the themes and sub-themes are. Researcher triangulation was conducted with a sample of 10% of the quotes being selected at random and these were sent along with the coding frame to the primary supervisor. This was done to assess the validity and replicability of the coding frame. The first attempt at this yielded a 50% agreement between researchers. Following this some of the sub-themes were renamed to better describe their meaning and some of the quotes were lengthened to provide context.

Following the first attempt at research triangulation, several changes were made to the coding frame, it was hoped that these would bring more clarity to the themes and sub-themes and increase the level of agreement between researchers. These changes include renaming the previous sub-theme of “being an expert patient” to “patient expertise and knowledge”. This was done as this sub-theme primarily consisted of examples of people with OI having to learn about OI and demonstrate their knowledge of the condition to HCPs. “Misconceptions and lack of knowledge of OI by HCPs” was renamed as “Misconceptions regarding OI by HCPs and lack of knowledge of OI by HCPs”, it was hoped that this would clarify the aim of this sub-theme as one which dealt with the misconceptions regarding OI specifically rather than generalised misconceptions regarding chronic conditions. “Inconsistency in treatments” was amended and became “Inconsistency in treatments offered for people with OI”, this was done to better differentiate between this theme and what became known as “Continuity in the health care setting” as there was some overlap and confusion between these sub-themes and their roles in the coding frame.

Despite these changes, there remained some confusion and overlap between a number of the sub-themes and the results of the second researcher triangulation affirmed these

confusions. A second attempt at this process improved the agreement to 54%, however according to Saldaña, J. (2021), this still falls short of the ideal minimum agreement of 75%. This ideal level of agreement was laid out by Roberts et al. (2019), however, they acknowledged that their study fell short of this level. Following a discussion regarding the issues with the coding frame, a number of actions were agreed upon and the following changes were made to the coding frame in order to address these. This should improve the validity of the coding frame.

Firstly, the sub-themes of "inconsistency in treatments offered for people with OI", "continuity in the health care setting", "poor record keeping and lost records" and "inequity in accessing services" have been combined to form a new sub-theme called "Coherence in healthcare". This new sub-theme will encompass all the discrepancies in health care available to people with OI. This will include differences between the UK home nations, health boards and HCPs.

Secondly, the sub-themes "Misconceptions regarding OI by HCPs and lack of knowledge of OI by HCPs", "Patient expertise and knowledge", "Access to specialist HCPs in OI", "Changes with age", "Missed diagnosis of fractures" and "Feeling lucky" will now be in their own theme known as "Knowledge". This new theme has been broken down into sub-themes, "Patient knowledge", "Knowledgeable HCPs" and "HCPs without OI knowledge". It was felt that these new sub-themes would lead to less confusion and ambiguity regarding the discussion of OI knowledge. It was important to group these sub-themes together as they are inter-related. There were multiple occasions during the discussions where participants talked about a spectrum of knowledge ranging from a lack of knowledge by some HCPs and then conversely, they would discuss their experiences of knowledgeable HCPs. This new theme embraces the ambiguity of some of the discussions without removing their importance to the research.

The theme of "Health care needs" has been altered with some sub-themes being combined and some being moved to other themes in order to address issues with interrelation between sub-themes. The sub-themes of "Appropriate referrals" and "Regular appointments" have been combined and this new sub-theme will be known as "Referrals to specialists and regular appointments". It is hoped that this will address the ambiguity that surround the previous sub-themes. In addition, the sub-themes "Lack of support" and "Lack of communication"

were both sources of confusion with some of the issues being raised by the participants being relevant to both discussions around communication and support. These sub-themes have been combined to address this. The sub-theme of "Support and communication from HCPs" provides clarity and will embrace the interrelated nature of the discussions that took place during the focus group and interviews.

The theme "Barriers to care" has been removed from the coding frame. The sub-themes that had been found within this theme have been moved to other themes or combined with other themes. This was seen as appropriate as the discussions regarding barriers to care often revolved around other issues which resulted in some confusion in the coding.

Finally, the sub-themes "Advocating for children" and "Fighting for care" were sources of confusion and the discussions in the focus group and interviews were often applicable to both of these sub-themes. These sub-themes have been combined to form "Advocating for care".

It is hoped that these changes to the themes and sub-themes included in the coding frame will correct issues regarding clarity and will better accommodate the interrelation between the issues discussed in the focus group and interviews.

3.2.5 Reflexivity Statement

As this phase of the study involved gathering qualitative data from a focus group and two one-to-one interviews, it was possible that biases may have impacted the discussion and the direction of the conversation. Biases may have existed as a result of personal experiences with the services which were being discussed, namely the transition to adult services and management of OI as an adult. In order to minimise the risk of personal biases from influencing the data gathered, a topic guide (appendix 3) was used to ensure the conversation remained on track and this ensured all participants were asked the same questions. Other input from the researcher in response to statements made by participants was limited to acknowledging their statements and reiterating their points to prevent miscommunication.

3.3 Results

The thematic analysis of the data led to the development of four themes and each of these themes contained three sub-themes. The final themes and sub-themes can be seen in Figure 3. This section of the chapter will explore these themes and sub-themes with examples of discussions that were had during the data collection phase.

Theme	Sub-Theme	Description
Knowledge	HCPs without OI Knowledge	A lack of understanding by HCPs of OI and how to treat the condition and the knock on effects of this such as missed fractures
	Patient Knowledge	Having knowledge of the condition and the treatments and being able to collaborate with HCPs
	Knowledgeable HCPs	Having HCPs who are knowledgeable and experienced in treating OI
Experiences of care	Frustration and fear	Feelings resulting from being dismissed, doubted and questioned by HCPs
	Dignity and respect	The need for or lack of respect from HCPs to ensure their dignity and self-worth is preserved
	Coherence in health care	Inequities and inconsistencies in treatments and services available to people in the NHS, differences between HCPs, health boards or UK home nations
Being proactive	Advocating for care	Being an advocate for care needs, such as treatments for self and for children
	Engaging in research	Getting involved in research studies for OI
	Engaging with OI Community	Utilising the knowledge and experience of people with OI and organisations working with people who have OI
Health care needs	Support and communication from HCPs	Needing support from HCPs and communication from HCPs regarding treatment, support and services available to them
	Referrals to specialists and regular appointments	Needing to have access to necessary specialists through referrals and needing to have regular appointments with HCPs
	Multidisciplinary team management	The need for health care management to involve HCPs from different disciplines to work collaboratively

Figure 3 Themes and sub-themes from qualitative data collection

3.3.1 Theme 1 – Knowledge

One topic which was often discussed by the participants was knowledge. This was not limited only to their own knowledge but also the knowledge HCPs have regarding OI. The participants described how the knowledge HCPs have regarding OI varies and also explained how they felt this had affected their care. The discussions around knowledge led to the development of the following sub-themes; patient knowledge, knowledgeable HCPs and HCPs without OI knowledge.

3.3.1.1 Sub-theme – HCPs without OI knowledge

The most common issues raised by the participants was the lack of OI knowledge by HCPs and the subsequent misconceptions surrounding OI. Misconceptions regarding the condition and HCPs not having sufficient knowledge of the condition would impact the treatment of the condition. Several participants discussed that "*doctors keep mistaking OI for osteoporosis*" (F, 32yo, T1/4) and because "*they think osteoporosis and OI are exactly the same and they don't differentiate between us at all*" (F, 33yo, T3). As a result, participants expressed how HCPs "*wanted to treat me very much like an osteoporosis or a generic rheumatology patient*" (M, 26yo, T4) where treatment plans are oriented "*on the treatment plan for an osteoporosis person"* (F, 33yo, T3).

Lack of knowledge of OI also leads to HCPs not understanding how easily fractures can occur in a person with OI. One participant described when they went to hospital for a suspected fracture but was told ""*oh no it's fine if only taking a small spill". Yes, a small spill could be like a car crash to you I try to tell them*" (F, 32yo, T1/4). Two participants described similar experiences where HCPs did not believe they had fractured because "*you couldn't have broken it you're walking on it fine*" (M, 35yo, T1) and "*there's no fracture there she's clearly walking fine on it"* (F, 32yo, T1/4).

A lack of knowledge regarding OI and how easily fractures can occur also results in the need for additional care when moving people with OI or doing procedures on them. One participant had seen a "*physio at the hospital after a really, really bad break and she said I'm going to do manipulation on you*" (F, 66yo, T1). Another explained that when HCPs "*try and manipulate me and I'm like DO NOT touch me*" (F, 33yo, T3) in order to prevent fractures. As

previously mentioned in the sub-theme missed fracture diagnosis, when fractures are not diagnosed, some are not given pain medications, this could also occur as a result of HCPs not understanding that people with OI "*do not get used to the pain*" (F, 32yo, T1/4).

Several of the participants also talked about the misconceptions regarding pain when they have fractures. The perceived attitude from HCPs was described as "*they break all the time, they probably don't feel the pain*" (M, 35yo, T1). Another said they had been asked "*Do you still feel the bones hurting? I say yes, I still need pain relief because it's agony*" (F, 32yo, T1/4).

A number of participants also expressed concern that some HCPs are unaware of OI altogether, "*the senior radiologist had never ever heard of it*" (F, 66yo, T1). Another participant said that "*my GP was pretty useless*" (F, 56yo, T4) and that they "*hadn't a clue about brittle bones, they still don't even now*" (F, 56yo, T4). Encountering HCPs who do not fully understand the condition meant they were not "*able to give me anything going forward*" (F, 32yo, T1/4) whereas another participant said they were told that "*there's nothing wrong with me*" (F, 66yo, T1) and was discharged as a result.

During the discussion, several of the participants disclosed that they had experienced difficulty in getting fractures diagnosed or felt that had fractures treated incorrectly by HCPs. Several of the participants indicated that they had experiences of fractures being missed, some as a result of HCPs doubting the presence of a fracture "*I had went to the GP and that said, insisted that it was muscular initially*" (F, 56yo, T4), another said they were told "*no there's no fracture there she's clearly walking fine on it*" (F, 32yo, T1/4).

Participants also said that fractures were missed because "*you do have to look a lot more thoroughly and closely at X rays with someone with brittle bones"* (M, 35yo, T1) and as a result of fractures being missed, one participant found "*the fractures worsened because they haven't locked properly or X rayed a different part of the leg*" (F, 32yo, T1/4).

Another issue with fractures not being identified that was discussed was the issues with treating the fracture. If the fracture is not seen, they are told "*we can't plaster you because there's nothing there"* (F, 33yo, T3). Participants also explained that they had gone to hospital for a fracture but "*didn't get offered painkillers"* (F, 33yo, T3) and "*even if you have to sit there 12 hours. You'd sit there without any pain relief*" (F, 66yo, T1).

Another issue that arose during the discussion regarding a lack of OI knowledge by HCPs was the changes that occur in OI as people age. A number of participants mentioned how their condition has changed as they have aged, there were several changes observed by the participants discussed. One participant expressed that compared to fractures as a child "*as an adult, it [fracturing] does actually hurt a lot more*" (F, 33yo, T3). This was seconded by another participant also stated that "*as you get older, with me every bone I've broken now has been more painful than the last one*" (F, 66yo, T1).

Participants also shared how as adults they felt that "*we disappear, we're off the map then*" (F, 66yo, T1). This was referring to the services available and another participant suggested that when "*you turn 18 and it will disappear, I don't exist anymore*" (F, 33yo, T3). Another participant expressed how the services available to them changed once they were in adults services as they "*went from very regular physio to having to fight tooth and nail to even get six weeks with the physio in adult care*" (M, 26yo, T4).

3.3.1.2 Sub-theme – Patient knowledge

A common topic of discussion was the need to know a great deal about OI, the need to be an expert patient and have a great deal of OI knowledge. There was discussion by the participants regarding their knowledge regarding fractures, as they have so many fractures "*we're not we're not silly to just go and get willy nilly X rays, we kind of know when we need one*" (M, 35yo, T1). They expressed how they have had to "*demand an X ray because I knew there was a fracture*" (F, 33yo, T3).

Participants also expressed how they need to have as much information as possible, this would allow them to be expert patients. One participant indicated that they wanted to know more about ongoing research as it "*would be nice to be kept up to date with what's happening and the new drugs that are available*". (F, 66yo, T1). The benefits of the internet were also discussed and how "*the introduction, really, of the internet. That's when I started to learn more myself about brittle bones*" (F, 56yo, T4). However, the risks of the internet were also discussed, the potential for "*misinformation on the internet it's really hard to know what to believe*" (M, 26yo, T4) and the issues with "*all those discrepancies between the different countries variations and how they deal with things*" (M, 26yo, T4) may also lead to people

being misinformed. This reinforces the findings in the lack of support and lack of communication sub themes.

The benefits of having OI knowledge was also discussed as a person who is fully informed would be equipped to deal with HCPs who lack OI knowledge, as one person said, "*I've often ended up having to correct their management of my condition in inpatient admissions*" (M, 26yo, T4). This knowledge and expertise also comes from living with the condition, as one participant discussed "*those with rare disease like us, where it's like you have a rare disease of course, you know what's going down*" (M, 26yo, T4).

3.3.1.3 Sub-theme – Knowledgeable HCPs

The need for knowledgeable HCPs was discussed and participants said that "*it's really good to see a doctor once a year that knows your condition and can reassure you*" (F, 66yo, T1) and this is also helped by having an HCP who is "*very good and they respect my knowledge*" (F, 66yo, T1). Participants also described knowledgeable HCPs, who were not experts in OI "*a rheumatologist in Swansea, and he has some knowledge of OI, like he managed to tell my GP to prescribe oramorph when I needed* it" (F, 33yo, T3). Participants indicated that they would be happy not seeing a specialist because "*if you get a good doctor that he knows what he's talking about. He can, you know, he knows what you're talking about, as well*" (F, 66yo, T1) and in addition "*a good doctor should really listen to you*" (M, 35yo, T1).

Participants were asked if they felt their care was being individualised to meet their needs. One participant described how their care is being overseen by "*a rheumatologist and mine is being very individualised*" (M, 35yo, T1) and they went on to explain how "*he [rheumatologist] took the time and he sort of sat down and we went through lots of things, you know, he went through my family tree of how it was. It did seem more personalized because he, that was his sort of area*" (M, 35yo, T1).

Some children with OI are seen in specialist OI centres but there are relatively few of these and therefore people have to travel significant distances to get to them, "*when I had [my son], we have to go all the way up to Bristol, because he's inherited it as well. The closest hospital with a department for OI was Bristol*" (F, 32yo, T1/4). This issue is further explored in the "experiences of care theme", within the "coherence in health care" sub-theme.

Participants also discussed how having access to a specialist helped as the HCPs were able to signpost them to other HCPs who had an interest in OI or were experts in managing OI. One participant described how the consultant "*picked out what surgeon would be best to do my hip and knee and he chose Mr A*" (F, 66yo, T1).

A surprising topic which emerged within the sub-theme of knowledgeable HCPs was luck, participants repeatedly stated that aspects of their care was due to luck. It was suggested that having a good consultant was lucky, a participant stated that "*I'm very lucky because now, my main doctor is Dr B, who does a lot of research*" (F, 66yo, T1), another participant said that they "*managed to get Prof C because I was very lucky and got him in with him*" (F, 33yo, T3).

3.3.2 Theme 2- Experiences of care

The experiences of the participants within the healthcare setting were a major and recurrent theme throughout the focus group and interviews. This included how they were treated by HCPs, how care differed in different hospitals or departments and how their experiences affected them. The experiences they described varied and allowed for the exploration of the following sub-themes; frustration and fear, dignity and respect and coherence in health care.

3.3.2.1 Sub-theme – Frustration and fear

Many participants expressed feelings of frustration or fear regarding their care. Participants expressed how they felt there was a lack of services available to them after they transitioned to adult services, "*I don't feel as if there's any aftercare*" (F, 32 years old (yo) Type 1 or 4 (T1/4)). Another participant also mentioned how "*I never got offered it [physiotherapy]"* (F, 66yo, T1).

Fear and worry were also mentioned repeatedly by participants in a number of ways. One participant expressed that as a child they were "*too scared to be touched, because I was so worried of fracturing*" (M, 26yo, T4) and this fear of fracturing or causing harm was also mentioned in reference to pregnancy, "*I was scared. In case I hurt the baby*" (F, 32yo, T1/4). A participant also expressed their concern for the co-morbidities associated with OI, "*makes*

me worried is could I have something wrong with my heart and just not realise it" (F, 56yo, T4).

Being dismissed was also discussed, "*anything I brought up was always dismissed*" (F, 56yo, T4). Another participant mentioned how they were "*constantly explaining myself as well*" (F, 32yo, T1/4) and the impact of this being that "*it's exhausting sometimes*" (F, 66yo, T1).

3.3.2.2 Sub-theme – Dignity and respect

The perceived attitudes of HCPs from the participants were also discussed at length and appeared to be quite impactful on the care and the feelings of the participants. Having HCPs who do not listen to the person with OI was mentioned as having a detrimental impact, as previously mentioned in the missed fractures sub-theme, a participant had a fracture become "*a full-blown fracture because they didn't listen at the beginning*" (F, 32yo, T1/4). Another mentioned that HCPs would not listen when they "*tried to demand an X ray because I knew there was a fracture, but they still said no*" (F, 33yo, T3).

A recurrent topic of discussion within this sub-theme was feeling as though some HCPs would have "*no respect whatsoever*" (F, 66yo, T1). One stated "*we should have that level of respect as does everybody else*" (F, 32yo, T1/4) but others seemed to also have missed out on this as another explained that they were "*laughed at by the GP*" (F, 56yo, T4) who doubted their hearing loss.

Another participant explained that they had been taking a medication but "*it wasn't working for me. So, he [consultant] said, right, let's get rid of that*" (M, 35yo, T1) and was able to see the benefits of having an HCP who listens and respects their patients.

3.3.2.3 Sub-theme – Coherence in health care

There were many aspects to coherence in health care discussed during the focus group and interviews. Firstly, participants discussed how their care had differed and the inconsistencies in the methods for treating OI. A topic that was mentioned by multiple participants was the lack of consistency in the HCPs that they see in appointments and in the hospital in general. One participant described how when they "*fracture something, there's no consistency. I haven't seen the same doctor twice*" (F, 32yo, T1/4).

Participants also described how this is not only the case when being seen for an emergency. One explained that they have "*gone through, you know, three or four different doctors in specialists*" (M, 26yo, T4). Another described how their consultant asked them "*do you mind if my senior registrar sees you*" (F, 66yo, T1) and went on to explain how they "*went, then for my annual appointment I saw another doctor"* (F, 66yo, T1).

One participant also described difficulties with getting treatments as "*sometimes my local hospital will agree to something which then I know that my specialist hospital won't agree to*" (M, 26yo, T4). They also explained that they are seen in both a local hospital and a specialist hospital "*because the local hospital can administer care more conveniently and more quickly than a specialist*" (M, 26yo, T4).

In addition to differences in treatments, participants also discussed the differences in the services available for OI around the UK. Participants discussed how "*there's not a lot anywhere for OI*" (F, 66yo, T1). As a result, people have to travel further to access specialist care, one explained that they have to travel to "*London because they have no one in Wales for me*" (F, 33yo, T3).

The difficulty in accessing the specialist centres was discuss, as not everyone is able to travel around the UK to have appointments, as a result "*people who need specialist services, but don't live near a local Centre of Excellence but miss out on the specialism*" (M, 26yo, T4).

One participant also acknowledged that "*there are doctors and surgeons out there that do take an interest in that they, you know, they're brilliant and it's just finding them and they're all in different areas*" (M, 35yo, T1).

A number of participants also expressed how the continuity seen in paediatric services was something that they missed and felt benefitted them. Participants described how in paediatric services "*you tend to get to know the nurses and the staff and the clinical team, whereas in adults. You know, there's in any given hospital, there can be upwards of 10/11 wards*" (M, 26yo, T4) and how HCPs would be very welcoming and reassuring, "*they used to say to me, or next time you break a bone just come back to us*" (F, 66yo, T1).

One of the participants described how "*for my annual appointment I saw another doctor*" (F, 66yo, T1), they were repeatedly passed on to other doctors which would have been very disruptive to their care. This was also discussed in the inconsistency in treatment sub-theme as each new doctor would not have been familiar with their medical history.

Continuity in treatments was also discussed as one participant explained that his paediatric consultant "*was keen to sort of keep me on, to keep getting that treatment and because he knew that I could get it as long as I was being seen by him*" (M, 26yo, T4). He described how the "*transition from Dr D my adolescent physician till I was about 20 years*" (M, 26yo, T4).

One participant described how they would have "*one doctor here and there I see and never the same one twice*" (F,32yo, T1/4), this would mean they would have to start from scratch with each doctor and it would not be clear how much they know about OI. Conversely, another participant described how they "*didn't have to repeat myself because Dr D filled in the adult physician a lot about the context about the specifics of my case*" (M, 26yo, T4).

There was also mention of limitations facing HCPs, where they may not know that a person has OI and how they may not have easy and immediate access to a person's diagnosis and medical history. Participants discussed how it should be possible for notable diagnoses to "*be across the top of the computer screen, you know, OI type 1. Even with my GP I have to remind him every time*" (F, 66yo, T1). This would ensure HCPs knew before beginning any treatment and it would ensure the most appropriate and experienced HCPs treat them," *I just think if there's something written, then surely, you want the senior radiographer or senior doctor to look at these X rays*" (M, 35yo, T1).

If HCPs had easy access to records, it would prevent people with OI from needing to go to the hospital with "*all these bits of paper for them to read*" (M, 35yo, T1).

3.3.3 Theme 3- Being proactive

A common topic of discussion was the need for participants to be very active in regard to their healthcare management. Participants discussed the actions they took to improve the care they and their children received. These discussions were focused on the following sub-themes; advocating for care, being active in research and OI community.

3.3.3.1 Sub-theme – Advocating for care

It was made clear that getting good quality healthcare has not always been easy for the participants as they all shared times when they had to "*fight for everything*" (F, 66yo, T1). One of the things participants said they had to fight for was to be seen in the best possible hospital for them, one participant mentioned that there had been several occasions where they had "*insisted upon going back to [a hospital in London]*" (F, 66yo, T1) and "*I insisted 15 years ago on being referred to [a hospital in London]*" (F, 66yo, T1). Another participant talked about how they had difficulties in getting seen in the correct hospital for them as "*Wales decided that I wasn't to be seen in London anymore. So, I had another fight on my hands*" (F, 33yo, T3).

Participants also mentioned many times when they have had to advocate for treatments or access to services which they needed. Physiotherapy was mentioned by participants, one explained that "*even now when I break a bone, I have to fight and fight for physio or anything and it's exhausting*" (F, 66yo, T1). Another reflected on how they "*went from very regular physio to having to fight tooth and nail to even get six weeks with the physio in adult care*" (M, 26yo, T4), this links back to the point made in changes with age as participants suggested that they had access to fewer services as they aged.

Also mentioned by several participants was that "*even X rays sometimes as well I have to fight for*" (F, 32yo, T1/4) but despite fighting for them, participants have found that "*even if you demand an X ray half the time, they say no anyway*" (F, 33yo, T3).

A participant also talked about their attempts to get referred for a DEXA scan by their GP. They explained that "*the brittle bone society recommended that I get a DEXA scan*" (F, 56yo, T4) and "*I don't know how many times I asked for DEXA scan, and I was told no*" (F, 56yo, T4). Despite best efforts to fight for care, as their GP would not listen, they could not get referred, as was previously mentioned in "HCPs without OI Knowledge" sub-theme.

Participants also highlighted that fighting for care results in a lot more work for them as "*it's just a thing that you have to fight for everything*" (F, 66yo, T1) and if they do manage to get seen by the HCPs they need it means "*there's more for me to manage, in terms of trying to*

make sure that I can go to basically, to double the amount of appointments" (M, 26yo, T4) which would be exhausting in the long term.

There were several participants who shared that they have children with OI, all those who did this, also expressed their desire to protect them from the difficulties that they themselves have faced by advocating strongly for their care. There were a number of key topics which were discussed by the participants regarding advocating for their children.

One of the strong motivators discussed was the "*need to make sure that things get better for their sake or my daughter's sake*" (M, 35yo, T1) and "*wouldn't want him going through the same kind of problems later on in life*" (F, 32yo, T1/4).

Participants also discussed their desire for their children to be seen by the best possible HCPs "*I had her referred there to see a whole team of people and we, you know, so she's now under their care*" (M, 35yo, T1). One participant also expressed how they were more confident in pushing for things from HCPs and they "*think I'm a lot more stubborn now as well. And I put my foot down*" (F, 32yo, T1/4).

Being able to advocate for children also depends on having information about OI, one participant shared how having a son with OI has meant they "*learned a lot more through him, As the years sort of rolled by*" (F, 56yo, T4). This links back to the sub-theme being an expert patient, as they learnt more about OI it would put them in a better position to advocate both for themselves and for their children. When asked whether they feel like they have the kind of skills you need to advocate for yourself, they responded, "*No. For my son. Yes, I did*." (F, 56yo, T4).

3.3.3.2 Sub-theme – Being active in research

There were two participants who shared that this was not their first time assisting with research regarding OI, they both stated that they are participating in the TOPaZ study (Treatment of Osteogenesis Imperfecta with Parathyroid hormone and Zoledronic acid). TOPaZ is a randomised open-label clinical trial for people with Osteogenesis Imperfecta. One mentioned that they "*went up to the hospital a year ago, not even a year and I only went because I was on this TOPaZ study*" (M, 35yo, T1), the other stated that "*I saw an*

endocrinologist, just because of joining the TOPaZ study" (F, 56yo, T4). They did not indicate if they had not been part of the study whether they still would have done these things.

One participant showed an interest in ongoing research, this was mentioned in the being an expert patient sub-theme but also shows that people with OI do want to engage and "*know research that's happening now and the outcome of it*" (F, 66yo, T1).

3.3.3.3 Sub-theme – OI community

Engaging in the OI community was also discussed, and participants shared how this community has helped them. The brittle bone society (BBS) was mentioned by a number of participants, the charity has a network of members and offers information and advice for people with OI and the parents of children with OI. One participant stated that when looking for information "*I do start in the Brittle Bone Society*" (M, 26yo, T4). However, another participant stated that although they offer information, "*it still feels that they are geared up to paediatrics*" (F, 33yo, T3) and that suggests that there is a lack of information regarding adults.

The network formed by the BBS also means they can offer advice and recommendations to people with OI. One of the participants also stated that "*the brittle bone society told me they were the best to go to and to ask Doctor E*" (F, 66yo, T1). Another discussed how the BBS "*recommended that I get a DEXA scan*" (F, 56yo, T4), as previously mentioned in the advocating for care sub-theme, although they are given the information without further support or increased awareness of OI by HCPs, people with OI will still have to fight and may not always be successful.

The OI community also exists outside of the BBS network, with many people utilising online community groups through social media sites such as Facebook. Participants said that they used these platforms to get OI information "*I mean that's only in the last couple of years or so that I've learned that, again, mainly through the internet, Facebook and brittle bones society*" (F56yo, T4). They also provide support for people with OI as one person said that they "*got more support out of the OI community generally than I did specifically from health care or social care professional*" (M, 26yo, T4).

3.3.4 Theme 4- Health care needs

The third theme identified from the focus group and interviews centred on the health care needs of the participants, specifically what they felt would aid their care and what they would like to see change in the future. There were three areas identified which form the following sub-themes: support and communication from HCPs, referrals to specialists and regular appointments, and multidisciplinary team management.

3.3.4.1 Sub-theme – Support and communication from HCPs

Participants discussed the needs they have in the healthcare setting at length with two issues being discussed a great deal. These are the need for support from HCPs and the need for communication with them in order to manage their care. These have been discussed together as the issues are interrelated and were often discussed together by participants.

Participants conveyed how a lack of support from HCPs had affected them. All of the participants stated that they did not have a transition "*there was no hand over as such from paediatric to adult*" (F, 66yo, T1) or that the "*transition hadn't really happened*" (M, 26yo, T4). Without a transition period, participants said they felt the attitude was "*you're on your own type of thing*" (F, 32yo, T1/4) and the whole process was "*very higgledy piggledy*" (F, 33yo, T3). One participant even said, regarding their discharge from paediatric services, that they "*was never actually formally told*" (F, 56yo, T4). One participant explained that after they were discharged from paediatric services "*it was a massive gap. Years and years*" (M, 35yo, T1) with no consultant.

The lack of support was also discussed when participants were asked if they had ever been seen by multiple services together. One participant explained that "*Not for me no but my daughter, there has been so you go, she had the appointments with Great Ormond Street, and there must be six people in the room*" (M, 35yo, T1). Another explained that a video conference call during the pandemic "*was the first time and probably the only time that I was ever given that opportunity*" (M, 26yo, T4).

Participants also explained that a lack of information meant they did not feel supported, one said that "*it still feels that they [BBS] are geared up to paediatrics and not that adult sector*"

(F, 33yo, T3). Another stated that "*I didn't really trust or know to look for information*" (M, 26yo, T4).

Issues with communication with HCPs were also discussed during the focus group and interviews. As previously mentioned, a lack of information regarding OI is confounded by a lack of communication from HCPs. A participant described how they "*had very little condition specific information. It was never given because the expectation was that I already knew*" (M, 26yo, T4).

One participant also described how they did not get a surgery as a child because they were not told "*it would have been beneficial as an adult*" (F, 33yo, T3) and "*only the fact that I would be in pain*" (F, 33yo, T3) was explained to them.

A participant also discussed the lack of communication between HCPs, "*I've had to wait weeks or months for them to liaise with the specialist team*" (M, 26yo, T4), the long wait could make the health issues worse. They also described incidents where they were expected to "*be the conduit of information*" (M, 26yo, T4) which would be difficult without a thorough understanding of the condition.

3.3.4.2 Sub-theme- Referrals to specialists and regular appointments

Being able to get to the correct services was also seen to be a topic of significance during the discussions. As several participants mentioned in the "support and communication from HCPs" sub-theme, they did not have transitions from paediatric to adult services, one participant discussed how ensuring they had access to HCPs after being discharged would be simple as "*all it would have taken was a referral to an adult service and surely that would have been it*" (M, 35yo, T1).

Having appropriate referrals is contingent on HCPs having some understanding of OI, as seen in the "HCPs without OI Knowledge" sub-theme, there are some who do not understand the condition. One participant described how they had not been able to access physiotherapy and at one point were told "*well if you're desperate go and pay for it*" (F, 66yo, T1).

Referrals to specialist services often depend on GPs, as was seen in "HCPs without OI Knowledge", there are some GPs who do not understand OI and as "*everything has to be*

done through your GP. So, if you're GP doesn't listen to you, you don't get anywhere" (F, 56yo, T4).

Participants also described how getting an HCP to do the referral is not the only issue as getting referrals to be completed can take a very long time before even getting onto a waiting list. One participant had been told to "*go back to your specialist and get them to write to us with a referral to do whatever they want you to do*" (M, 26yo, T4) and another was told they have to "*go back to my GP and get my GP to refer me*" (F, 66yo, T1).

Being able to rely on the same HCP and seeing them at regular intervals was also mentioned by several participants. Participants discussed how "*it's really good to see a doctor once a year that knows your condition*" (F, 66yo, T1) as they would get to know the individual and be in the best position to offer treatment options which meet their needs.

One participant mentioned that they believed "*we should be entitled to like a once a year, sort of multi-disciplinary check-up*" (F, 56yo, T4) as this would allow for all aspects of the condition to be monitored.

The use of technology to assist with regular appointments was also discussed "as having the option to do video conferencing or telephone consultations going forward as an option or an alternative as an additional resource" (M, 26yo, T4). This was also mentioned in the "Support and communication from HCPs" sub-theme and would also help to tackle some of inequities in accessing services by making specialities more accessible.

3.3.4.3 Sub-theme- Multidisciplinary team management

Participants mentioned that there was a need for them to be seen by a MDT, some had that in childhood but lost that when moving to adult services. The participants were asked if they had ever been treated by multiple services who are working together to treat you, the responses of participants were consistent; "*Nope, never*" (F, 33yo, T3), "*that's what I kind of remember as a child might have happened. But when you're an adult. No*." (M, 35yo, T1), "*Never*" (F, 56yo, T4) and "*No, so it's been what I'm 26 now. So, within the eight years I've transitioned at the sort of specialist level*." (M, 26yo, T4).

The benefits of having an MDT were discussed by the participants and were not limited to the people with OI but "*I think that it just needs to be a more disciplinary focused approach. Multi-disciplinary, that's what I meant. That it would be cheaper for the medical profession. If they provided the correct services*" (F, 33yo, T3).

Also, one participant discussed how the use of an MDT would allow for HCPs and the person with OI to work "*collaboratively and co-productively [and] can then work towards ensuring that the outcomes or whatever is being worked on suits both people, are not too heavy handed for one side or the other*" (M, 26yo, T4).

3.4 Discussion

The aim of this phase of the study was to explore the experiences of people with OI in regard to the transition to adult health services in the UK and the management of their on-going care as adults. These findings would then be used to design an online semi-structured survey which would further explore these issues and to ascertain whether they are reflective of the whole of the UK. A topic guide was developed using the existing literature and this was used in one focus group and two one-to-one interviews were conducted via zoom.

There were four themes identified in this phase, each of the themes could be broken down into three sub-themes. The "Knowledge" theme identified issues relating to a lack of understanding of OI, both by individuals with the conditions and by the HCPs responsible for treating the condition. The "Experiences of care" theme included issues relating to how experiences in both paediatric and adult health services affected them emotionally and the difficulties arising from a lack of continuity in health care. The "Being proactive" theme indicated that self-advocacy was important in ensuring people with OI get the right care and that utilising the OI community plays a significant role. Finally, the "Health care needs" theme highlighted that support and communication with HCPs is vital and that ensuring access to the correct services is critical.

3.4.1 Existing Literature

There is a lack of evidence in this field and the previous studies identified in the literature review exploring these issues in OI were conducted in the US and Canada. Three studies

identified were conducted in Canada and one was conducted in the US, these were Dogba et al. (2014), Jeong et al. (2018), and Carrier et al. (2018). These studies are discussed in turn below, although these studies did echo many of the points raised by the participants in this study, there are some limitations to the use of these studies in the UK as a result of the differences between the health systems in both the US and Canada in comparison to the UK. This section will explore this existing literature relation to their similarities with this research, their strengths, any limitations, and the implications of these findings on this research and vice versa.

Dogba et al. (2014) conducted an analysis of the transition programme in the Shriners Hospital for Children. There were some similarities between their findings and the findings of the focus group and interviews in this research. A lack of availability of resources to support the transition to adult services. This is comparable to the UK as the only information readily available to people with OI comes from the BBS, with most of their information being focused on the care of children with OI.

There was a notable difference in the discussions made between the study by Dogba et al. (2014) and the focus group and interviews for this research. It was found that individuals with OI were beginning their transition process at around age 14 and it was subsequently reported that participants were feeling well prepared for the transition to adult services. These findings were not reflected in this research but support the statements of the participants who were advocating for greater support during the transition process.

There was discussion by Jeong et al. (2018) that individuals with OI felt that they had no one to turn to and that HCPs did not know how to treat OI. This is similar to the findings of the focus group and interviews where it was discussed at length by participants. Jeong et al. (2018) also stated that those who utilised the tool achieved a greater understanding of the condition. When taken together, these arguments would appear to support the assumption that having access to resources to aid transition to adult services would be better prepared to manage their care as adults.

There were discussions in the focus group and the interviews regarding individualised care. Some participants in this research stated that their care had been individualised to a degree,

this was not the same for all participants. It was stated by Carrier et al. (2018) that flexibility and individualisation was necessary when planning the transition to adult services. However, there is an inconsistency with this argument as they later go on to emphasize the importance of a structured transition programme and the benefits this has with regards to patient satisfaction. Although the need for a structured programme is clear, the need for that to also be flexible cannot be overlooked as the needs of individuals can be very different and can evolve over time.

Although the findings of the Canadian studies do echo the needs of individuals with OI in the UK, those findings cannot be applied directly to the UK. There are differences in policy and practice which will affect how effectively recommendations can be carried over. Firstly, the study by Carrier et al. (2018), discussed the age at which preparations for transfer should begin, they state that transition to adult services typically happens between age 18 and 21 and as a result, preparation should begin around age 14. In the UK, transition to adult services typically happens between age 16 and 18, assuming the preparation process would take the same duration in the UK, preparation in the UK would need to begin by age 12 . Whether children are able to understand their condition sufficiently at this age would need to be investigated and the process for preparing children for transition to adult services in Canada may have to be revised and amended in order to suit the UK.

The Canadian health system is similar to the UK system as like the NHS, healthcare in Canada is free at the point of care. Some health services such as prescriptions and at home care are funded by a combination of government funds and by the individuals which is also true in some parts of the UK (Martin, 2018). Despite this similarity, a higher number of people in Canada report difficulty in paying for these services, 30% in Canada and 12% in the UK (McAlister, 2018). This could be due to some of the home nations paying for these services out of their NHS budget and therefore reducing the burden on the individual (Bevan et al., 2014).

The 2012 study by Shapiro and Germain-Lee conducted in the USA, revealed that individuals reported a lack of continuity in their care when moving between paediatric and adult services. This does reflect the discussions in the focus group however, they also reported that issues with insurance would hinder the access to specialists and therefore this lack of

continuity could be the result of insurance coverage rather than the availability of OI specialists.

The US health system is vastly different to the UK with care being paid for by health insurance, which is the responsibility of the individual, although some people are able to get public health insurance, which is government funded, through schemes like Medicaid. Despite public health insurance schemes, in 2020 8.6% of the US population had no medical insurance at any point during the year (Keisler-Starkey & Bunch, 2022).

As the health system is vastly different to the UK health system, it makes it difficult to compare findings of US studies to UK studies as the experiences of individuals are the result of different factors and will affect their lives differently. In the UK, cost of healthcare is not the principal barrier to accessing healthcare and therefore the experiences of individuals would be focused on other factors such as waiting times.

Shaprio and Germain-Lee (2012) also discuss health burden and how the responsibility of communicating between HCPs often falls to the individual. This was reflected in the focus group as it was expressed that on many occasions people have had to relay information between HCPs in the same way. This is in spite of the technology which allows for instant communication, even in remote areas which was also referenced as a possible solution by participants in the focus group and in the interviews.

3.4.2 Theories and Models

There were a number of theories which could have been used to frame this research. The main discussions in this research can be split into topics regarding the transition to adult health services and the topics regarding the ongoing care of adults with OI. It was observed that no one theory perfectly encapsulated both of these topics. As a result, it was necessary to use multiple theories for this research.

For the transition arm of the research, there were two possible theories which would cover the majority of the issues raised in the focus group and interviews. These were ‘transitions theory’ and the ‘Movin' on up health care transition model’. When comparing the elements included in each of the models, it was made clear that there were many overlapping features

of these models but that each model also included some elements that were missed by the other.

One limitation of the transition's theory is the use of ambiguous language, and it is not made clear what the meaning of some the elements included are. Conversely, the Movin' on up health care transition model uses lay-terms and is more user friendly than the transitions theory. This is important as information will be accessible, and language will not be a barrier. This is essential as it is considerate of both health literacy and general literacy. Health literacy is the ability of a person to understand information and to make decisions about the health, this is critical to empowerment and allows for self-determination (World Health Organization, 2021).

Both of these models appear to include self-management and advocacy. However, neither of the models include individualisation, the need for this during transition for people with chronic conditions has been highlighted in many research studies. Gleeson & Turner (2012) expressed that individualisation would allow for transition to be a collaborative process which would account for and be respectful of an individual's needs. The need for this specifically for people with OI was discussed by Tournis & Dede (2018b). Although not all of the participants had experienced individualised care, one participant did express that his care *"did seem more personalized because he, that was his [consultant's] sort of area"* (m, 35yo, T1). However, participants did agree when asked that they needed individualised care. Therefore, individualisation should be considered when discussing the transition to adult services for people with OI.

The transitions theory includes the 'transition time span' and the 'change and difference'. It is assumed that 'change and difference' refers to the change of the individual and their needs over time. Both of these issues arose during the focus group and interviews and the transition time span was discussed in the existing literature. The change over time in their condition was discussed by the participants, "*when you were a child, you kind of cry for effect, I find. But, as an adult, it does actually hurt a lot more*" (F, 33yo, T3) and this was echoed by another participant who stated that "*as you get older, with me every bone I've broken now has been more painful than the last*" (F, 66yo, T1).

The Movin' on up health care transition model included some elements that were missing from the transition's theory. These include, surveillance, family and case management. Surveillance is important during the transition to ensure that any changes to health are identified early, and it would also ensure the individual is supported during the transition. The family element is also important as the transition to adult services would mean a shift in responsibility from the parent to the individual which would have an impact on family members. This was discussed by participants, when discussing advocacy in child services one person said "*I suppose my parents would have dealt with it more*" (F, 56yo, T4) and the importance of being able to take over the responsibility was highlighted that transition should involve a shift and includes "*making you start to feel a bit more like an adult that you do have your own life to lead and they may have your own expectations and your desires that are separate from your parents"* (M, 26yo, T4).

It is unclear what the case management element covers, this could provide the opportunity for individualisation. This is important for OI as the data showed that individual needs can vary greatly and therefore treatment plans need to be individualised to account for these disparaging needs. This model does include surveillance, this may include monitoring for changes over time, which was missing from this model, this aspect was included in transitions theory as 'change and difference'. This is important for OI as it was made clear in the data that the impact of OI on an individual can vary over time and as a result, the treatment needs can increase or decrease at any time. This will also be affected by the length of the transition process as even during this process the needs of the individual may change and therefore the transition process needs to have the flexibility to account for these changes.

Given that this model includes some elements that were missed by the transitions theory and is only missing transition time span and individualisation, it is felt that the Movin' on up health care transition model is most appropriate for use when discussing the findings relating to transition from paediatric to adult services.

For the ongoing care of adults with OI arm of the research, there are a number of models which could be suitable. These models would not be appropriate for the transition arm and therefore these models will be used for discussions of the issues related to adult care as these issues will be viewed independently.

The chronic care model includes six elements, three of these elements relate to the needs of the individual. These are decision support, self-management support and community including organisations and resources for patients. These elements are all crucial issues that were discussed during the focus group and interviews. However, the remaining elements are centred on the systems which allow for the delivery of care and are aimed at HCPs and not the individual. This is not relevant to the current research as this research is focused on individuals with OI rather than the HCPs.

The minimally disruptive medicine (MDM) care model is a comprehensive tool which includes elements which are aimed at individuals, HCPs and some are aimed at both to allow for co-production. The MDM does not address self-management and many of the HCP focused elements would not be suitable as the experiences gathered in the focus group and interviews was that of the individuals and not of HCPs. Self-management is important in chronic conditions as it allows for people to take more control over their health and to work with the HCPs. This was discussed by participants, and it was felt that "*there is a certain thing to be said about co-collaboration and co-production . I think there should be a 50/50 split where there is a recognition, especially with those with rare disease like us, where it's like you have a rare disease of course, you know what's going down*" (M, 26yo, T4).

The MDM does address burden of illness and treatment which is not included in the chronic care model. This is relevant to this research as the burden of illness was discussed by participants and they emphasized the difficulty they faced "*trying to get them [HCPs] to communicate or trying to be the conduit of information does put a lot of pressure on me because it's that either they expect me to share information between them or they expect me to be able to relay stuff*" (M, 26yo, T4).

All three of the applicable elements included in the chronic care model are included in the MDM and therefore the chronic care model will not be used. The MDM is the most appropriate model for the ongoing care of adults with OI.

3.4.3 Implications

The focus group results showed that there are significant issues regarding misconceptions and lack of knowledge of OI by HCPs. One of the issues relating to misconceptions was the

lack of knowledge surrounding co-morbidities of OI. There are numerous co-morbidities, some of which can be very dangerous such as neurological defects (Marini & Dang Do, 2000). As these issues are not understood or known about by some HCPs responsible for the care of adults with OI, there is a risk that issues such as neurological defects would not be diagnosed early and this would lead to worsening of the condition. The consequence of such issues not being identified and treated early is the increased burden on the health service. It is often stated that prevention is better than cure, this not only refers to the health of the person with the condition but also the health services as the intervention required to resolve these issues later would be more significant. This would add to the cost of managing OI for an already struggling NHS (Iacobucci, 2023).

It was clear that the general lack of knowledge of OI by HCPs has a detrimental impact on people with OI. However, it can be argued that this is partly fuelled by the lack of OI specific services for adults in the UK. There is nowhere for adults to go where they can see a specialist. This puts them in the untenable situation of having to educate their HCPs on OI themselves, but this is not a realistic prospect as time is limited in the healthcare setting (Salisbury, 2019). It would not be possible for people with OI to ensure each HCP they see has an in-depth understanding of their medical history and OI generally.

The lack of specialist HCPs ultimately results in them having to rely on the abilities of HCPs who do not have the specialist knowledge required to manage a complex condition such as OI. It was found by Kolovos et al. (2021) that 54% of the admissions of people with OI were of people under the age of 14. A significant proportion of admissions therefore come from adolescents or adults with OI. There is a large demand for specialist services and as those services do not exist, adolescents or adults with OI have to carry the burden of ensuring their HCPs understand OI and can give them the treatments they need. This is a substantial burden to place on the shoulders of people who are already having to deal with a chronic condition which is associated with chronic pain (Eton et al., 2013).

The burden of having to educate HCPs on the issues relating to OI are also compounded by the issue of OI knowledge of people with OI. It is understood that living with a chronic condition allows for the development of knowledge relating to the condition and an understanding of the impacts that the condition has on the life of the individual (Wilson et

al., 2007). However, this knowledge takes time to develop. It is possible that at the point of entering adult services, they may not have sufficient experience to draw on in order to guide the HCPs in the management of their condition. That is not to say that with time or training for the individual that it would not be possible, it should be encouraged and supported in order to maximise the chances of positive health outcomes (Lindsay & Vrijhoef, 2009). A successful expert patient relationship would ease the burden of care on the person with OI and the health system as they would be aware of the risks associated with their condition and could act early when problems arise (Donaldson, 2003).

The development of expert patients is also contingent on the availability of information regarding the condition. They need to be aware of treatments available to them and this information is often difficult to find or can be very overwhelming to lay-people. At the present time (February 2023) there is no page on the NHS Choices website for OI and the information from the BBS is more focused on paediatric care. As a result, people have to rely on other websites, from other countries to get information regarding OI. This information may not be applicable to the UK as health systems around the world differ greatly (Papanicolas et al., 2019). In addition, there may be differences in the treatments available in the different nations of the UK as healthcare is devolved to the governments of each of the nations (Bevan et al., 2014).

As it is difficult to find information, the availability of knowledgeable HCPs remains the best hope for people with OI. There were discussions around HCPs who are not specialists being excellent at caring for people with OI as they were aware of research in the condition and ensured they were updated on new developments in treatment. This emphasises the need for continued research in OI and the management of it. The availability of such research will enable HCPs who are not specialists to provide appropriate care. The availability of research would also help to bridge the gap in the guidance from the NHS and other organisations responsible to recommending suitable care for specific conditions. There are not OI specific guidelines and instead the condition is mentioned in general guidelines for rheumatology services and orthopaedic services (NHS England, 2013c, 2013e). In the absence of guidelines, there is often a requirement to tailor the treatment plan to the needs of the individual. This again highlights the importance of research into OI and the availability of information. Without freely available research and information, there is a risk that HCPs without specialist

knowledge would not be able to provide suitable care for people with OI and their health would be put at risk.

The lack of knowledge of HCPs not only has an impact on the treatment options available but can also affect how the HCP is viewed by the person with OI. There was discussion around fear associated with OI and part of that was a fear of injury, both of injuries in general but also injuries caused by HCPs. These injuries could occur as a result of HCPs not being aware of how easily fractures can occur or of not knowing how to safely move a person with OI. The need for education of OI to HCPs cannot only be limited to the treatments or comorbidities, but also how to prevent harm. HCPs are trained in manual handling of patients and are trained to ensure they are safe, it is imperative that they be prepared to handle people with OI as fractures can occur easily and given the difficulties in accessing services such as physiotherapy, avoiding fracturing is a vital method of protecting the health and quality of life of people with OI.

There was also a fear of comorbidities discussed which links both with some HCPs lack of knowledge and patient knowledge. Without OI knowledge, HCPs may not know what they should look out for, which tests need to be run or how to prevent issues from developing. Without OI knowledge, people with OI will not know how they can protect themselves from comorbidities or how to spot early signs of issues developing. There is a need for partnership between HCPs and people with OI to ensure their health is preserved and protected but this is not possible without information being readily available.

A major factor impacting the lives of adults with OI is rooted in coherence within the health system. An issue whereby they were not able to see the same HCP on multiple occasions presented as a problem in the discussions. The result of this is that they would not have point of contact in the event of issues. Although they could go to their GP, there are a limited number of options available to GPs and the most likely is a referral to a specialist. Referrals can take a great deal of time, with many people in England having to wait over 52 weeks (Baker, 2022). During the wait for treatment, it is possible that the issue would worsen, and the medical issue would have a greater impact on their life.

A crucial part of managing OI is getting the right support. This would not be limited to support from family members, the support would also need to come from HCPs. This support would be needed both during the transition and whilst being seen in adult services. A smooth transition to adult services provides better health outcomes and would put the individual in a better position to manage their condition (Crowley et al., 2011).

In addition to support from family, support from the BBS is also needed to manage the condition. Although they offer some information, much of it is aimed at parents of children with OI and not adults with OI. This may be the result of assumptions surrounding the improvement of OI with age but as was seen by Kolovos et al. (2021), a large proportion of admissions into hospitals is that of people with OI over the age of 14. There is still a need for information and support after leaving paediatric services.

The matter of coherence was also raised in relation to treatments offered. It is likely that this issue is a result of the absence of guidance for treating OI and subsequently, there is no standard treatment offered to people with OI. The impact of the inconsistency in treatment offered is inequity between people with OI, with some being offered treatments and other being denied treatment. This would also be compounded by the lack of specialists and the resulting postcode lottery affect, whereby those who are fortunate enough to live near a hospital with a specialist able to get the best treatments for OI. Those who do not live near specialists or those without the means to travel to receive specialist care must hope that their HCP is receptive to them and is willing to learn from their knowledge and experience. This further supports the need for OI specific guidance to allow for consistency and great equity.

The ability to share knowledge and experience with HCPs is contingent on their ability to advocate for themselves. This process was often described as a fight. This need to advocate or fight, for treatments appears to be necessary as soon as they enter adult services. This would not be easy for them as it would likely have been their parent's responsibility while they were in paediatric services. It is an important skill to have when dealing with a chronic condition to ensure they get the care they need (Grant & Pan, 2011).

Self-advocacy is essential as a way of managing the reduction in available services as an adult. Not having access to services such as physiotherapy can be very detrimental to bone health in OI. Without regular exercise, bones can become osteoporotic and become even more susceptible to fracturing (Sam & Dharmalingam, 2017). Without access to physiotherapy in adult services, there is an increased risk of osteoporosis developing as they may no longer be able to engage in regular activity. Standard exercise methods such as visiting a gym or swimming may not be possible and subsequently, they would be placed at risk.

The need for self-advocacy links back to the discussion of educating HCPs. In order to do this, people need to be able to stand up for themselves and be willing to fight for their right to appropriate care. Despite the work of the BBS and their efforts to assist people with OI by recommending interventions such as bone density scans, as there are not guidelines in place, the responsibility would still fall on the person with OI to push for what they needed.

The responsibility of managing care would be an arduous task, especially when first leaving paediatric services. The ability to manage care, appointments and education of OI for both themselves and HCPs would be very time consuming and tiring over time (Kleman et al., 2021).

In order to mitigate the burden of managing OI, some would rely on the support of others with the condition and would seek to learn from their experiences. There are a number of Facebook groups for OI, these are akin to virtual support groups. These are very beneficial as they are available at any time of day and are not limited by geography. This method of sharing experiences is a very valuable tool and could prove to be a helpful learning tool for HCPs.

The involvement in other research projects was also discussed with multiple participants indicating that they were actively involved in research for OI. It is possible that they have a desire to engage in research as a result of negative experiences and do not want others to share them in the future. This would suggest that not all people with OI have negative experiences in the health system and given the small sample size, the experiences and issues discussed could be that of the minority of the OI population.

It is also possible that they were more likely to engage in research as they had previously utilised the BBS or the Facebook groups as a source of advice and support and as a result saw this research project and wanted to participate to share their stories. Those with positive experiences may not have the same need of Facebook groups or the BBS and as a result, did not see the recruitment post for this research.

Conversely, they may have wanted to participate in this project or others to increase their understanding of OI. This draws back on the issue of availability of information regarding OI and the need to learn about the condition in order to manage it.

3.4.4 Strengths and Limitations

An exploratory study investigating the transition to adult services and the ongoing management of care of adults with OI in the UK has not previously been conducted. There are some limitations to this study and the impact of these limitations has minimised where possible.

One limitation of this study relates to the application for ethical approval. Approval for this phase of the study was obtained from the ethics committee prior to recruitment. At which point it was hoped that multiple focus groups could be conducted. However, as only one participant attended the meeting, which was intended to be the second focus group, it became a one-to-one interview instead which was not stipulated in the ethics application. Following a discussion with the supervisory team, it was concluded that further recruitment for a second focus group or second one-to-one interview (depending on the success of the recruitment) could be conducted without amendments to the ethical application as there was an absence of ethical ramification of the change. However, in an ideal world and if this study was to be conducted again, amendments would have been made to the application for ethical approval, to gain approval for the additional recruitment and the interviews.

The small sample size of the focus group presented a potential limitation of this research. The recruitment phase was extended in order to attempt to obtain more participants but despite these attempts at further recruitment through Facebook groups and the BBS, there were only six participants in total for this phase. There was a significant amount of data gathered during these groups and this allowed for the development of an online semi-structured

survey to further explore the issues raised. There are many possible reasons for the small sample size. The method of recruitment, Facebook groups and the BBS social media accounts and website, would only reach a small population. These methods are contingent on suitable persons being present on those websites regularly.

This small sample size also resulted in a sample which did not include all of the home nations of the UK. There were three participants from England, two from Wales and one from Northern Ireland. There were no participants from Scotland and as a result any issues that are specific to that area were not identified. Despite all being part of the UK, there will most likely be differences between all of the home nations as health care is a devolved issue and therefore the structure and policies of each country can be tailored to meet the needs of that area.

Despite the low response rate, these methods of recruitment have been shown to be beneficial when attempting to access hard-to-reach populations (Topolovec-Vranic, 2016). Given the limited number of people in the UK who met the inclusion criteria, it can be argued that this method of recruitment was most appropriate for this research.

The use of an online focus group and interviews may have also negatively affected the sample size. This type of group has reported to have a higher no-show rate than in-person focus groups (Rupert, 2017). It was not possible to have in-person focus groups as a result of the coronavirus pandemic and therefore online focus groups were the only option. There are some limitations to using online focus groups and interviews. Firstly, some people are not as experienced in using technology and if they have not used software such as zoom before this will be a barrier to them. Secondly, technology does not always work, connection issues and issues with installation of software may prevent some from participating. Finally, online recruitment limits the available population as those who are not online will not be aware of the research study and will be prevented from participating.

Focus groups were initially chosen in lieu of one-to-one interviews because it was felt that the discussion between participants would allow for greater exploration of the topic and increase the likelihood of achieving data saturation. This method also reduces the need for prompting from the researcher as participants would be engaged in a conversation and

issues may be brought up by some participants that others would have also experienced but may not have thought to bring up themselves.

A benefit of the sample of this phase was that the participants did offer a diverse group, including ages range from 26 years to 66 years. It was felt that this was important to the research as it is important to understand both the historic experiences of people with OI and the current experiences of people with OI. This would allow for the research to learn from both previous best practices and past mistakes. However, it is important to remember that those experiences may not reflect the current practices and understandings of OI in the medical field.

There were both male and female participants, however there was a greater number of female participants. This was expected as it is understood that males are less likely to engage in research and male participants are harder to find. It was important to have both male and female participants as the experiences between them will differ. Both due to societal biases and also as some of their needs and health concerns will differ.

All of the common types of OI were represented, this was important as different types and different severities may have vastly different experiences and may face different barriers. Of the four UK nations, three were represented in the sample. This was vital as since devolution, the policies and practices in these nations may differ and therefore the recommendations or advice given to people with OI may not be applicable to all in the UK and may need to be tailored according to those countries policies.

3.5 Conclusion

This chapter has discussed the findings of the qualitative phase of the study and has explored the issues which were raised by the participants. The data has shown that transition is an issue for people with OI and that there are a number of other issues facing people with OI, including a lack of continuity in their care, misconceptions of the condition among HCPs, difficulties in accessing information, and the need for advocacy. These findings will be taken forward and used to frame the quantitative phase of the research. The following chapter will discuss the quantitative phase of the research and will further explore the issues raised in this chapter.

Chapter 4 – Quantitative Phase

4.1 Introduction

The previous chapter discussed the qualitative phase of the study and discussed the need for further investigation into the topics raised. The findings for the qualitative phase of the research have led to the generation of a number of research questions. The research questions have been broken down into primary and secondary priorities, the primary question of this research is "Do people who have a smooth transition report a better experience in adult services?". This question addresses the main issue which led to this study being conducted and was one of the main topics of discussion during the focus group and interviews. This topic was also central to the online semi-structured survey.

The secondary research questions allow for deeper exploration of other issues and topics which arose from the literature and during the focus group and interviews.

1. Is there a connection between continuity of care and how people rate their care in adult services?

2. How does knowledge of HCPs vary between child and adult services?

3. Is there a connection between poor care for adults and a lack of adult specialist services?

4. Is there a link between being dismissed by HCPs and the occurrence of missed fractures?

5. Is there a connection between missed fractures and poor HCP knowledge?

6. Are people who are members of the BBS more likely to have more knowledge and/ or to advocate for themselves?

7. Is there a connection between GPs not giving referrals when requested and whether adults have a consultant and/or regular appointments?

These research questions were discussed in the qualitative chapter and were a reflection of the major findings of the research thus far. The objective of this phase of the study is to further understand these research questions primarily from a quantitative epistemological approach, but will be supported with the use of open-ended questions to gather some qualitative data. This phase will further explore the issue of transition from paediatric services to adult services. This will be used to work towards achieving the aim of the thesis which is to investigate the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta. This will be achieved through the use of an online semi-structured survey that has been designed specifically for use in this research. The design of the online semi-structured survey will be discussed in the method section of this chapter. This phase of the study will also contribute to fulfilling the objectives of the research, including identification of any changes in care given between paediatric and adult services from the perspective of adults with OI, which was listed as the second transition objective in the previous chapter. This chapter will discuss the quantitative phase of the research, the results of the data collection and the implications of those results.

4.2 Method

4.2.1 Participants

The sample was recruited according to the specific inclusion and exclusion criteria laid out in the qualitative phase. The same requirements were made with regards to the participants and those had to be met in order of participate in the study. They had to be over the age of 21, had to live in the UK and had to have a diagnosis of osteogenesis imperfecta. In order to allow for generalisability, it was hoped that the sample for this phase of the study would be more diverse than that of the qualitative phase, however sample still had to fall within the bounds of the inclusion and exclusion criteria.

When calculating the intended sample size, data from the Office for National statistics and the Brittle bone society was used. The population size has been calculated using data from the Office for National Statistics. Based on their most recent data regarding age of the population, approximately 25.9% of the general population is under the age of 21 (Office for National Statistics, 2019). Applying this to the estimated number of people with OI in the UK,

4453 (which was calculated by dividing the general population figure by the estimated incidence of OI), only 3300 people in the UK have OI and are over the age of 21 (Brittle Bone Society, 2022).

This information was then used to calculate required sample size for differing confidence intervals and margins of error (Table 4). Based on these calculations and arguments made by Wilson Van Voorhis & Morgan (2007) regarding reasonable sample sizes, the intended sample size for the online semi-structured survey was between 50 and 100 participants as this would allow for an exploration correlation relationships between variables. Wilson Van Voorhis & Morgan (2007) stated that there could be no fewer than 50 participants. Given the small population and the challenges experienced during the recruitment process for the qualitative phase, it was felt that a target sample of between 50 and 100 participants would be satisfactory.

Table 4: Confidence interval, margin of error and required sample size for the quantitative phase

Confidence interval	95%	90%	95%	90%
Margin of Error	5%	5%	10%	10%
Ideal Sample Size	345	251	94	67

Participants were recruited through social media, the same procedure for recruitment was used for both the qualitative phase and the quantitative phase. With permission from gatekeepers, a recruitment post was shared in the Facebook group 'Osteogenesis imperfecta UK and Ireland (Adults)'. This was a pre-existing private group and members have to be approved by the administrators of the group in order to join. 45 participants took part in the online semi-structured survey with 31 completing the entire survey. There were multiple attempts made to increase the number of responses, the survey was shared in the OI Facebook group on several occasions and the BBS shared the study on their social media accounts multiple times too. Snowball recruitment was attempted, as the participants from the qualitative phase were reached out to and notified of the questionnaire. Type I, III, IV were represented in the sample and there were also options for people to identify has having

unknown types with those categories being mild, moderate, severe, and other and these were selected by 15 participants (Table 5).

Table 5: OI type distribution of the sample

What OI type do you have?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Type 1	13	28.9	28.9	28.9
Type 3	12	26.7	26.7	55.6
Type 4	5	11.1	11.1	66.7
Other	2	4.4	4.4	71.1
Unknown mild	6	13.3	13.3	84.4
Unknown moderate	5	11.1	11.1	95.6
Unknown severe	2	4.4	4.4	100.0
Total	45	100.0	100.0	

Types I and III were most common in the sample population with 13 and 12 participants respectively. With type I being a mild type, when combined with the unknown mild cases, there were a total of 19 people with a mild form of the condition in the sample or 42.2%. Type IV being a moderate type, using this same principle, combining with the unknown moderate, there were ten people with a moderate type in the sample or 22.2%. Severe forms of the condition, type III and unknown severe accounted for 14 people or 30.1%. The remaining 4.4% (two participants) selected other for their type of OI.

Age of diagnosis was also collected from the participants and can be seen in Table 6. The range of age of diagnosis was 0-35 years. The mean age was 3.31 years. The most common response was 0 years, accounting for 53.3%. 95.6% of participants were diagnosed at 14 years or younger. There were only two participants who were diagnosed in adulthood. Both of these participants stated they had an unknown mild type of the condition. Given the average age of diagnosis, the two participants who reported their diagnosis happening at ages 29 and 35, it can be assumed that this is not the norm for OI.

Table 6: Age at which the participants received their OI diagnosis

At what age was your OI diagnosed? - Years				
	Frequency	Percent	Valid Percent	Cumulative Percent
0	24	53.3	53.3	53.3
1	4	8.9	8.9	62.2
2	5	11.1	11.1	73.3
3	1	2.2	2.2	75.6
4	2	4.4	4.4	80.0
5	1	2.2	2.2	82.2
7	2	4.4	4.4	86.7
8	1	2.2	2.2	88.9
9	1	2.2	2.2	91.1
10	1	2.2	2.2	93.3
14	1	2.2	2.2	95.6
29	1	2.2	2.2	97.8
35	1	2.2	2.2	100.0
Total	45	100.0	100.0	

51.1% of participants (23/45) stated they were the first in their family to have OI. 31.1% (14/45) of people stated one or more of their parents had OI. 20% (9/45) of people stated they had another family member with OI. 17.8% (8/45) stated they have one or more siblings who also have OI.

As the demographics were not collected until the end of questionnaire, we do not have the demographics for all 45 participants. Of the participants who completed these demographics, 28 out of 35 identified as female and 7 of 35 identified as male. These are 80% and 20% of the valid percent respectively. The home nations of the UK were all represented, of the 33 participants who made it to this question, 20 were from England, one from Scotland, six from Wales and six from Northern Ireland.

Ethical approval was sought and received from the College and Human Health Sciences Ethical committee. There were amendments requested from the ethics committee for this application, the only stipulation of the approval from the committee was that permissions from any relevant organisations to be forwarded to the chair of the ethics committee prior to advertising the survey.

4.2.2 Materials

The online semi-structured survey to be used in this phase of the study was designed using the results from the qualitative phase of the study. This data from the thematic analysis and using pre-existing questionnaires, the questions to be included were written and formulated into the questionnaire which was launched (Appendix 6). In order to formulate the questions, the themes developed from the qualitative phase were reviewed and the major findings from participants were considered. This then allowed for missing elements to be identified and this then would present an area of interest to be included in the online semi-structured survey. This process was repeated and was supported with the use of information gathered in the literature review and from existing questionnaires.

There were two pre-existing questionnaires that were used as a basis for this survey, firstly was The Generic Short Patient Experiences Questionnaire (GS-PEQ) (Sjetne, 2011). The element of this survey which was used for the development of the survey for this research was the use of Likert scales. These scales are useful for quantifying data which would otherwise be difficult to measure. For example, when asking whether participants felt they were respected by their HCP, if this question had given the options of 'yes' or 'no', participants may have been unsure of how to respond if they had some HCPs who did respect them and some who did not. The use of the Likert scales offered more flexibility in the responses.

The second was the Ready Steady Go questionnaire used by Southampton Children's Hospital (Nagra, 2015). The structure of this questionnaire was used as a basis for the development of the survey used in this research. This survey had divided questions into a number of domains, some of which included knowledge, self-advocacy and transfer to adult care. It was felt that

this structure would make the survey easy to follow and would increase the likelihood of participants answering all of the questions included.

The rest of the survey, including the domains to be included and the questions within those domains was designed from the results of the qualitative phase.

4.2.3 Design

This phase of the research was designed to be an online semi-structured survey with an associative observational design. There were no conditions imposed and the sample was not compared to another group. This phase of the study was designed to be cross-sectional as there was only one point of data collection from the participants, this method is effective for studying rare diseases as it does not require repeated access to a limited population (Rezigalla, 2020).

The use of a descriptive design can provide an understanding of characteristics and trends of the population but cannot draw conclusions on cause and effect. A number of variables have been considered which may affect the results, including type of OI, age, gender, and country of residence. These variables were compared to the results of the questions in the online semi-structured survey in order to test whether they were related to findings such as support during transition and knowledge.

4.2.4 Procedure

The online semi-structured survey was disseminated online using Qualtrics (Qualtrics XM - Experience Management Software, 2023). It was decided to have virtual semi-structured survey as this method of data collection had several benefits and few limitations.

Firstly, online surveys and questionnaires are quicker to disseminate, and responses can be received straightaway, whereas the time for hardcopy responses to be received would be dependent on the speed at which they were sent back. Secondly, the cost of printing and sending hardcopy questionnaires to prospective participants would be very high whereas an online survey or questionnaire can be run at no cost to the researcher.

A potential and significant limitation of using an online survey is the inability to confirm the participant fulfils the inclusion criteria. In this case, it was not possible to confirm the participants had OI, were over the age of 21 or lived within the UK. If this happened, it may affect the quality of the data. The data will be checked for any potential anomalous results in order to mitigate the risk of data being skewed as a result of this.

The online semi-structured survey was launched on Qualtrics on the 12th of May 2021 and was closed on the 30th of December 2021. This was to allow as many responses as possible and to allow the number of participants to increase which would improve the reliability of data collected.

The online semi-structured survey was shared in a UK based OI Facebook group numerous times during this period to try and reach as many of the members of the group as possible. The recruitment post used in this Facebook group (Appendix 7) included information about the online semi-structured survey and contact information to allow prospective participants to ask questions prior to completing the online semi-structured survey. The online semi-structured survey was also shared by the BBS, it was posted in the research section of the BBS website and shared the online semi-structured survey on their social media accounts periodically during that time period.

At the beginning of the questionnaire, participants were asked to read the participant information page and the consent page (Appendix 8). In this page participants were asked to confirm they consented to participating and met minimum age requirement of 21 years old. If they selected 'No' on this page, they were not able to complete the survey.

After completing the survey, participants were taken to a debrief page (Appendix 9) which offered contact details for researchers to allow for participants to ask questions and follow up with the research and also provided them with the link to the BBS website in case they wanted further information on current OI services available in the UK.

4.2.5 Analysis

The analysis of the data gathered in the online semi-structured survey was centred around the key research questions developed for this research. These research questions were

formulated using the aims and objectives of this research study, the existing literature and the data gathered in the focus group and allow for a more focused analysis of the data gathered, these questions are listed in the introductory section of this chapter. It was these eight questions which framed the analysis and determined which statistical tests would be run on the data. The data was moved over from Qualtrics and the analysis was done using SPSS software. As the majority of the questions were closed-ended, quantitative data was gathered, and a number of tests could be run on this data. The remaining open-ended questions yielded qualitative data and content analysis was conducted on this data.

Prior to analysing the quantitative data, the power of the data was calculated using the sample size, the population size, and confidence interval, and it was found that there is enough power in the sample to detect a medium to large effect size for a one-tailed hypothesis at 80% power. Before attempting to run any inferential statistics, descriptive statistics tests were conducted, such as frequencies, means, median and range. Following this, cross tabulations were run to allow for question data to be compared and the p-value for these crosstabulations were calculated using Fisher's exact test. This was chosen as in the tables generated during the analysis, there were more than two cells with a count lower than 5. The open-ended questions in the survey yielded qualitative data which would be analysed first using the themes and sub-themes that were developed through the content analysis of the focus group data. Following this, the data would be reviewed for any new themes that were not present in the focus group or interviews. This was done as the purpose of the online semi-structured survey was to explore whether the same issues and concerns which were raised by participants in the qualitative phase, were consistently present across a larger sample of the OI community.

4.3 Results

As the online semi-structured survey generated both quantitative and qualitative data, a number of different analyses needed to be conducted. This chapter section will explore the data gathered which will allow for a discussion of the findings later in this chapter, starting with the quantitative data and then moving on the qualitative data.

4.3.1 Quantitative Results

This section will discuss the quantitative findings of the online semi-structured survey and how these findings address the eight research questions which were developed following the focus group and interviews. This allows for a broader exploration of the issues raised during the focus group and interviews. Each of the research questions will be explored in turn with findings from various questions in the survey being utilised.

4.3.1.1 Do people who have a smooth transition report a better experience in adult services?

The first research question which was explored was the primary research question, 'Do people who have a smooth transition report a better experience in adult services'. This question is central to the research as a whole. In order to explore this issue, the data from a number of questions needed to be explored to allow for a picture to be developed of the experience in adult services for people with OI.

Firstly, the data from the questions directly related to the transition to adult services were analysed. Participants were asked if they had support when being discharged from paediatric services. 34 of 43 participants stated that they did not have support from healthcare professionals when being discharged. This data was then compared to the question which asked people to directly compare their experiences in paediatric services and adult services (Table 7). 31 of 43 participants who answered both of these questions stated that care was better in paediatric services.

*Table 7: Crosstabulation - Did you have support from healthcare professionals when being discharged from paediatric services? * Do you feel the care you received for your OI was better as a child or not?*

Did you have support from healthcare professionals when being discharged from paediatric services? * Do you feel the care you received for your OI was better as a child or not? Crosstabulation					
		Do you feel the care you received for your OI was better as a child or not?			Total
		Yes - it was better	No - it was not better	No difference	
Did you have support from healthcare professionals when being discharged from paediatric services?	Yes	9	0	0	9
	No	22	5	7	34
Total		31	5	7	43

The chi-square finding for this crosstabulation (table 8), using fisher's exact test showed a two-tailed p-value of 0.134 which is not statistically significant.

Table 8: Chi-square analysis for table 7 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	3.356	0.134
N of Valid Cases	43	

The question of support from HCPs during discharge from paediatric services was then compared to the responses regarding overall experience in adult services (Table 9).

*Table 9: Crosstabulation - Did you have support from healthcare professionals when being discharged from paediatric services? * How would you rate your overall experience in adult services?*

Did you have support from healthcare professionals when being discharged from paediatric services? * How would you rate your overall experience in adult services? Crosstabulation							
		How would you rate your overall experience in adult services?					Total
		Excellent	Good	Average	Fair	Poor	
Did you have support from healthcare professionals when being discharged from paediatric services?	Yes	1	2	2	2	1	8
	No	0	5	6	3	14	28
Total		1	7	8	5	15	36

The chi-square finding for this crosstabulation can be seen in table 10, using fisher's exact test showed a two-tailed p-value of 0.103 which is not statistically significant.

Table 10: Chi-square analysis for table 9 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	6.587	0.103
N of Valid Cases	36	

36 of the participants completed both of these questions and of those 36, 28 stated they did not have support from HCPs when being discharged from paediatric services and half of those participants also rated the care they received in adult services as poor. Only eight of these 36 participants stated they had support during their transition, there was not a clear pattern in this group as responses were split across all options.

The responses to the questions were further examined by crosstabulating them with the results for the OI type question, this was done as it would allow for patterns in the care for different types of OI to be identified and explored. Table 11 shows the crosstabulation between the question regarding support during their discharge from paediatric services and OI type. Table 11 showed that all of the OI types represented by the sample, had some participants who stated that they did not received support when being discharged from paediatric services. For most of the OI types, more participants selected 'No' for this question, only the 'Unknown Severe' type had an equal number of 'Yes' and 'No' responses.

*Table 11: Crosstabulation - What OI type do you have? * Did you have support from healthcare professionals when being discharged from paediatric services?*

What OI type do you have? * Did you have support from healthcare professionals when being discharged from paediatric services? Crosstabulation				
		Did you have support from healthcare professionals when being discharged from paediatric services?		
		Yes	No	Total
What OI type do you have?	Type 1	3	9	12
	Type 3	3	8	11
	Type 4	1	4	5
	Other	0	2	2
	Unknown mild	0	6	6
	Unknown moderate	1	4	5
	Unknown severe	1	1	2
Total		9	34	43

Table 12 shows the chi-square finding for this crosstabulation, using fisher's exact test showed a two-tailed p-value of 0.807 which is not statistically significant.

Table 12: Chi-square analysis for table 11 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	3.725	0.807
N of Valid Cases	43	

4.3.1.2 Is there a connection between continuity of care and how people rate their care in adult services?

Another question which was asked in the online semi-structured survey was in regard to continuity of care. This issue was raised several times during the focus group and interviews. The majority of participants stated they did not feel they had experienced continuity in their care. In order to address whether this impacted their experience in adult services, the responses to both questions were compared (Table 13). Table 13 shows that those who stated they had not experienced continuity in their care also most commonly rated their overall experience in adult services as poor. Whereas, those who stated they had experienced continuity in their care as adults, rated their overall experience as either good or average.

*Table 13: Crosstabulation - As an adult, have you experienced continuity in your care? * How would you rate your overall experience in adult services?*

As an adult, have you experienced continuity in your care? * How would you rate your overall experience in adult services? Crosstabulation							
		How would you rate your overall experience in adult services?					Total
		Excellent	Good	Average	Fair	Poor	
As an adult, have you experienced continuity in your care?	Yes	0	3	2	0	0	5
	No	1	2	5	4	15	27
	Don't know	0	2	1	1	0	4
Total		1	7	8	5	15	36

The chi-square finding for this crosstabulation as shown in table 14, using fisher's exact test showed a two-tailed p-value of 0.007 which is statistically significant. This suggests that there is a correlation between continuity in care and overall experience of adult services.

Table 14: Chi-square analysis for table 13 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	15.846	0.007
N of Valid Cases	36	

4.3.1.3 How does knowledge of HCPs vary between child and adult services?

As knowledge of OI was a prevalent theme in the focus group and in the interviews, it was important to explore the issue regarding a change between the level of knowledge between paediatric care and adult care. In order to explore this, participants were asked a number of questions regarding knowledge of OI, not only their own knowledge but also their impression of the level of knowledge held by their consultants. Firstly, participants were asked to retrospectively rate the knowledge of their consultant in paediatric services. The results (table 15) show that the majority of participants rated their paediatric consultant's knowledge as either 'Excellent' or 'Good'. Of the 45 responses to this question, seven rated their paediatric consultant's level of knowledge as poor.

Table 15: How would you rate your paediatric consultant's knowledge of OI?

How would you rate your paediatric consultant's knowledge of OI?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Excellent	14	31.1	31.1	31.1
Good	14	31.1	31.1	62.2
Average	10	22.2	22.2	84.4
Poor	7	15.6	15.6	100.0
Total	45	100.0	100.0	

In addition to rating their paediatric consultant's knowledge of OI, participants were also asked to rate the knowledge of their adult consultant. These findings can be seen in table 16. This table shows that fewer than 50% of participants would rate their adult consultant's knowledge as either 'Excellent' or 'Good', in comparison with the 62% rated as 'Excellent' or 'Good' in paediatrics.

Table 16: How would you rate your consultant's knowledge of OI?

How would you rate your consultant's knowledge of OI?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Excellent	5	11.1	12.2	12.2
Good	15	33.3	36.6	48.8
Average	12	26.7	29.3	78.0
Fair	3	6.7	7.3	85.4
Poor	6	13.3	14.6	100.0
Total	41	91.1	100.0	
Missing		4	8.9	
Total		45	100.0	

These questions were then crosstabulated in order to establish whether any correlation between the ratings of paediatric and adult consultant existed (Table 17). This shows that only three participants rated the knowledge of their consultants as excellent for both paediatric services and adult services. The most common combination of response was 'Excellent' for paediatric services and 'Good' for adult services. This would suggest that the level of OI knowledge decreases between these services, this could be due to the lack of specialist OI services for adults with OI.

*Table 17: Crosstabulation - How would you rate your paediatric consultant's knowledge of OI? * How would you rate your consultant's knowledge of OI?*

How would you rate your paediatric consultant's knowledge of OI? * How would you rate your consultant's knowledge of OI? Crosstabulation							
		How would you rate your consultant's knowledge of OI?					Total
		Excellent	Good	Average	Fair	Poor	
How would you rate your paediatric consultant's knowledge of OI?	Excellent	3	8	2	0	0	13
	Good	1	2	5	2	1	11
	Average	1	2	4	0	3	10
	Poor	0	3	1	1	2	7
Total		5	15	12	3	6	41

Table 18 shows the chi-square finding for this crosstabulation, using fisher's exact test showed a two-tailed p-value of 0.093 which is not statistically significant.

Table 18: Chi-square analysis for table 17 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	16.012	0.093
N of Valid Cases	41	

4.3.1.4 Is there a connection between poor care for adults and a lack of adult specialist services?

As was discussed in previous chapters, these is no specialist OI service for adults in the UK and as a result there are a limited number of specialists for adults with OI. It was important to explore this and the potential consequences of the lack of services. In order to address this

research question, participants were asked whether they had a consultant who is an OI specialist, as an adult (table 19). There were 42 responses to this question, this table shows that 76% of the respondents do not have a consultant who is an OI specialist.

Table 19: Do you have a consultant who is an OI specialist?

Do you have a consultant who is an OI specialist?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Yes	10	22.2	23.8	23.8
No	32	71.1	76.2	100.0
Total	42	93.3	100.0	
Missing	3	6.7		
Total	45	100.0		

As was previously discussed, participants were also asked to rate their overall experience in adult services. Table 20 shows the responses to this question. There were 36 responses to this question, 41.7% of the respondents stated that their experience had been poor.

Table 20: How would you rate your overall experience in adult services?

How would you rate your overall experience in adult services?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Excellent	1	2.2	2.8	2.8
Good	7	15.6	19.4	22.2
Average	8	17.8	22.2	44.4
Fair	5	11.1	13.9	58.3
Poor	15	33.3	41.7	100.0
Total	36	80.0	100.0	
Missing	9	20.0		
Total	45	100.0		

In order to answer the research question, 'Is there a connection between poor care for adults and a lack of adult specialist services?', the results from the question regarding adult consultants and the overall experience in adult services were crosstabulated (table 21).

*Table 21: Crosstabulation - How would you rate your overall experience in adult services? * Do you have a consultant who is an OI specialist?*

How would you rate your overall experience in adult services? * Do you have a consultant who is an OI specialist? Crosstabulation				
		Do you have a consultant who is an OI specialist?		Total
		Yes	No	
How would you rate your overall experience in adult services?	Excellent	0	1	1
	Good	2	5	7
	Average	4	4	8
	Fair	1	4	5
	Poor	1	14	15
Total		8	28	36

The chi-square finding for this crosstabulation (table 22), using fisher's exact test showed a two-tailed p-value of 0.147 which is not statistically significant.

Table 22: Chi-square analysis for table 21 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	6.140	0.147
N of Valid Cases	36	

Table 21 shows that, of the 36 participants which responded to both questions, 14 stated that they did not have a consultant who is an OI specialist and their experience in adult services has been poor, this equates to 39%. Only one participant stated that their experience in adult services was excellent, this participant also stated that they did not have a specialist consultant. This suggests that it is possible to have a positive experience in adult services without a specialist. It is not possible to identify how their experience was 'Excellent', but this would be important to explore.

4.3.1.5 Is there a link between being dismissed by HCPs and the occurrence of missed fractures?

During the focus group and interviews, participants had discussed how they had been dismissed by HCPs and the occurrence of fractures being missed by HCPs. The online semi-structured survey included questions on both of these issues, and this allows potential correlations to be explored. Participants were asked about experiences of being dismissed or doubted by HCPs (table 23). This question gave the options of 'Yes' or 'No', 36 of the 39 which responded selected 'Yes' to this question. Which suggests that being dismissed or doubted is common for people with OI.

Table 23: Have you been dismissed or doubted when raising concerns to healthcare professionals?

Have you been dismissed or doubted when raising concerns to healthcare professionals?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Yes	36	80.0	92.3	92.3
No	3	6.7	7.7	100.0
Total	39	86.7	100.0	
Missing	6	13.3		
Total	45	100.0		

Participants were then asked whether they had experiences of fractures being missed by HCPs (table 24). In response to this question, 32 out of the 39 selected 'Yes', this suggests that for this sample of people, fractures being missed is not uncommon.

Table 24: Have you had fractures missed by healthcare professionals?

Have you had fractures missed by healthcare professionals?				
	Frequency	Percent	Valid Percent	Cumulative Percent
Yes	32	71.1	82.1	82.1
No	7	15.6	17.9	100.0
Total	39	86.7	100.0	
Missing	6	13.3		
Total	45	100.0		

These questions were then crosstabulated (table 15), this shows that 29 of the 39 which responded to these questions selected 'Yes' for both questions. This would suggest a correlation between being dismissed or doubted by HCPs and the incidence of fractures being missed by HCP'.

*Table 25: Crosstabulation - Have you been dismissed or doubted when raising concerns to healthcare professionals? * Have you had fractures missed by healthcare professionals?*

Have you been dismissed or doubted when raising concerns to healthcare professionals? * Have you had fractures missed by healthcare professionals? Crosstabulation				
		Have you had fractures missed by healthcare professionals?		Total
		Yes	No	
Have you been dismissed or doubted when raising concerns to healthcare professionals?	Yes	29	7	36
	No	3	0	3
Total		32	7	39

The chi-square finding for this crosstabulation (table 26), using fisher's exact test showed a two-tailed p-value of 1.000 and a one-tailed p-value of 0.543. Neither of these results are statistically significant.

Table 26: Chi-square analysis for table 25 crosstabulation

Chi-Square Tests			
	Value	Exact Sig. (2-sided)	Exact Sig. (1-sided)
Fisher's Exact Test		1.000	0.543
N of Valid Cases	39		

4.3.1.6 Is there a connection between missed fractures and poor HCP knowledge?

In addition to exploring whether the incidence of missed fractures was correlated to being dismissed or doubted by HCPs, the incidence of missed fractures was also compared to the results regarding consultant knowledge of OI in adult services. The previous section showed that 82% (32 out of 39 respondents) had experienced fractures being missed, previous results also showed that the level of knowledge held by consultants in adult services was most commonly rated as poor. In order to explore any correlation between missed fractures and poor HCP knowledge of OI, these questions were crosstabulated (table 27).

*Table 27: Crosstabulation - Have you had fractures missed by healthcare professionals? * How would you rate your consultant's knowledge of OI?*

Have you had fractures missed by healthcare professionals? * How would you rate your consultant's knowledge of OI? Crosstabulation							
		How would you rate your consultant's knowledge of OI?					Total
		Excellent	Good	Average	Fair	Poor	
Have you had fractures missed by healthcare professionals?	Yes	4	11	8	3	6	32
	No	1	3	3	0	0	7
Total		5	14	11	3	6	39

Table 28 shows the chi-square finding for this crosstabulation, using fisher's exact test showed a two-tailed p-value of 0.765 which is not statistically significant.

Table 28: Chi-square analysis for table 27 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	2.309	0.765
N of Valid Cases	39	

Table 27 shows there were incidents of fractures being missed at all levels of consultant knowledge. For those who stated that they had not experienced fractures being missed by HCPs, none rated their consultant's level of knowledge as 'Fair' or 'Poor'.

4.3.1.7 Are people who are members of the BBS more likely to have more knowledge and/ or to advocate for themselves?

The BBS and their role was discussed in the focus group, therefore it was important to ask about the BBS in the online semi-structured survey and to explore the impact of the BBS on adults with OI. In order to answer the research question above, the question asking whether the participants were members of the BBS needed to be crosstabulated with a number of questions. Firstly, the responses to the question regarding the participants knowledge of OI was crosstabulated with the responses regarding membership of the BBS (table 17). This table shows that of the 39 participants that responded to this question, 35 are members of the BBS. Of the participants who reported being members of the BBS, the majority of participants who rated their knowledge of OI as either 'Excellent' or 'Good'. Those who stated that they are not members of the BBS, 75% of those also reported their knowledge as excellent with the remaining participant rating their knowledge of OI as average.

*Table 29: Crosstabulation - Are you a member of the brittle bone society or social media groups for OI? * How would you rate your knowledge of OI?*

Are you a member of the brittle bone society or social media groups for OI? * How would you rate your knowledge of OI? Crosstabulation							
		How would you rate your knowledge of OI?					Total
		Excellent	Good	Average	Fair	Poor	
Are you a member of the brittle bone society or social media groups for OI?	Yes	10	20	3	1	1	35
	No	3	0	1	0	0	4
Total		13	20	4	1	1	39

The chi-square finding for the crosstabulation in table 29, using fisher's exact test showed a two-tailed p-value of 0.143 which is not statistically significant.

Table 30: Chi-square analysis for table 29 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	7.457	0.143
N of Valid Cases	39	

Secondly, the responses to the question regarding sharing opinions with HCPs was crosstabulated with the responses regarding membership in the BBS (table 31). This table shows that, despite their membership status with the BBS, the majority of participants selected 'Sometimes' or lower for the frequency with which they share their opinion with HCPs. This suggests that not all participants who are members of the BBS had sufficient confidence and/or knowledge to share their opinion with their HCPs.

*Table 31: Crosstabulation - Are you a member of the brittle bone society or social media groups for OI? * Do you share your opinion on these treatments with your healthcare professionals?*

Are you a member of the brittle bone society or social media groups for OI? * Do you share your opinion on these treatments with your healthcare professionals? Crosstabulation							
		Do you share your opinion on these treatments with your healthcare professionals?					Total
		Always	Often	Sometimes	Rarely	Never	
Are you a member of the brittle bone society or social media groups for OI?	Yes	5	6	14	4	6	35
	No	0	0	2	1	1	4
Total		5	6	16	5	7	39

The chi-square finding for this crosstabulation (table 32), using fisher's exact test showed a two-tailed p-value of 0.939 which is not statistically significant.

Table 32: Chi-square analysis for table 31 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	2.074	0.939
N of Valid Cases	39	

Finally, responses for the question regarding membership in the BBS was crosstabulated with the responses regarding the need for advocacy (table 33). This shows that on the whole, the majority of participants selected 'Sometimes' or higher for the frequency with which they need to advocate for themselves. The results between the groups who are members of the BBS and those who are not members of the BBS are not consistent. The group who are not members are equally split between 'Never' and 'Often'.

*Table 33: Crosstabulation - Are you a member of the brittle bone society or social media groups for OI? * Have you ever had to advocate with healthcare professionals for treatments/ tests to be provided?*

Are you a member of the brittle bone society or social media groups for OI? * Have you ever had to advocate with healthcare professionals for treatments/ tests to be provided? Crosstabulation							
		Have you ever had to advocate with healthcare professionals for treatments/ tests to be provided?					Total
		Always	Often	Sometimes	Rarely	Never	
Are you a member of the brittle bone society or social media groups for OI?	Yes	3	16	10	3	3	35
	No	0	2	0	0	2	4
Total		3	18	10	3	5	39

Table 34 shows the chi-square finding for the crosstabulation in table 33, using fisher's exact test showed a two-tailed p-value of 0.169 which is not statistically significant.

Table 34: Chi-square analysis for table 33 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	4.823	0.169
N of Valid Cases	39	

4.3.1.8 Is there a connection between GPs not giving referrals when requested and whether adults have a consultant and/or regular appointments?

The online semi-structured survey included questions regarding appointments and availability to get referrals to hospital services. The impact of GP's on access to other services was explored through several questions which have been crosstabulated. Firstly, the question regarding GP referrals was crosstabulated with the question which asked whether the participants have had regular appointments with a consultant since entering adult services (table 35).

*Table 35: Crosstabulation - Has your GP referred you to appropriate services when needed? * Have you had regular appointments with a consultant under adult services or not?*

Has your GP referred you to appropriate services when needed? * Have you had regular appointments with a consultant under adult services or not? Crosstabulation				
		Have you had regular appointments with a consultant under adult services or not?		Total
		Yes	No	
Has your GP referred you to appropriate services when needed?	Always	5	2	7
	Often	5	5	10
	Sometimes	5	6	11
	Rarely	1	3	4
	Never	1	3	4
Total		17	19	36

The chi-square finding for this crosstabulation (table 36), using fisher's exact test showed a two-tailed p-value of 0.552 which is not statistically significant.

Table 36: Chi-square analysis for table 35 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	3.156	0.552
N of Valid Cases	36	

The participants are split fairly equally between 'Yes' and 'No', 17 respondents and 19 respondents respectively, for the question regarding regular appointments, and more participants reported that they 'Sometimes' or 'Often' receive referrals to appropriate services from GP's when they are needed. The responses to the question regarding appropriate referrals from GP's was then crosstabulated with the question regarding referrals when they have been requested by the participant (table 36). Table 36 shows that 'Sometimes' was the most common response for both of these questions and only three respondents selected 'Always' for both questions. There were two respondents who selected 'Never' for both questions.

*Table 37: Crosstabulation - Has your GP referred you to appropriate services when needed? * Have you been able to get referrals to appropriate services when you have needed or asked for them?*

Has your GP referred you to appropriate services when needed? * Have you been able to get referrals to appropriate services when you have needed or asked for them? Crosstabulation							
		Have you been able to get referrals to appropriate services when you have needed or asked for them?					Total
		Always	Often	Sometimes	Rarely	Never	
Has your GP referred you to appropriate services when needed?	Always	3	4	0	0	0	7
	Often	1	5	3	1	0	10
	Sometimes	0	2	8	1	0	11
	Rarely	0	1	2	0	1	4
	Never	0	0	2	0	2	4
Total		4	12	15	2	3	36

Table 38 shows the chi-square finding for the above crosstabulation, using fisher's exact test showed a two-tailed p-value of 0.005 which is statistically significant. This shows that there is a correlation between receiving referrals from GPs and being able to access the appropriate services.

Table 38: Chi-square for table 37 crosstabulation

Chi-Square Tests		
	Value	Exact Sig. (2-sided)
Fisher-Freeman-Halton Exact Test	24.655	0.005
N of Valid Cases	36	

4.3.2 Qualitative Results

This section will explore the qualitative data gathered through the online semi-structured survey. There were 16 questions which gathered qualitative data in the survey, this section will go through the results of those questions and will consider whether there are similarities between the findings of the interviews and focus group, and the survey.

The first question to gather qualitative data in the online semi-structured survey asked for participants to describe the ways in which the care they received as children was better, if at all. The most common comment that was made by participants was in regard to the frequency of appointments they had as a child. Statements such as *"I received regular care and treatments as a child"* and *"I had annual check ups"* appeared regularly. Of the 29 responses to this question, 12 expressed that they used to have regular appointments as a child. With some stating that they are no longer under the care of a consultant. In addition to this reference to the 'Referrals to specialists and regular appointments' sub-theme from the qualitative phase, several of the participants also expressed that they struggle to access care. Responses to this question made reference to the ease of access and the *"quick access to surgeons and specialists".* The consistency of services and better support as a child was also discussed in multiple responses, it was stated that HCPs in paediatric services "*were consistent with care"* and were "*willing to listen to any concerns".*

The next question asked participants to explain how they were supported during their transition. Responses to this question were mixed with some reporting a positive experiences, one participant *"met [their] adult consultant before moving services"* and another reported that they had "*regular check ups".* Conversely there were some participants who reported that they "*didn't have any support"* and another *"was discharged and told to go to [their] GP to ask for a referral as an adult if they had a problem".*

Knowledge of OI was mentioned in several of the questions, one question asked participants to explain how they felt their consultants level of knowledge of OI had affected their care. A recurring issue raised by the participants was a lack of knowledge of OI by their consultants. Some of these issues might arise when "*OI is 'new' to your doctor"* or when their *"consultant isn't a specialist".* One of the consequences of this is the need to "*correct doctors and nurses".* With this comes the need to advocate for themselves, some expressed that they "*question all decisions"*, not only did they need to question decisions made but they also felt that they *"had to fight for any help".* This could also be due in part to feeling "*abandoned"* by their consultant. This question also revealed that participants have difficulty trusting their consultants as one person expressed how they "*would like to trust [their] doctor"*.

Participants were also asked where they get information from if they are not given clear explanations from their HCPs, this question gave options and allowed for participants to enter a text response. The results for the pre-set responses can be seen in table 39.

Table 39: Primary sources of information for participants in the quantitative phase

	Brittle Bone Society	Osteogenesis Imperfecta Foundation	Other Websites	GP	Specialist/ Consultant	Other healthcare professional	Social media	Friends/ Family
Valid	22	14	1	2	7	2	10	9
Missing	23	31	44	43	38	43	35	36

In addition to those responses, there were three text responses to this question. One person stated they would speak to their son's OI specialist team at Bristol, another stated they had watched a programme on television and the last stated they would use "general web

searches". Following the question regarding sources of information, participants were asked where they would go first to access information. Those responses were gathered as qualitative data but can be seen in the figure below.

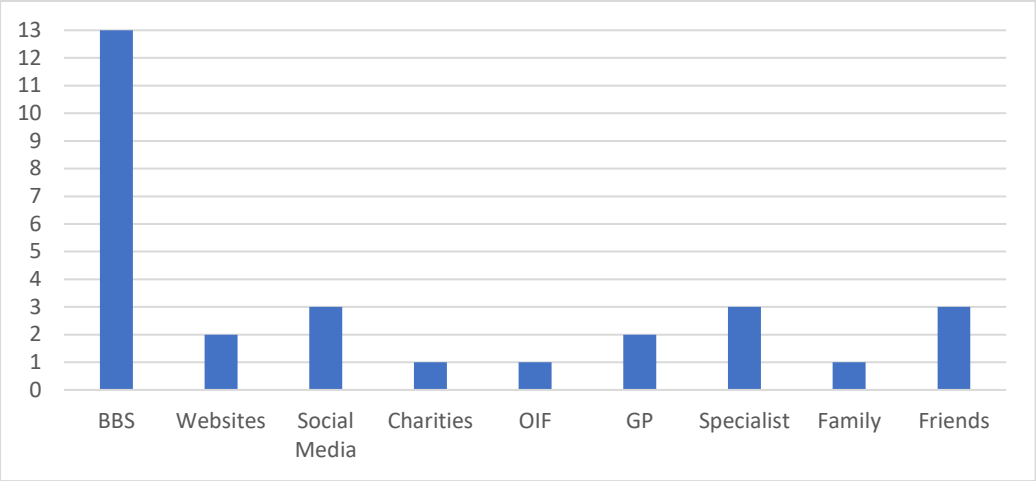

Figure 4: Sources of information and the frequency of use reported by participants in the quantitative phase

In addition to asking participants about the sources of their information, they were also asked what had prompted them to look for information about OI. The motivating factors behind looking for information about OI were varied, some wanted to "*be more knowledgeable regarding treatment options"* themselves, whereas others were driven by the desire to help family members or other people, stating reasons such as wanting "*to be able to help them understand".* There were some responses which stated the reason was that their "*health is deteriorating"* or that they have "*unexplained health problems"*. Which also connects with comments made which state that the reason for looking for information about OI was that they had "*concerns about [their] health"* and were "*worried about [their] future health"*. A contributing factor to this could be that participants feel there is a "*lack of help or advice from medics".*

Further to these discussions around availability of advice and issues of lack of information of OI by both individuals and HCPs, participants were asked whether they had experienced fractures being missed by HCPs and why they felt this had happened. In response to this question, there were two recurring issues. Firstly, participants reported that they felt HCPs *"never listened to [them]"* and *"didn't believe [they'd] fractured".* One participant reported

that a "*previous fracture [had] been disregarded"* with another stating they felt their HCP showed a "*lack of interest"* and was *"not listening to symptoms".* The second recurring issue in this question was a lack of knowledge of OI, specifically that the HCPs had a "*lack of knowledge about how to read x-rays"* and that the HCPs "*couldn't read the x-rays as things like hairline fractures aren't as clear with OI".*

Further to the discussions of knowledge in OI and the lack of knowledge held by some HCPs, participants were asked what they do when they receive conflicting advice from HCPs. The responses to this question were varied with some stating they would "*take the advice of the specialist above all"* and others state they would "*think about what would be best for [them]"* and acknowledge that they "*go with [their] own instinct".* The ability to trust their own instincts and advocate for themselves seen here was also seen in response to the question regarding continuity of care and how this affects care. Participants stated that they "*have to chase things up"* and this could be due to them "*not receiving any care"* which was an issue expressed by many of the participants. These issues of not receiving care were discussed further by participants in response to the question regarding referrals to services. Multiple participants stated that their GP "*wouldn't refer*" them to services they needed. The need for advocacy was also seen here as one participant stated that their "*persistence paid off".*

Participants also discussed the ways in which they have advocated in response to the question which asked for the ways in which they have advocated for their children if they have OI. Some of the driving factors for this advocacy could be due to parents with OI having *"learnt a lot from [their] experiences*" and feeling the need to "*fight for the correct treatments"*. Another participant reported that they had to push for monitoring of their child and that if they "*hadn't push for monitoring this [scoliosis] may have been missed*".

These responses regarding children with OI also highlighted the importance of MDTs, participants reported that these services were available for their children. When asked about these services for themselves, few participants reported having an MDT experience as an adult, one stated that they *"had [their] first multi-disciplinary appointment as an adult recently".* Participants felt that MDT appointments would be "*most beneficial*" for people with OI, this could be due to the fact that HCPs "*don't realise all the other problems that come with OI*". This would also allow for knowledge between HCPs to be shared which would

help as participants felt that "*nobody seems to know what to do"*. The absence of such services for adults left participants feeling "*extremely let down"* and they emphasize that "*it's not good to feel forgotten".*

Factors that could contribute to feeling let down by services include "*being dismissed by an arrogant consultant"* or the fact that some participants "*couldn't get [their] GP to do anything*". These issues could also be a contributing factor to the reports by participants that the lack of support "*can feel lonely".* These issues around the level of support they received were also addressed when participants were also asked how the BBS had helped them if they were members. The majority of responses were centred around the peer-support that they received as a member of the BBS. Participants stated that *"without the BBS I would have felt alone with my OI"* and that they see being a BBS member as "*being part of an extended family".* This peer support enabled them to see that "*other people go through the same things"* and this allowed them to develop a *"connection with other OIers"* and know that they *"are not alone".*

4.4 Discussion

The aim of this phase of the study was to further explore the experiences discussed in the qualitative phase and to consider the generalisability of the findings to a broader sample of people with OI. Specifically in regard to the transition to adult health services in the UK and the management of their on-going care as adults and to answer the research questions which were developed from the results of the qualitative phase. The findings of this phase have been consistent with the findings of both the literature review and the qualitative phase. This section will discuss these findings and will consider how these align with the findings from the literature review and form the qualitative phase. All of these findings were then to be used to design a patient guide for adults with osteogenesis imperfecta.

4.4.1 Existing literature

Although there is limited evidence for the issues raised in the research, there are some studies which have discussed both transition and ongoing care of adults with OI. As previously mentioned in the discussion of the literature in both chapter one and chapter three, these studies were not conducted in the UK, however, implications of issues relating to

poor transition and limited services for adults with OI will still be useful for comparison with the findings in this research. This section will discuss the findings of this phase of the research in relation to these existing studies in the field of OI and will also look at findings from research looking at transition for other chronic conditions.

The principal issue for this research is the transition from paediatric to adult services, the qualitative phase showed that transitions did not always occur and in this quantitative phase, the survey showed that 34 out of 43 participants did not receive support during their transition. The need for a smooth transition was highlighted a number of times throughout the literature with the drivers of this need being varied. Hill et al. (2019) discussed the emotional impact of transition, stating that the combination of the burden of illness, changes in care, changes in HCPs and the new setting would have a significant emotional impact on people with OI. This suggests that people who do not have a smooth or supported transition would have issues with these things and would experience an impact on their emotional wellbeing and this could present long term issues for adults with OI but this aspect was not discussed.

A way of combatting issues associated with transition is preparation which was discussed extensively in the existing literature. This preparation phase would be dependent on HCPs having an understanding of OI and passing their knowledge onto the individual. The results of the survey showed that individuals feel their paediatric consultants had more knowledge of OI compared to their adult consultants. This would suggest that HCPs are in a position to prepare individuals for their transition, but this did not occur for this sample. It was highlighted by multiple studies that preparation for transition needs to commence as early as possible to ensure a smooth transition. In studies by Dogba et al. (2014) and Shapiro & Germain-Lee (2012) transition is described as occurring at 18 years of age and 16 years of age respectively, despite this, the transition process begins at age 14. This gives both the HCPs and the individual ample time to prepare for the transition which would allow for a smooth transition to occur. Commencing preparation for transition early is not limited to OI, Hilliard et al. (2014) observed that early preparation is vital for a smooth transition and can allow for common transition issues to be dealt with and problems prevented. In addition to this, Gleeson & Turner (2012) argue that early preparation for transition should be seen as part of the best practice for transition.

The transition tool developed by Jeong et al. (2019) includes an assessment of readiness to transition. The importance of assessing the readiness of individuals to transition is also discussed by Dogba et al. (2014) who emphasised that without this assessment, potential barriers to transition cannot be identified and the individual could not be adequately prepared for their transition. The preparation for transition also extends beyond identifying HCPs and ensuring continuity of treatments as it also requires consideration of external challenges which may affect the transition. These factors were discussed by Stewart (2015) who argued that transition programmes need to cater to individual challenges associated with school, work or leaving home which could also influence where a person would receive their adult care. This supports the need for a comprehensive transition tool which takes into account the differences between the needs of people with OI.

Carrier et al. (2018) expressed how the preparation for transition to adult services is complex for people with OI and should be dealt with collaboratively with HCPs from various fields to allow for treatments to be prioritised and also establishes a support system for the individual as they would have multiple HCPs whom they would know and trust. This support during the transition is vital but the source of this support is not limited to HCPs. Jeong et al. (2019) discussed the importance of peer support and highlighted that a major part of their transition program involves the prioritisation of establishing peer support and connecting individuals and families with OI. The results of the survey showed that the majority of participants are members of the BBS (35 out of 39 respondents) which would suggest they are part of the OI community and have access to peer support. This need for peer support was reflected in the findings of the survey as multiple responses to the open questions reference the OI community and state that without this support network, they would experience loneliness. Shapiro & Germain-Lee (2012) emphasised that developing this support network can be difficult for adults with OI. Chung (2014) discussed these difficulties in relation to chronic conditions and stated that online support groups, such as Facebook groups, can be extremely useful in providing these networks and allowing people with the same conditions to support each other. This could also extend to organisations such as the BBS which have an online presence and allow people with OI to meet which may not otherwise be possible.

The results of the qualitative phase showed that continuity was an issue for adults with OI. The results of the survey suggest that this is the case for the wider OI community with 27 out

of 36 respondents stating that they had not experienced continuity in their care. In a study looking at the transition programme in Shriners Hospital for Children, Dogba et al (2014) discussed the importance of a smooth transition to adult services for people with OI not only to provide support for the person with OI but also to allow for continuity. They found that people with OI understood the need for them to leave paediatric services but wanted to remain in paediatric services. This could be due in part to knowledge that their care would change after leaving paediatric services. Carrier et al. (2018) describe the continuity as one of the cornerstones of the transition to adult services, they highlighted the importance of this as a way of ensuring good health outcomes for individuals. This argument was also made by Dogba et al. (2014) who state that a lack of continuity in care are one of the potential outcomes of a poor transition which can also lead to an increase in hospital admissions and an increase in morbidity. There are barriers to achieving continuity in care, especially during the transition to adult services, Shapiro & Germain-Lee (2012) recognised that there was great importance in ensuring continuity but that this would be difficult as many HCPs have little or no experience in dealing with OI. They suggest that communication between HCPs and collaboration, could enable continuity and this argument was also made by Jeong et al. (2019). Beyond the realm of OI, these arguments have been made generally in regard to continuity and chronic conditions. Iyengar et al. (2019) recommends communication not only to ensure continuity in care but also to ensure that the individual will be comfortable with their new HCP and will be able to trust that they not only understand OI but also that they have a good understanding of their individual medical needs.

HCP knowledge has been a recurring issue throughout this research. The findings of the survey are consistent with this finding with the knowledge of adult consultants being rated lower than that of the paediatric consultants. Having an understanding of OI was a significant issue throughout this research and this finding was mirrored by the literature. Hill et al. (2019) discussed how the issue of a lack of knowledge of OI is prevalent among GP's in the UK. This presents an issue as GP's act as a gatekeeper to other services within the NHS and without referrals from them, people with OI may not be able to access specialist services they need. Hill et al. (2019) also stated that outside of specialist services, there is limited OI knowledge and that HCPs have less experience dealing with OI. As there are fewer specialists dealing with OI in adults, people with OI would struggle to find a HCP who could provide the

same care as their paediatric HCPs. This would explain the finding by Dogba et al. (2014) who discussed that people with OI would prefer to remain in paediatric services and express frustration at the differences between paediatric and adult services. Similar findings were reported by Viner (1999) who report that one of the factors making the transition difficult is the reluctance of the paediatric team to let go of their patient. Their lack of trust in the adult services could be passed on to the individual and make them hesitant to trust their new HCPs.

A possible cause of this difficulty in trusting adult services could be the knowledge that adult services have less knowledge of chronic conditions such as OI and therefore cannot provide the same level of care as specialist paediatric services. The survey asked participants if they had an OI specialist as an adult and 32 out of 42 stated that their consultant was not a specialist. Not only could this explain why knowledge is reported to be lower among adult consultants when compared to paediatric consultants but in addition, this could explain the findings in other studies which report HCP knowledge as poor. Viner (2008) reported that adult consultants have little knowledge of congenital heart disease, although this is not directly comparable to OI, as a chronic condition, similarities can be drawn upon as issues around knowledge of the condition are fundamental to the treatment of the condition. Without this knowledge, HCPs would not be aware of best practices for the condition. This was also discussed by Rettig & Athreya (1991) who highlighted that some chronic conditions are seen as childhood conditions and as a result adult consultants and other HCPs may not have had adequate training for dealing with these conditions. In the case of OI, the idea of it as a childhood disease may be contributed to by the high number of specialist paediatric clinics which exist in contrast to the absence of specialist adult services.

This lack of specialists and specialist clinics was discussed in the literature and supports the findings of this research. Hill et al. (2019) discussed the impact of this on individuals with OI and expressed that being seen by an HCP who is not an expert in OI can amplify feelings of anxiety and distress which would increase the emotional burden on the individual. Without specialist management, the burden of managing the condition falls to the individual, as reported by Jeong et al. (2019). They found that people were left to fend for themselves when they were under the care of an HCP who was not an OI specialist. This discussion of specialists was not limited to specialist consultants, the benefit of specialist nurses was also

seen as an important part of both the transition out of paediatric services and in adult services. Carrier et al. (2018) stated that a strong transition programme requires an MDT, but specifically should include a specialist nurse who can facilitate the transition. Further to this they expressed that this role is also needed in adult services, as this would allow for adults with OI to be supported and have someone they can turn to in the event of fractures or other health issues. The importance of this role has also been recognised in the care of other chronic conditions. Research into the transition to adult services for people with diabetes conducted by Wagner et al. (2015) also found that the role of a specialist nurse was important. They state that this role is needed not only to coordinate care and facilitate the transition to adult services, but also to retain patients within the health service. Viner (1999) explain that having a specialist nurse is the practice in Great Ormond Street Hospital (GOSH) and that this reflects the needs for an MDT approach to both transition and adult care. Viner (1999) went on to explain that one of the reasons the role of the specialist nurse within the MDT is so important is because it would not be possible for all the needs of patients with complex chronic conditions to be fulfilled without this role. Therefore, the absence of this role in adult services for people with OI further supports the need for resources to provide information to people with OI.

The need for an MDT in the management was also discussed by Tosi et al. (2015). They discussed how the lack of specialists was recognised by the Osteogenesis Imperfecta foundation (OIF) and how they established a multidisciplinary committee whose objective was to conduct and facilitate research focused on issues facing adults with OI. This committee included HCPs and people with OI, this highlights the importance of involving people with the condition as they would have a different perspective to HCPs.

The qualitative phase showed that a consequence of being seen by HCPs who do not have a thorough understanding of OI is having concerns and questions ignored and being doubted by HCPs. This was observed in the online semi-structured survey responses as 36 out of 39 respondents said they had been dismissed or doubted by HCPs. Further to this, 32 out of 39 respondents reported having fractures missed by HCPs. These results were compared and 29 out of 39 respondents who stated they have been dismissed and had experiences fractures being missed. This would suggest a connection between these factors although the p-value from Fishers exact test could not support this. There were discussions of these issues in the

literature, when discussing the impact of being dismissed by HCPs, Hill et al. (2019) emphasised that having their concerns dismissed could amplify feelings of anxiety or distress which might discourage people from seeking help. In addition, Hill et al. (2019) being dismissed by HCPs can start early, as early as the point of diagnosis, at which point the effects of this are no longer limited to the individual with OI. In this case, family members would also have experiences of being dismissed or doubted by HCPs. This could have a negative effect on the mental health of both the individual and the family members. Tsimicalis et al. (2016) expressed that the psychosocial needs of both the individual and the family members need to be considered as a result of experiences like this as they can contribute to feelings such as anxiety, depression, and guilt. They argue that support systems need to be in place in order to preserve the mental health and wellbeing of people in these situations. The importance of these services was discussed by Viner (2008) and Nieboer et al. (2014) who both discussed the benefits of these systems in order to preserve and protect the voice of the individual which would in turn allow for a better relationship between individuals, family members and HCPs. This better relationship would facilitate better communication and could work to protect people from experiencing doubt or dismissal from HCPs.

This issues of dismissal from HCPs could be aided through an improvement of the relationship between individual and HCP. Better communication between individuals and HCPs would also be beneficial as it would give individuals the opportunity to express their opinions and treatments and other aspects of their care without needing to advocate for themselves. This need for advocacy was seen in the qualitative phase and in the findings of this phase. The survey results showed that only five out of 39 respondents reported never having to advocate for themselves, therefore 34 out of 39 had needed to advocate for themselves at some point in their lives. This finding is supported by the literature as many of the studies reviewed recognise advocacy as a skill which is developed during childhood and is further developed through a smooth and coordinated transition. Carrier et al. (2018) found that structured transition programmes had been associated with an increase in skills such as advocacy and self-management. These skills are vital for adults with OI, as was seen in the discussion of continuity and the lack of services, there are gaps in the current service provision which could be bridged by individuals if they had the necessary skills. Dogba et al.

(2014) made similar observations and suggested that a key part of preparation for transition should be ensuring the individual is able to advocate for themselves.

Tsimicalis et al. (2016) found in their review that children with OI developed skills needed to manage their condition at a young age, this included advocacy. This suggest that the development of these skills was done out of self-protection and was a necessity for children with OI. It could be argued that this might not be the case for children who are treated by specialist HCPs, as they would know how to treat a child with OI and perhaps the urge to protect themselves would arise only after encountering HCPs who do not have a deep understanding of OI. For any children who did not develop these skills, it would likely be a more difficult transition for them as they would not be adequately prepared for dealing with less knowledgeable HCPs. This highlights the need for education of both individuals and HCPs. This draws back on a point made by Hill et al. (2019) who stated that guidelines for transition need to prioritise preparing individuals to manage their condition themselves and to advocate for themselves when needed. Shapiro & Germain-Lee (2012) discuss how developing these skills at a young age would also ensure they were prepared for the transition to adult services, which as previously mentioned, can be very difficult due to the number of changes that occur simultaneously.

Advocacy is needed for adult services not only to allow adults with OI to get the referrals from their GP but to also have their voice heard during appointments with HCPs. The findings of this phase showed that only seven of the 39 respondents reported never sharing their opinion with HCPs. This would suggest that individuals with OI are happy to tell their HCP what they think. However, the finding of the high rate of people being dismissed must be considered. Although they do share their opinions at a high rate, they experience dismissal or doubt at a higher rate, and this may explain difficulties in obtaining both referrals and appointments. This was also seen in both the literature and the results of this research. Hill et al. (2019) state that a possible explanation for the difficulty facing people with OI when trying to obtain referrals and appointments could be the lack of knowledge of OI help by HCPs in adult services. This could be due in part to the previously discussed issue that some chronic conditions like OI are seen as childhood diseases. Dogba et al. (2014) describe how their transition programme aims to combat some of these issues by providing individuals being

discharged to their local care institutions with contact information for their paediatric team so they can remain in contact and be involved in their care if needed as adults.

Another issue with appointments and referrals is the burden they put on the individual. This was discussed by Hill et al. (2019) and Shapiro & Germain-Lee (2012), both expressed concern that the number of appointments individuals face under adult services could be overwhelming. This would be due to the fact that individuals are less likely to have MDT management as an adult and would need to have separate appointments. Hill et al. (2019) emphasized that this could create an unreasonable emotional burden on the individual and might make them less likely to engage in their care. Which in turn could lead to worsening issues and more comorbidities in the future. A high volume of appointments could also interfere with other everyday activities such as work, education, and social activities. Compromising these could affect their finances, future prospects and their mental health. Shapiro & Germain-Lee (2012) emphasised that adult care should be considerate of these issues. In regard to appointment load and appointment frequency, Iyengar et al. (2019) recommends quarterly appointments for adults with diabetes following the transition to adult services to allow for continuity and coordination of care. Conversely, Nakhla et al. (2017) advises that the perception of adult services held by HCPs in adult diabetes services is that appointments are less frequent for adults. In addition, they state that there is an expectation that appointments are less flexible, and patients should expect to have multiple appointments with different HCPs. This supports the concerns raised by Hill et al. (2019) and Shapiro & Germain-Lee (2012) and highlights the issues not only in the current services but also the expectation of those services.

Wagner et al. (2015) suggests that these issues are further support for the specialist nurse role in adult services, they can ensure adults with chronic conditions like OI are followed up and not overwhelmed by the volume of appointments they might require as an adult. Another potential barrier to appointments in adult services is the different structure utilised during appointments. Robertson (2006) discussed how these differences such as appointment length and having fewer notes on hand during the appointment might make adults less trusting during the appointment. Although the issue of availability of hard copies of notes should no longer be an issue, difficulty in accessing medical records has been

discussed in the findings of this research and may still be a barrier to ensuring people attend their appointments and feel able to trust their HCP.

4.4.2 Theories and Models

The theories and models that were explored as part of the discussion for the qualitative phase of the research, the MDM and the Movin' on up health care transition model were explored further in this phase. This was to assess in what ways they aligned with the findings and to identify any elements of these models that did not align with the findings.

The Movin' on up health care transition model was used for the transition aspect of the research. This was done as no one model or theory encapsulated all aspects of the research. This model explores different domains and considers how these domains can be interconnected. The findings of the online semi-structured survey supported several elements included in the Movin' on up health care transition model. The four intervention elements of the model were all found to be supported by the findings of this phase of the research. 'Teaching' was included as one of the desired interventions of transition and the need for education both of HCPs and individuals was prominent in the results. This would allow for better management of the condition and would allow for individuals to advocate for themselves which was also included as part of the individual domain of the model.

'Case management' is another intervention listed in the model and the importance of this was seen in the results. The need for management of adults with OI was highlighted as the majority of participants had not experienced this. In addition, another element of 'case management' was seen in the literature as the need for a dedicated point of contact in the form of a nurse specialist, the role of this person is to coordinate the care of individuals with OI and this would allow for the condition to be managed. This links to another element of the model, 'surveillance', the need for this was seen in the results as many of the participants do not have regular appointments but would need this in order for their condition to be properly managed and for comorbidities to be identified and treated swiftly. For the 'treatment and procedures' elements, this was also reflected in the results of the survey, as the results showed that people with OI are frequently dismissed by HCPs and have fractures missed.

In the qualitative phase, it was identified that some of the drawbacks of this model were the lack of individualisation and the time of the transition. Both of these elements were highlighted in the literature as important aspects of successful transitions. The absence of individualisation has been discussed in both the qualitative and quantitative phases of this research. The time of transition was not discussed as the majority of participants did not have an official transition between paediatric and adult services but is important to be considered.

As this model covers the majority of issues raised by the quantitative phase, the Movin' on up health care transition model will continue to be used for the discussion of issues related to the transition from paediatric services.

For the discussion of issues relating to the ongoing management of adults with OI in the qualitative phase the MDM was used. This model was used to assess the findings of the quantitative phase of the research. There are several elements of this model which align with the findings of the research. Firstly, the burden of illness and treatment was observed in the findings as the participants expressed that they had to advocate for themselves, and this would take a toll on them overtime. The MDM highlights the importance of accounting for these issues. The second aspect which the findings reflected was the 'shared-decision making', the need for this was seen in response to question regarding respect from HCPs, as the results showed that there is a need for mutual respect in order for shared-decision making. This aspect could also be seen in the results which emphasise the need for MDT management as treatment plans should be agreed upon by HCPs from varying disciplines. Finally, the need for 'community navigators' who can support individuals in adult services. This was observed not only in the need for HCPs to support individuals but also in the need for peer support which was provided through the community groups established through the BBS and social media platforms.

The results of this phase also highlighted limitations of the MDM. This limitation is the absence of self-management from the model. Self-management was a recurring issue raised both during the qualitative phase and the quantitative phase of the study. Grady & Gough (2014) emphasised that self-management is vital in the management of chronic conditions and that properly preparing people with chronic conditions to self-manage would aid to mitigate complications and comorbidities.

As self-management is such an integral part of this research, the suitability of the MDM for this research is brought into question. However, there is not a suitable alternative to this model and this suggests that a new model is needed which can more appropriately discuss the issues relating to the ongoing management of adults with OI.

4.4.3 Implications

The online semi-structured survey showed that the majority of the sample did not have support when leaving paediatric services. The impact of this on the individual would not be limited to the time of their transition but would extend into their experience in adult services. Without guidance in adult services, they would not be equipped with the skills for self-management and would struggle to advocate for themselves. Further to this, they would be unsure of aspects of their future care, such as who would be their consultant and who they should contact in the event of complications.

This is vastly different from the care they would have received in paediatric services; in those services they would know their consultant and their first point of contact. This change in services could be one of the reasons behind the majority of the sample stating that the care they received in paediatric services was better. It is possible that the participants rate their care in paediatric services as better than adult services because they did not have the uncertainty seen in adult services. It could also be due to them having more support from family members who would have attended the appointments with them. This could also be the result of hindsight, their care may not have actually been better, but they may look back at that time in a positive way for other reasons, which gives them the false impression that their care was better on the whole.

The idea of hindsight influencing the rating of paediatric services in comparison to adult services can be explored using the results of the question regarding the overall rating of adult services. Of the 36 participants which responded to this question, 15 rated adult services as poor. There are a number of potential reasons for this. Firstly, participants likely decided on this rating by comparing it to their experience in paediatric services. This does suggest that their care was better as a child, and this would support the findings of the question comparing the two services. Secondly, there is a lack of specialists for adults with OI and the

services available to them are limited. This could easily result in a poorer experience as they might be unable to access the treatments they need.

There was also consideration made to the influence of OI type on the care received by the participants. The survey showed that the type of OI the participants had did not affect the level of support they received during their discharge from paediatric services. This is arguably a positive for the OI community as it suggests that the best care is not reserved for certain types of OI and instead the differences in care such as level of support and available treatments are due to external factors such as availability of specialists.

Another issue identified in the survey was that of continuity in adult services. The majority of participants stated that they did not experience continuity in their care. This could have a number of negative consequences on the individuals. Firstly, not having consistent recommendations or treatments offered to them by HCPs. Frequent changes in their medications or other medical interventions could cause their condition to worsen and affect their quality of life (Viner, 2008). Secondly, not having continuity in the HCPs they are seen by would increase the burden on the individual as they would have to go through the same conversations repeatedly. They would need to explain their condition and their medical history to each HCP, this would take up a great deal of time in the appointments. In addition, the burden this puts on the individual could affect their capacity for other activities such as education, work or social activities (Siboni et al., 2019). This could have a detrimental impact on their mental health.

The results of the survey also showed that participants rated the knowledge of their consultants in paediatric services as higher than the knowledge of their consultants in adult services. The possible reasons for this finding are closely linked to the previous issues discussed. Firstly, more participants stated that they were under the care of a specialist as a child and as such those HCPs would have more knowledge of OI. In addition to this, these HCPs are more likely to be working as a part of an MDT, as clinics for OI adopt an MDT approach more frequently. This would mean those HCPs have access to more knowledge from differing specialties and fields (Bregou et al., 2016). Finally, as children they would have had less knowledge of their condition and by comparison to them an HCP would have seemed more knowledgeable, and this also could be contributed to by the effects of

hindsight and subsequently their view of their paediatric consultant may have been skewed by other external factors and may not be an accurate representation of the consultants' level of knowledge.

In comparison to this, when asked about the level of knowledge held by their adult consultant, fewer people selected excellent which suggests the participants did not find them as knowledgeable. There are a number of potential explanations for this finding. Firstly, when looking at this finding, it must be compared to the question which asks people whether they are seen by a specialist in adult services. Fewer than a quarter of the participants stated that they are seen by a specialist as an adult, this could be due to the lack of specialist services. As many participants reported that they are not seen by a specialist, it would be reasonable to assume that they do have less knowledge of OI as it is a rare condition. Further to this, the availability of information regarding OI must be considered, as information is difficult to access, HCPs may struggle to research the condition prior to appointments with people who have OI. Finally, as people with OI age, they acquire more information about their condition, they are able to learn more about their condition by experiencing it and HCPs cannot have this same level of understanding (Cordier, 2014). This might make HCPs seem less knowledgeable when compared to the individuals' knowledge.

The survey showed that a large proportion of participants have had a poor experience in adult services. 15 out of 36 participants rated their experience as poor and of these 15, 14 stated that they do not have a consultant who is an OI specialist. This, along with the other findings of the survey support the need for specialists for adults with OI as a means of improving their experience in adult services. Having access to a specialist as an adult would allow individuals to be better informed which will allow them to make decisions which are best for them. They would be able to be treated according to the best practices for people with OI, which could lead to fewer complications and early identification of comorbidities. Individuals could also have better support as their HCP would be more knowledgeable about issues facing people with OI and the impact of that to their general health and wellbeing.

Another finding of the survey was that the vast majority of participants had at some point been dismissed or doubted by the HCP. Of the 39 participants which responded to the question, 36 had been dismissed and of that 36 participants, 29 stated that they had

fractures missed by HCPs. It is possible that being dismissed or doubted can lead to fractures being missed and this could lead to worsening health status and prolonged healing. As without a diagnosis of a fracture, individuals would not be able to access services which might aid in their recovery, such as physiotherapy. Without these services, they might struggle to regain mobility and might heal incorrectly which could lead to an increase in pain (Lafage-Proust & Courtois, 2019b). Combined this could impact quality of life and the emotional toll this would take on the individual must be considered. Not only the emotional burden of a slow and painful recovery but also the impact of being doubted or dismissed by HCPs on their mental health. This also supports the need for transition services and adult services for people with OI which can prepare them for self-management and self-advocacy.

The finding of missed fractures was suspected to be due in part to the lack of knowledge of OI held by HCPs who are not specialists. When these two questions were compared, there was no clear link between the incidence of missed fractures and the level of consultant knowledge. However, the question regarding consultant knowledge was aimed at gauging the knowledge of the individual's primary consultant and not that of other HCPs that they might encounter, including any HCPs in accident and emergency departments which are more likely to be responsible for diagnosing fractures. The individual's primary consultant may only learn about new fractures at scheduled appointments.

When considering the level of knowledge held by individuals, the impact of the BBS on knowledge was explored. Of the 39 participants which responded to the question, 35 stated that they are members of the BBS. Of this 35, 30 rated their knowledge of OI as either good or excellent. Although this might suggest that having access to the information from the BBS has a positive impact, there are other considerations to be made. Firstly, of the four participants which stated they are not members of the BBS, three rated their knowledge as excellent. Secondly, the information which is distributed by the BBS is freely available on their website and a membership is not required to access it. Further to this, the impact of the BBS on whether individuals would share their opinions on treatments with HCPs was explored. The data showed that of the 35 participants who are members of the BBS, only five stated that they would always share their opinions with their HCP. This suggests that knowledge alone is not enough. If this result is compared to the question regarding knowledge where ten participants rated their knowledge as 'Excellent', there are a number of participants for

whom this high level of knowledge was not enough to enable them to share their knowledge and opinions with their HCP. This suggests that in order to enable people with OI to become empowered expert patients, they need to be educated on their condition and also need to be given the confidence to discuss their condition, share their opinions and to challenge their HCPs.

The access to appointments and referrals to specialist services was also explored in the survey and the findings showed that in regard to appointments, the participants were split fairly evenly. 17 participants stated that they did have regular appointments with a consultant in adult services, and the remaining 19 stated that they did not receive this. This finding could in part explain the findings of the question regarding overall experience in adult services. Given that such a high number of participants are not seen regularly by a consultant, they might struggle more to access services when they need them as they would have to go through the referral process which can be quite lengthy. When asked about referrals, participants for both the GP referral question and for the question regarding referrals in general, the majority of participants stated that they are able to get referrals often or sometimes. This supports previous discussion that there is inconsistency in the treatment of OI and that this does impact the individuals. If individuals are unable to obtain referrals to specialist services, their condition could worsen, and this can impact not only their physical health but also their mental health (Dahan-Oliel et al., 2016). This again emphasises the importance of transition and adult services to ensure individuals with OI continue to have access to services which they need.

4.4.4 Strengths and Limitations

This phase of the research aimed to further explore the issues raised in the qualitative phase and was designed to ascertain whether those issues raised were shared by the wider OI community. A limitation of this phase of the study was the small sample size that was obtained. Due to the small number of participants, the results lacked the statistical power required to answer some of those questions definitively. This small sample size was obtained despite numerous attempts to recruit participants. The recruitment post was advertised in the aforementioned Facebook group for adults in the UK with OI, in addition to being shared by the BBS on their website, on their social media accounts and in their monthly newsletter

to members. As this condition is rare and affects fewer than 5000 people in the UK, there is a small community from which participants could have been recruited. In addition, when taking into consideration the inclusion criteria for participants, that left an estimated 3300 potential participants across the UK.

Although the sample was small, it did include participants from all home nations within the UK and the OI types were varied which allowed for a variety of experiences to be included and therefore represented in the results. The benefit of this is that the results can take into consideration, the variety within the OI community and the diverse experiences which are the consequence of varied severities of the condition. In addition to the small size of the sample, this sample is not representative of the general OI population as type I accounts for 50% of cases in the general population and accounted for only 28.9% of the sample population (OIF, 2020).

Another benefit of the method selected for this phase is that as it was online, participants were not inhibited by geography and were able to take part from anywhere in the UK and at a time that was suitable for them. Unlike the qualitative phase where the data collection had to be arranged for times which best suited the participants, this phase of the study could be conducted at their own pace and at a time which was convenient for them and their schedule.

Another limitation of this phase of the study, was that the analyses could not show that the results were statistically significant for the majority of the crosstabulations. This does not mean that there are not correlations but rather that this sample did not have sufficient power to show these relationships. This was another limitation that resulted from the small sample size, and this would support the need for a further research study.

4.5 Conclusion

This chapter has discussed the findings of the quantitative phase of the research. These findings have shown that in addition to transition being an issue for people with OI, managing the condition in adult services and self-management is also a significant issue. The findings of this phase of the research, combined with the findings of the literature review and the

qualitative phase will be used to develop a patient guide for adults with OI. The following chapter will discuss the development of the guide and the uses of the guide.

Chapter 5 – Resource development phase

5.1 Introduction

The previous chapters have discussed the qualitative and quantitative research which has been undertaken for this study. The data collected in these phases revealed a number of issues faced by people with OI both during and after their transition to adult services. One of the key findings identified was a lack of knowledge of OI and the difficulties faced by people when trying to learn about the condition and the subsequent issues faced when attempting to manage their condition and educate their HCPs. This chapter will focus on the development of the patient guide which aims to address the issues identified in the previous chapters. This chapter will discuss the how the guide was developed and the potential implications of this guide on the OI population.

Although the primary aims of this research were to explore the issues around transition and the ongoing management of adults with osteogenesis imperfecta, there was intent to create a resource for adults with OI from the beginning of the project. The resource would support adults with OI and allow them to be more active in the management of their condition by equipping them with the information needed to make informed decisions and to advocate for themselves.

The original aims and objectives of the research as laid out in the introductory chapter were clear and the development of the guide either served to fulfil these objectives in a number of different ways or was a consequence of the fulfilment of objectives through another phase of the research. The focus group, interviews and online semi-structured survey achieved the third objective, “To develop an understanding of the ongoing care needs of adults with osteogenesis imperfecta in the UK from their perspective” and did allow for an understanding of the needs of people with OI. This understanding of OI and the needs of individuals could then be taken forward and was an integral part of the development of the patient guide. The focus group, interviews and online semi-structured survey also served to achieve the fourth objective, “To identify any gaps in knowledge of people with osteogenesis imperfecta” and did explore the issues around gaps in knowledge people with OI have. This information was also taken forward and a significant contributor to the patient guide development.

The final objective of this research was to develop a guide for people with OI, "To develop a guide for adults with osteogenesis imperfecta to assist them in navigating the healthcare system in order to facilitate their access to the necessary services". This guide addresses the gaps in knowledge and provides people with OI with the tools needed to navigate the health system and be proactive in their care. Self-management is a vital part of chronic conditions management and is key for easing the burden on the limited resources available within the NHS. Allowing people with chronic conditions to be cocreators in their treatment plans benefits both health providers and the individuals by enabling collaboration and compromise to ensure treatment plans have the intended health benefits as well as meeting the needs and wants of the individuals.

In addition to considering the original aim and objectives of the research, the data gathered was vital for the development of the guide. The discussions from the qualitative phase yielded a vast amount of useful data and this included comments which supported the need for a resource for people with OI. Participants stated that both HCPs and people with OI struggle with misconceptions "*that is why I think we need more education in OI"* (F, 32yo, T1/4). The participants highlighted that *" there was lots of gaps"* (M, 26yo, T4) in the information available and that there *"much so much information and misinformation on the Internet it's really hard to know what to believe"* (M, 26yo, T4). This would make it difficult for people with OI to find information and would make educating themselves and being proactive with their health more difficult. They also discussed the existing information available from the BBS but stressed that "*it still feels that they are geared up to paediatrics"* (F, 33yo, T3) and there was a limited amount of information which was appropriate for them. One participant said that *"the information, side of things does need to be a lot broader"* (F, 32yo, T1/4). This is important for OI as it has a wide range of potential complications and is not limited to fractures.

It was also mentioned that part of what makes accessing information difficult is the uncertainty with where to go for information. As previously mentioned, the NHS choices website does not have a page for OI (Correct as of February 2023), one participant expressed that *"Just knowing where to go if I had a query or a worry really"* (F, 56yo, T4) would help them. The participants all agreed that resources were needed for people with OI, *"something like an information pack that's available for you to click on at any time"* (F, 33yo, T3) was

suggested and this aligns well with the aforementioned objectives of the research and further supports the need for a patient guide for people with OI to be produced.

The focus group and interviews, in addition to providing valuable data to address the aim and objectives of the research, also led to the development of a number of research questions. These questions were to be taken forward into the development of the online semi-structured survey. When exploring these questions using the data gathered in the focus group, interviews and online semi-structured survey responses, it was clear that there are limitations in current services and that a patient guide for people with OI would be beneficial.

Firstly, the sixth research question which poses the question; Are people who are members of the BBS more likely to have more knowledge and/ or to advocate for themselves? This question explored the relationship between being a member of the BBS and the level of knowledge people with OI had and their likelihood to advocate for themselves. It was found that even people who are members of the BBS rated their knowledge as poor. It was important to explore why this could be, perhaps a lack of adult centred information was the cause or that people with OI do not fully understand the scope of OI and subsequently do not know what comorbidities they are at risk of developing. Although it is unclear what the cause of the result is, it was clear that there was a need for adult centred information. This would give people with OI information pertinent to them and would inform them of the risks OI poses to adults.

The third research question was in regard to adult services and asks, Is there a connection between poor care for adults and a lack of adult specialist services? The data revealed that a great many of the participants were unaware of the risks associated with OI and found adult care for OI to be poor and not comparable to the care they received as children. When taking these points together we can establish that as some adults with OI are unaware of the risks associated with OI and do not receive good adult care. It is possible that early signs of comorbidities will go unnoticed as they are not being monitored by HCPs. In addition to the lack of monitoring, without an understanding of what they are at risk for, people with OI will not be able to be vigilant and act proactively in the event of complications. The combined effect of these points is that there would be a higher risk of complications for adults with OI. A patient guide could also address this issue by providing adults with OI the information and

enabling them to be more vigilant and proactive and this would allow for better self-management.

The fourth research question also address issues which are relevant to the development of a patient guide; Is there a link between being dismissed by HCPs and the occurrence of missed fractures? It was important to explore any links between being dismissed and fractures being missed by HCPs. This exploration led to a further exploration of a connection between lack of knowledge, advocacy and being dismissed. The purpose of this question was to explore whether a patient's lack of knowledge would influence them in regard to advocating for themselves. It is known that the possession of knowledge empowers people with chronic conditions, and this could enable them to advocate for themselves. For OI, this could allow people to advocate for x-rays to be performed or give them the confidence to question the HCPs. This could allow for fewer fractures to be missed and for better care to be given to people with OI. This also supports the need for a patient guide.

The quantitative phase also contributed to the development of the guide. This was achieved as the online semi-structured survey further explored the topics discussed in the focus group, interviews, and online semi-structured survey. This made it possible to explore what areas of information people were most interested in receiving and also how they would want to receive the information. It was important to explore both of these aspects in order to give people with OI a say in the patient guide as it was being produced for their use and if it did not meet their needs, it would not be beneficial and would not achieve the objectives laid out in the beginning of the research.

Based on the data gathered and the discussions of the previous chapters, it was decided that there was sufficient justification for the development of the patient guide. The remainder of this chapter will discuss the methods undertaken to develop the guide, the results of the feedback collection and will discuss the implications of guide.

5.2 Method

This chapter has so far discussed the overall purpose of this research and the knowledge gap in the field of OI. The overarching aim of the research has been to provide an evidence-based resource for people with OI and this section will discuss the methods undertaken in the

development of the patient guide and the results of the feedback of the guide which was obtained from people with OI, research supervisors and the BBS.

5.2.1 Participants

Participants for feedback on the guide were recruited from a private Facebook group, Osteogenesis Imperfecta UK & Ireland (Adults). This group has 249 members (as of January 18th 2023). This group was selected as, unlike some other Facebook groups for people with OI, it is a UK based group, and this research has been focused on individuals in the UK with OI. The desired population for this data collection was adults with OI who are living in the UK. However, as personal information was not being collected in this phase of the study, it was not possible to check that these criteria were met. It was felt that by posting the guide and asking for feedback in this Facebook group, and not in the other international groups, that these criteria were likely to be met. Ethical approval was sought from the Faculty of Medicine and Life Sciences. Approval was granted by chairs action following amendments being made to the ethics application for the online semi-structured survey phase of the research. This was done as this round of data collection was seen as an extension of that phase of the research.

5.2.2 Materials

The guide was developed using the data that was collected in the focus group, interviews, and through the online semi-structured survey. The information included in the guide was informed by the findings from the literature review conducted prior to the commencement of the focus group and interviews. Information was then corroborated using sources such as the NHS website, NICE guidelines and other publications. This was done to ensure information included in the guide was reliable and to prevent misinformation. The guide was broken down into topics which were identified through the literature review. These were then prioritised in the guide according to the responses to the online semi-structured survey where participants were asked what topics they would like to have more information on. These topics were then researched individually, and the findings were summarised and explained. The guide does not assume prior knowledge of OI and gives the reader the basics as well as addressing the gaps in knowledge that were identified in the focus group and the

interviews, and in the survey. The content is aimed at people with OI rather than HCPs and therefore it does not contain much jargon and any jargon is defined in the guide.

5.2.3 Procedure

Feedback for the guide was collected online through the use of Facebook. This method of data collection has a number of benefits and drawbacks. The first benefit of this method is the quick response time. Members of the Facebook group were able to see the recruitment post immediately and would be able to provide feedback quickly. The second is the ability to attach the guide and the participant information sheet to the recruitment post, this would make it easier for participants to see the information without having to download the documents, this makes this method more accessible and more user friendly. A drawback of this method is the inability to verify the eligibility of participants. It would not have been possible to confirm the participants had OI, were over the age of 21 or lived within the UK although this data was anonymised and this was not relevant to this data collection, this is a limitation of the use of social media and other similar websites as it is not possible to verify identity.

The first version of the guide was posted in the private Facebook group on October 5th 2022 and again on November 3rd 2022. The second post of the guide stated that no responses would be collected after 5pm on the 10th of November 2022. The guide was shared in the group twice in an attempt to reach more of the members of the Facebook group and to give more people a chance to participate. The guide was only posted on this Facebook group as the group was private and therefore the comments would not be visible to who were not members of the group. This group was chosen as it is a group for people with OI in the UK and Ireland. The post which was placed in the Facebook group (Appendix 7) included information about the research and contact information to allow prospective participants to ask questions about the research. The post also included a link to a participant information page which participants were asked to read prior to participating. It was made clear in the post that participation through commenting was voluntary, and that no personal information would be taken. The post also stated that by commenting on the post and providing feedback on the guide, they were giving their permission to their comments being used for the purposes of the research. Screenshot images were taken of the comments, the names and

profile pictures were covered, and these can be found in Appendix 10. Individuals were assigned numbers; this was done to track how many contributors there were to the comments.

5.2.4 Analysis

The comments made on the Facebook posts were extracted and a qualitative analysis was conducted. This method of analysis was chosen as the aim was to obtain feedback on the guide in order to make amendments to the guide. A full thematic analysis did not need to be conducted as the aim was not to identify or explore relationships between themes or sub-themes. Comments were left by eight individuals with some individuals making multiple comments, some individuals were discussing OI related topics in the comment section and these have been included in Appendix 10. One individual left a comment on the post but did not provide feedback on the guide. That comment has been omitted from Appendix 10.

After the comments were collated and feedback was extracted, amendments were made to the guide and a second version was created. The changes can be seen in Appendix 11. This amended version was then sent to both the primary and secondary supervisor for additional feedback. The feedback provided was then used to make additional amendments to the guide, these changes can be seen in Appendix 12.

Once these amendments were made, the guide was sent to the Brittle Bone Society and their Medical Advisory Board and the Scientific Advisory Board to be reviewed. The members of their boards were able to ensure all of the information in the guide was accurate and they provided some points which needed to be clarified in the guide and some minor amendments were made, these can be seen in appendix 13. This feedback came from people who are experts in OI and work with people who have OI, they were able to provide feedback based on their experience and knowledge of what does and does not work for OI. This is beneficial for the guide, as was seen in the review of the literature, information on OI and best practice treatments are limited.

Following these three stages of review, it was felt that the guide did not need any further amendments, the final version of the guide can be seen in Appendix 14. The guide could then be made publicly available, and the final version was then disseminated in the Facebook

groups where participants were recruited from and was also shared by the Brittle Bone Society on their website and through their social media accounts.

5.3 Results

The feedback on the guide was received in three waves. The first from members of the OI group on Facebook, the second from the supervisory team and the third from the BBS medical advisory board (MAB) and BBS scientific advisory board (SAB). The results of each wave of feedback collection will be discussed separately in this section of the chapter and the changes made to the guide following the feedback will also be included in this section.

5.3.1 OI Facebook Group Feedback

There were comments left by eight people on the Facebook group, one person did not provide feedback in their comment and so this will not be included in the results. Of the seven people to leave feedback on the guide, there were a number of suggestions for additional information which could be included, some questions regarding information included and a number of general comments which reflected a positive impression of the guide.

The fractures page yielded a few comments. The first comment stated that "*green fractures are more common in kids"*, this comment was referring to greenstick fractures, the information regarding these fractures in the version of the guide uploaded to the Facebook group did not specify that these fractures are more common in children. Following this comment and further research was reviewed and it was determined that this comment was accurate, and the guide was updated accordingly. Another comment on this page of the guide questioned the recommendations on where to go for medical treatment in the event of fractures. This information was based on the information provided on the NHS website, but this information was limited and following this comment, the types of fractures which might require the use of 999 services was extended to include open fractures, pelvic or femur fractures or unstable tibia fractures.

The page regarding surgery for OI also received some feedback. One comment in reference to telescopic rods stated that "*although they grow with the child they may need changing if*

the child outgrows the rod". Based on this feedback, additional information regarding the limitations of telescopic rods was added to the guide which emphasised that these rods have a maximum length and can fail for other reasons which may results in them needing to be exchanged.

Two people left comments regarding the bisphosphonates page in the guide. One person suggested that the description of the risks associated with bisphosphonates were not detailed enough. Another stated that "*not all individuals have a DEXA scan before starting treatment"*. Based on these comments a fuller description of the risks associated with bisphosphonates was given and clarification on the use of DEXA scans prior to bisphosphonates being prescribed was added to the guide.

The page on physical therapies also received some feedback. One comment highlighted hat *"building muscle mass also helps to reduce fractures"*. This statement was added to the summary of the use of physical therapies for OI. This person also added that "*specialist OI Centres have PTs/Ots on their OI team"* they go on to say that these people would be the *"first port of call".* This feedback was not included in the guide as the guide is aimed at adults and OI teams primarily treat children with OI. Another person raised a query about the hydrotherapy section by saying that "*hydrotherapy [is] more than just swimming more like physio in the water".* The guide did not state that hydrotherapy was just swimming and following on from this comment, the description of hydrotherapy, including the uses of it and how it is conducted was expanded.

The page on dental problems associated with OI received two comments. Both of these comments questioned the use of dental implants in OI as their use is dependent on bone density. As OI severity can vary and implants might be possible for those with higher bone density, the following statement was added to reflect the implications of the use of dental implants, 'this will depend on whether the bone is strong enough to support the implant and may not be possible for some people with OI'. One person left feedback for the page on hearing loss. They first state that they loved the "*mention of bone anchored hearing aids*" and went on to suggest the addition of information on cochlear implants and mixed hearing loss. They then made a further comment asked for the guide to include *"stapedectomy surgery*

which has been successful in some OI patients". Based on these comments a number of additions were made to this page and all of the above recommendations were acted upon.

There were two comments for the page regarding heart and lung issues. One person exclaimed that they did not previously know about the issues affecting the heart in people with OI. They said they were *"especially interest in the heart section, [as this was] not an area [they] know anything about"*. The second person commented on the section regarding atrial fibrillation and asked for more information about the risks of this for people with OI. They stated that atrial fibrillation can lead to an *"increase of blood clots that can cause strokes/ heart attacks"* they go on to mention that this is usually treated with an anticoagulant. This was explored in the literature and additional information regarding the risks of and treatment of atrial fibrillation was added to the guide.

There were a number of comments made regarding the spine page of the guide. This included two of the participants conversing about basilar invagination (BI) which is one of the complications associated with OI that affects the spine. In the comment which began this discussion of BI, the person stated that their son had BI and that they would "*love to see a list of symptoms and advice to be assessed by a neurologist"*. Another person replied to this advising that they have BI and as a result "*have a lot of knowledge on this [BI]".* Both of these participants then went on to discuss treatments and symptoms of BI. Based on this discussion, the BI section of the spine page was expanded with symptoms and advice on where to go in the event of these symptoms added to the guide.

One comment praised the page on pregnancy in OI, stating it was "*very interesting, as [it is] hard to find information about it"*. Another comment made reference to this page and suggested that the guide should include that people "*with OI who are planning to have children a referral can be made by GP/ OI team for genetic counselling"*. They go on to mention that this process led to their care being overseen by a consultant obstetrician and them being given extra scans and a birth plan. These additional details were added to the page on pregnancy. There was also a comment raised concerning the lack of a description of the BBS. They highlighted that the guide mentioned "*the BBS but [didn't] say who they are".* A description of the BBS was added to the guide in response to this comment. A comment was also left which queried the lack of information on "*the impact of mental health and the help*

available". This could not be resolved following this wave of feedback and presents an opportunity for future research.

There were a number of comments left which gave an overall positive review of the guide, with people stating they felt the guide was "*very comprehensive"* and that it was "*an interesting read and very informative"*. Another person commented on the guide reiterating that the guide was *"really interesting"* and that following on from reading it, they had "*lots of questions to add to [their] usual list"* for their consultant at their next appointment.

In addition to the positive feedback on the content of the guide, several comments also praised the aesthetics of the guide. The first comment left on the guide stated that at "*first glance love the colour and layout"* another comment said that the "*colours and layout are lovely and makes it easy to read*". There were two comments left which raised concerns over the accessibility of one of the pages due to the colour choices for the text and the text box. The first stated that "*black on dark purple and other colours are hard to read"*, they added that "*[they] are dyslexic so might be why [they] struggled"*. Another concerning the further reading pages, it was stated that these were difficult to read. In order to ensure the information in the guide was fully accessible, the colours for this page were revised and the website addresses for further reading were split into two pages and were divided into the pages they corresponded to.

A general comment that was left on the guide was concerning some typos within the guide. The guide was reviewed following this wave of feedback collection and typos such as spelling mistakes or missing punctation were remedied. Some other formatting concerns that were raised concerned the glossary; one comment recommended that the glossary should be alphabetical. This was resolved after this wave of feedback and the glossary was reformatted, it was alphabetised, the words were put in a bold font to make finding the desired definition easier and the colours of the page and text were revised to ensure the page was accessible.

5.3.2 Supervisory Team Feedback

After the changes were made to the guide based on the feedback from the members of the OI Facebook group, the second version of the guide was sent to the supervisory team for further feedback.

There were some more formatting issues which needed to be addressed, such as typos and missing punctuation. It was also suggested to change the order of the pages to improve the flow of the guide. The genetics page was to be moved further towards the beginning of the guide and would be placed after a new page containing the information regarding different types of OI, the genes affected and the inheritance pattern of those types. It was also recommended that the website addresses be made into hyperlinks to allow easy access to the sources of information.

The only remaining change recommended by the supervisory team was that the word on the back cover of the guide be revised to show that the information included in the guide was not directly taken from other sources and was a culmination of research using data from this research, existing literature and guidelines from recognised bodies such as the NHS and NICE.

5.3.3 BBS Feedback

After the changes were made on the advice of the supervisory team, the third version of the guide was sent to the BBS for review by their MAB and SAB. Once they had reviewed the guide they sent through feedback on the guide and recommendations for changes.

The first comment was in reference to the spine page of the guide. It was stated that some of the people on the MAB and SAB would not recommend bracing for spinal issues in OI. Based on this comment, an addition was made to this section of the guide stating that 'Bracing or surgery may not always be possible in OI as the bones may not tolerate these treatments'. The feedback also highlighted a typo in one of the gene names and this was corrected in version four of the guide.

There were several comments regarding the bisphosphonate page of the guide, these comments mostly centred on the language used to describe the actions of bisphosphonates. The first comment regarding this stated that "*bone is not made more quickly; better to say that the reduction in bone breakdown reduces bone fragility"*. Based on this comment the explanation of the action of bisphosphonates was amended and in version four would be 'Bisphosphonates work by reducing how quickly bone is broken down, the reduction in bone breakdown increases bone density and reduces bone fragility'. The next comment regarding bisphosphonates was in relation to the side effects associated with the treatment. It was

stated that although acute side effects such as fever and muscle aches are common for 48 hours after the treatment, these do not usually recur. This was added to the section which discusses the risks of bisphosphonates. An additional comment was left regarding side effects of bisphosphonates and that was that "the potential risks increase with the duration of treatment" and therefore bisphosphonates are often given for a limited time "*often 5 years".* Based on these comments the risks section was further amended to include this information.

There was also concern raised in the comments from the BBS that the page regarding physical therapies did not give an accurate representation of the roles these services play in the care of OI. The first comment made stated that *"OTs are heavily involved in wheelchair provision but not really mobility aids"* the comment then added that *"physiotherapists have many more skills to offer, particularly in relation to posture, pain and pacing."* Based on these comments, the role of the physiotherapist was expanded significantly to give a more in depth description of their capabilities. Also, the role of occupational therapists was amended.

There were also two comments which discussed the use of telescopic rods. These comments both highlighted that their use is limited to children and emphasised that "telescopic rods don't last". Based on these comments' clarification was made in the guide as to their limitations and the guide now stated that telescopic rods are 'Good for children who are still growing but are not used in adults'.

There was a comment included in the feedback was stated that babies with OI are *"more likely to lie in the breech position, increasing the likelihood of delivery by Caesarean Section"*. This was added to the guide in the section which discusses the choice between natural and caesarean birth. Another comment which was made reported that "despite the known lung changes, there was no reported issue for those with OI during the Covid pandemic", this finding was not yet apparent in the literature and this finding was added to the heart and lung page of the guide.

Finally, there was a comment which suggested that "The four English centres are presented as four UK centres". In the guide these were described as the four highly specialised services in the UK and in order to avoid confusion, clarification was made in the guide to ensure the

services available were clear. Version four of the guide therefore contains the list of the four highly specialised services in the UK and the regional centres which are spread across the UK.

5.4 Discussion

The aim of this phase of the study was to review the patient guide which was produced from the findings of this research. This was done in order to achieve the final objective of the research which was laid out in the introductory chapter. This final objective was "To develop a guide for adults with osteogenesis imperfecta to assist them in navigating the healthcare system in order to facilitate their access to the necessary services". Thus far this chapter has discussed the methods undertaken for the development of the guide, the methods undertaken for the review of the guide and the results of that review. This section will discuss the guide in relation to the existing literature, models and theories, the implications of the guide and the strengths and limitations of this phase of the research.

5.4.1 Existing literature

The existing literature, as has previously been discussed, is very limited and there are currently no other patient guides which have been reviewed in the literature. The BBS has produced a number of documents aimed at providing information about OI to individuals and parents, but these have not been tested, reviewed, or discussed in the literature. This research presents the first resource in the UK for adults with OI and as such the literature which can be used to review the guide and to draw comparisons from with regards to the results of the feedback are limited to one tool which was produced in Canada by Jeong et al. (2019). In addition, feedback on the guide was collected at a single point and therefore it is not clear what effect the guide would have on individuals, their health outcomes, knowledge, or hospitalisations.

Jeong et al. (2019) discussed the development of the 'Good2Go My Health Passport'. As discussed in previous chapters, this study showed that the provision of a tool was beneficial for individuals with OI as there is a lack of knowledge of OI among HCPs and individuals with OI. However, it has also been discussed that this tool does not provide information to individuals and is instead a resource aimed at providing HCPs with a summary of an individual's medical history. Jeong et al. (2019) stated that some of the drivers behind the

development for this tool was that individuals felt isolated during their transition, lack guidance and also that HCPs needed additional resources to assist in the assessment of individuals readiness to transition. The final version of this tool has not yet been tested and therefore it is unclear whether it does enable continuity, provide guidance or satisfy the needs of people with OI, all of which were described as desired outcomes of the tool.

In the absence of other equivalent resources for OI, the results of this study were compared to a systematic review and meta-analysis of self-management interventions by Panagioti et al. (2014). This study showed that self-management interventions can be effective in reducing hospitalisations and an improvement in health outcomes. The study also found that self-management interventions can reduce costs to the health system. A number of the studies included in this systematic review and meta-analysis were hindered by a small sample size, in a similar manner to this research. The results of Panagioti et al. (2014) suggest that this patient guide could be beneficial to individuals with OI as it provides individuals with knowledge and sign-posting to services, which could lead to the same positive outcomes observed in Panagioti et al. (2014).

5.4.2 Theories and Models

For this phase of the research, the MDM model was used, this is because this phase of the research is focused primarily on the management of OI in adults and does not directly discuss the transition from paediatric to adult services. This is suitable for this phase as the aim of the patient guide was to provide adults with OI with information, this guide was not directly aimed at supporting people with OI during their transition and as a result, the MOU model was not going to be used to explore these results.

As has been discussed in previous chapters, the MDM model is split into two sections, with the first, 'Tools to Identify the Right Care', being made up of seven elements, these are processes or actions that should be undertaken to allow people to have the best care possible. The second section, 'Tools to Make the Right Care Happen', consists of a further six elements which are processes or actions which HCPs should undertake to ensure they can provide adequate care. This second section has not previously aligned with the findings of the research as the research prior to the guide development phase was centred on the

experiences of people with OI. However, as the goal of this phase of the research was to produce an outward facing resource for people with OI, some of these elements are now applicable to the research and align with the findings.

The first element of the 'Tools to Make the Right Care Happen' section is 'Resource registries', the purpose of this is to establish a list of resources to ensure access to best practice information. This is to ensure that HCPs are able to provide individuals with information specific to their condition. This aligns well with the guide as the information included in the guide is specific to OI and includes many of the common comorbidities associated with the condition. In addition, the guide includes many links to further information and allows both individuals and HCPs to read further into treatments, make informed decisions and ensure evidence-based practice.

The second element within this section of the model is 'Lean consumption', this consists of making healthcare more efficient and ensuring good access to services for people with chronic conditions. It is possible that this guide could lead to more efficient care for people with OI, as the guide is designed to ensure that people with OI receive appropriate treatments and identify comorbidities early. This would then enable people to be referred to the correct services. This would be possible through collaboration between individuals and HCPs. Individuals would have information regarding their condition and the comorbidities, enabling them to identify any symptoms early and HCPs would have access to information about OI to ensure they know what the associated risks are and thereby what services would be required. This would then allow for the 'Lean consumption' element of the MDM to be achieved but this would be dependent on cooperation between individuals and HCPs.

The next element of the 'Tools to Make the Right Care Happen' section of the MDM is 'Medication therapy management', this involves ensuring that treatments are suited to the specific needs of the individual. Although the general principle of this element does align with the guide as it is routed in the ideology that care for individuals needs to be individualized, the guide could not offer a comprehensive discussion of all treatments and variants of medications or therapies that can be offered. Therefore, instead of giving HCPs all of the tools required to design treatment plans which would algin with this element of the model,

the guide instead offers HCPs with information to begin the process of providing individualised care design in cooperation with individuals.

Another element of this section of the model is 'Community navigators' which involves the utilisation of other individuals with the same chronic condition to share experiences and information with each other. Although the guide does not directly facilitate this peer-to-peer support, it does signpost individuals to the BBS which can in turn allow people to connect and create support networks. Another element of the model links closely to this as it encourages multidisciplinary team working, this element is 'Relational coordination'. The guide also can not directly facilitate this practice, but the information included highlights the systemic nature of the condition which reflects the need for multidisciplinary care.

The next element of this section of the model is the 'Wisdom leadership' which reflects the need for care to be expert-led and utilise the knowledge of specialists within the field. The importance of this for OI has been discussed throughout this research and as such there is signposting to specialised services for people with OI in the guide. The information for these services was obtained from the BBS who work closely with specialists. This allows for people with OI to seek referrals to HCPs who are experts in the field of OI. However, despite this it may not always be possible for adults with OI to be seen by a specialist as there are no specialist centres for adults with OI in the UK. As the guide cannot directly allow for wisdom leadership to occur, by signposting individuals to HCPs who have some expertise in treating OI, this gives them the opportunity to seek care from someone who is well versed in managing OI.

The final element of the 'Tools to Make the Right Care Happen' section of the MDM is the 'Choosing wisely campaign'. This element encourages HCPs to educate individuals on where to go for different types of care. This would ensure that they go to the correct facility to receive care whether that be their GP, a pharmacy or an accident and emergency department. The guide does offer some direction to individuals on where they should go for different issues. Despite the fact the guide cannot provide individuals with information on where to go for all potential issues, providing them with information on their condition and the potential comorbidities, it enables them to make the best decision for themselves.

Based on this section of the MDM, the patient guide that was developed and the feedback that was received, this model fits well with the findings and suggests that the guide does align with the principles behind the MDM including being theory-based and patient-centred. This would also mean that the guide does have the potential to benefit individuals with OI and enable them to manage their condition effectively.

5.4.3 Implications

The feedback from the patient guide was on the whole quite positive and supported the need for this guide. There are wide ranging implications for this guide and the effects of the guide are not limited to the OI community. This section will explore the implications of this guide and the potential impact of the guide.

The primary aim of this guide was to provide adults with OI with a source of information to allow them to manage their own condition. This would fill the gap in the service provision which was discussed throughout this research. The participants in the qualitative phase of the research raised concerns at the lack of support for adults with OI, they went on to suggest that a resource would assist them and provide some of the support that they felt they needed in order to manage their condition.

For people with chronic conditions such as OI, ensuring they are supported will have a range of benefits and will not be limited to the care and management of their OI. It has been documented that people with chronic conditions are more likely to experience mental health problems (Hopman et al., 2009) . The addition of mental health issues on top of a chronic condition such as OI would increase the burden placed on the individual and this would have a drastic effect on their quality of life and their capacity to participate in social or work activities. Although this guide is not aimed at helping people with mental health problems, it is hoped that the provision of information and signposting to services will ease some of the burden placed on people with OI. This will then reduce the overall burden placed on the individual which may increase their capacity to participate in social or work activities. This may then improve their overall quality of life.

In addition to this, support is also provided by the guide in the form of networking to the OI community. The guide not only includes signposting to NHS services but also directs people

to the BBS which is the UK charity for people with OI. This would enable people to interact with the OI community and receive peer support from others with OI. This was included in the guide as the importance of the BBS to people with OI in the UK was emphasised in both the qualitative and quantitative phases of the research. In the quantitative phase, the majority of the participants were members of the BBS, and this suggests that it plays a significant role in the OI community. In addition to the BBS, the online OI community exists on social media websites, and this was also referenced as a source of support and information for people with OI. It is felt that these platforms will allow people with OI to share the patient guide with each other and this will allow for more people to be educated on their condition and will allow them to manage their condition more effectively.

Another benefit of being able to manage their own condition will be their ability to advocate for themselves. The need for advocacy was a recurring theme throughout this research and this would be enabled by ensuring that they are fully informed and are able to make their own decisions about their treatment. This would then mean the patient guide would enable people with OI to advocate for themselves, this would then allow them to become empowered expert patients. The need for people with chronic conditions to be empowered expert patients was seen in the CCM which was reviewed as part of this research. Although this model was not utilised as it was not fully compatible with this research, elements of this model can be seen in other models and are vital for ensuring people with chronic conditions are supported and are enabled to take control over their treatment. Empowerment for individuals with chronic conditions is essential for advocacy and self-management. (McCorkle et al., 2011) states that in order for empowerment to occur, education of individuals and communication using layman's terms is essential. This guide uses this type of language to communicate with the readers, this is to allow individuals to be educated and to enable them to become empowered and to advocate for themselves.

As has been discussed, this patient guide will have a positive impact with regards to providing support to people with OI, will enhance their ability to advocate for themselves and will give people access to a community of others with OI. All of these benefits are dependent on people with OI being educated in their condition, potential comorbidities, and treatments. The main objective of the guide was to educate people with OI and this then allows for all of the aforementioned implications to occur and is the gateway for enabling self-management,

empowerment and self-actualisation as individuals will be able to make their own informed decisions and will be granted full autonomy over their health.

As previously mentioned, access to information regarding OI does not only depend on ensuring adequate resources are available, but it is also dependent on the information and resources being accessible. One element of this was discussed in the results section, ensuring the document was accessible for people with dyslexia and other similar conditions through formatting was important. In addition to this, the language used had to be carefully considered in order to ensure the information was clear and could be understood. Layman's terms were used where possible and if this was not possible, a glossary was included in the back of the document which offered definitions of medical jargon. Ensuring the document is easy to read was important as both health literacy and general literacy of the population could impede access to the information. Poor health literacy has been linked to increased hospital visits which may waste valuable time and resources within the NHS (Simpson et al., 2020). Providing easy to read resources can aid in the improvement of health literacy which in turn can enhance an individual's ability to manage their condition. This was essential for this research and was required in order to achieve the fifth objective of this research, to develop a guide for adults with osteogenesis imperfecta to assist them in navigating the healthcare system in order to facilitate their access to the necessary services.

Further to this, this patient guide will not only provide education for individuals with OI but could also serve as a resource for HCPs. It was made clear through the literature review and the review of current guidelines that there is a paucity of information available to HCPs to guide best practice for OI. As this guide was made with feedback from the MAB and SAB of the BBS, the information included in the guide reflects the best practice for individuals with OI. This guide can therefore fill gaps in the information available to HCPs and could improve the standard of care adults with OI receive from the HCPs. It is hoped that this will result in fewer complications and fewer incidents of missed fractures which according to the data from both the qualitative and quantitative phases of the research are frequent occurrence for adults with OI.

An additional benefit of using this guide to educate HCPs is that as they will be better informed of the wide-ranging impact that OI has on an individual. It is hoped that this will

then mean they will be less hesitant to give people the treatments and referrals to specialists that they need. This can then improve health outcomes by preventing complications or by treating them early. Prevention or early intervention for complications will not only improve the quality of life of individuals but will also reduce the number of or length of hospitalisations which will reduce the burden of the NHS (Eaton et al., 2015). Although it can be argued that additional appointments or testing that may occur as a result this will be more costly for the NHS in the short-term, the opportunity cost of this is that in the long-term, there will be fewer severe complications and it is known that early intervention results in less treatment and fewer hospital stays (Soares et al., 2020). Therefore, there is potential to save the NHS more money in the long-term and this method of monitoring and treatment for people with OI will be beneficial for the NHS.

In addition to improving physical health outcomes, receiving better care from HCPs and experiencing fewer complications and comorbidities can have a positive impact on the mental health of individuals. As was previously discussed, people with chronic conditions are more likely to experience mental health issues and this is contributed to by the development of comorbidities and overall deterioration of health. Therefore, educating HCPs can have a wide-reaching benefit. Prevention of mental health issues could be further aided with inclusion of information on mental health for people with OI in the guide, this presents an opportunity for future development of the guide and the potential for the impact of the guide to be improved.

5.4.4 Strengths and Limitations

This phase of the research was aimed at developing and reviewing a patient guide for adults with OI. A limitation of this phase of the research was the small sample size in the first wave of feedback. Comments with feedback for the guide were only left by seven people when it was posted in the OI Facebook group. There are a number of potential reasons for the small response rate from this group. Firstly, the group only has 259 members (as of June 2023). In addition, it is possible that not all of these members are actively engaging in the group. However, despite the small number of respondents, there was a lot of feedback received from those people as many of them gave extensive feedback and some left multiple comments on the guide. Some people even began conversations with other group members

in the comment section where they discussed their experiences with some of the complications listed in the guide.

A strength of this phase of the study was that it allowed co-production to occur. Using feedback from both the OI community in the form of feedback comments on the Facebook post and HCPs in the form of notes provided from the SAB and MAB of the BBS enabled an element of coproduction. The benefit of this to the guide is that it will better serve the group it is aimed at. Despite best efforts to coproduce the guide, the guide was not fully coproduced as feedback could only be gathered following the first edition of the guide being made. This was due to the time constraints placed on the research. Although it could be argued that these efforts to coproduce the guide are limited, these methods were the only ones which could be utilised as these methods allowed the release of the guide to be limited whilst changes were ongoing. Following the collection of feedback and the changes being made, the guide was made widely available (Morgan et al., 2023). As the guide was not made freely available prior to the this, it was possible to protect the guide during its development.

5.5 Conclusion

This chapter has discussed the final phase of the research, which was aimed at developing and reviewing the patient guide for adults with OI. The purpose of this guide was to provide adults with OI a resource which would allow them to be better informed of their condition and treatment options. This guide would also signpost them to necessary services. The feedback on the guide from people with OI, the supervisory team and the BBS provided both positive comments and some constructive criticism with suggestions on how to further develop the guide. The comments received supported the need for a patient guide for adults with OI. The following chapter will discuss the research as a whole and bring together all phases of the study for a final discussion.

Chapter 6 – General discussion

The previous chapters of this thesis have explored issues relating to the transition from paediatric to adult services and the management of adults with OI. This included multiple data collection points in the form of a focus group, interviews, an online semi-structured survey, and feedback on the patient guide. This chapter will discuss the findings of this research as a whole, will compare the findings of this research to current literature and will explore the implications of this research on future research and practice.

6.1 Literature review

The first stage of this research consisted of a review of the existing literature. A review was conducted which focused on literature relevant to the transition from paediatric to adult services and the ongoing management of adults with OI. This was done in order to establish any gaps in the current knowledge of OI, to explore the findings of previous research and to identify what information was readily available to both HCPs and individuals with OI.

This review of the literature found that there was a lack of research into the transition from paediatric services to adult services for people with OI. In addition, the majority of the existing literature was centred on the impact of OI on children rather than adults. As a result, there was a limited amount of information available discussing the issues facing adults with OI. The literature was broken down into themes and the findings of these themes were explored. The first theme was transitioning to adult services. The literature in this topic came from the US and Canada and there was consensus in these studies regarding the needs of adults with OI. The first of these studies was by Carrier et al. (2018) which reported that both children and adults with OI wanted to be educated and be knowledgeable of OI in order to engage in self-management. Shapiro & Germain-Lee (2012) supported this and emphasised the need for equipping individuals with the skills needed to overcome barriers and to be independent. This was also supported by Dogba et al. (2014) who stated that without these skills, individuals with OI would be unable to engaged in self-management. Further to this, both Dogba et al. (2014) and Shapiro & Germain-Lee (2012) stated that without continuity, reassurance and communication from HCPs during their transition, the process would contribute to anxiety and would lead to a poorer transition experience. All of these findings

align with the findings of this research as participants reiterated their desire for more education to allow them to manage their condition. The need for continuity, communication and reassurance or support were also recurring themes throughout the study which also shows more alignment between this research and the existing literature.

The second theme in the literature review findings was management of OI, this theme was broken down into five sub-themes. The first sub-theme discussed the general requirements for managing OI. Gil et al. (2017) discussed the need for individualised care for OI and this was supported by Tournis et al. (2018). The results of this research show that individuals with OI also feel that individualised care is important and is beneficial. This sub-theme also discussed the clinical needs for adults with OI, Marini et al. (2017) advocate the need for screening for adults with OI in order to identify complications early. This also aligns with the findings of this research as it was stated by many participants that they wanted regular check-ups and tests in order to keep track of their condition and provide them with reassurance.

The second sub-theme with the management theme was multidisciplinary team management. This has been discussed at length throughout this research and the importance of this for adults with OI was argued by Bregou et al. (2016) who stated that the use of MDTs led to improved health outcomes. This is due to the ability of the disciplines to work together and provide a holistic approach. Marr et al. (2017) stated that the use of MDTs was beneficial as it allowed for consideration of the wider determinants of health. These benefits of using MDTs were seen in the results of this research and participants stated that this method of management would ensure that their care met their needs and would prevent them from receiving conflicting treatment advice.

The third sub-theme for the management of OI, was the audiological management of the condition. The research into this area of OI revealed that hearing loss is common for people with OI, Swinnen et al. (2012) described the incidence of hearing loss and reiterated the implications of this on people with OI. However, Hald et al. (2018) stated that hearing loss is undertreated in OI and this was the result of a lack of screening for hearing loss. This further supports the need for frequent appointments, that are holistic in nature, providing thorough care. This need for screening of known complications and comorbidities associated with OI was discussed by Marini et al. (2017) which reflects the importance of monitoring of adults

with OI. Such monitoring or screening will prevent such issues from being missed and ensure that the health status of people with OI is preserved. This will enable their quality of life to be maximised. The absence of such screening was seen in the results of this research, where participants stated that they developed comorbidities, and that these were missed by their HCPs and as a result they failed to receive treatment when it was needed.

The fourth sub-theme regarding the management of OI was cardiovascular management. This sub-theme echoed the findings of the previous sub-theme. Matsushita et al. (2020) emphasised the need for screening for cardiovascular issues as Radunovic et al. (2011) state that issues with the cardiovascular system are associated with OI. The incidence of such issues was discussed briefly in the research as participants expressed that this was an area they knew little about. This finding was taken forward and incorporated into the guide.

The fifth and final sub-theme for the management of OI theme was pregnancy management. The literature in this field showed that complications are more common for individuals with OI, both Yimgang & Shapiro (2015) and Cozzolino et al. (2016) stated that as these are common, more care needed to be taken when deciding on treatment plans and that these should be individualised to meet their needs. Participants did not discuss incidence of complications during pregnancy but the need for individualised care was discussed at length.

The third theme identified in the literature review was guidelines. This revealed that there are no OI specific guidelines and that information on treating OI comes from guidelines aimed at general services such as a rheumatology or orthopaedics. Little reference was made to OI in the guidelines and those that did discuss OI were from NHS England and may not be applicable to the devolved nations of the UK. It can be argued that these guidelines facilitate misconceptions of OI as the recommendation for people with OI following their transition to adult services is to refer them to an osteoporosis specialist. This research showed that the majority of adults with OI are not seen by OI specialists and a great many people report that their HCPs mistake OI for osteoporosis. OI specific information is needed in order to address these misconceptions and to ensure people with OI receive the correct treatments.

The final theme identified in the literature review was living with OI. This theme discussed impact that OI has on individuals and the ways in which this impact can be diminished. The

literature shows that quality of life is negatively impacted by OI, Balkefors et al. (2015) found that people with OI had a reduced health related quality of life score. This reduction in quality of life is the result of a number of factors. Harsevoort et al. (2020) discussed the impact of fatigue on quality of life, this is a common symptom of OI and has a detrimental effect on quality of life. Further to this Nghiem et al. (2018) stated that quality of life is significantly impacted by pain. Both of these symptoms are known to be common for people with OI. The occurrence of pain was discussed in the qualitative phase of this research and the effect it had on participants was not insignificant. The literature also discussed some manners in which quality of life can be improved for people with OI. Balkefors et al. (2013) discussed the positive impact that surgery had on quality of life for people with OI. The findings of this research reflected this in multiple ways, some participants described the positive impact that surgery had on them, while others described how they felt surgery would have improved their quality of life.

The review of the existing literature has shown that there is a lack of understanding with regards to the management of OI in adults. This presents a challenge for both individuals with OI and for HCPs, this lack of information precipitates the misinformation with regards to OI. This perpetuates misconceptions and endorses a lower standard of care for adults with OI.

6.2 Qualitative phase

The first phase of data collection was the qualitative phase which consisted of a focus group and two interviews. These discussions were guided by the information gathered from the literature review and were focused on the participants experiences of transition and their care as adults. The transcripts from the qualitative phase were analysed using thematic analysis and this generated four main themes, each of these themes had three sub-themes.

The first of the four themes is ‘Knowledge’. The first sub-theme relating to knowledge was ‘HCPs without OI knowledge’ this showed that the lack of knowledge among HCPs led to many misconceptions regarding OI. This included mistaking OI for osteoporosis. The impact of these misconceptions ranged from not receiving adequate pain relief, to having fractures not diagnosed. This would prevent them from receiving treatment or referrals that they needed and ultimately would worsening their health status. This ‘Knowledge’ theme also contained

the sub-theme ‘Knowledgeable HCPs’ which discussed the benefits of HCPs with OI expertise. This showed the benefits of being treated by a knowledgeable consultant were not limited to receiving the best standard of care which could be individualised to suit them, but also led to the individual having a better understanding of OI. This was due to the specialist being able to answer their questions and provide them with the support they needed to manage their condition. The final sub-theme within the ‘Knowledge’ theme was ‘Patient knowledge’ where participants discussed their desire to have more information about OI and how they were unsure on where they should go for information and that this would leave them with questions and uncertainties about their condition which would hinder their efforts to manage it themselves.

The second theme identified was ‘Experiences of care’. Here the participants discussed their experiences within the health care system and from HCPs. The first sub-theme was ‘Frustration and fear’, here participants shared their experiences of being dismissed by their HCP and how their lack of knowledge of the condition left them fearful of complications. Here the impact of being dismissed was also seen as they described their frustration at the HCPs who had not listened to their concerns. The second sub-theme was ‘Dignity and respect’, here participants referenced the lack of respect that HCPs showed them. They stated that HCPs would not listen to them and that their knowledge and experience as someone who lived with OI was not respected. The final sub-theme within this ‘Experiences of care’ was ‘Coherence in health care’. This sub-theme showed that there were many inconsistencies in the treatment of OI and that this was dependent not only on the HCPs generally but that the region the individuals lived in also played a role as some were able to access specialist centres while others were restricted by distance.

The third theme was ‘Being proactive’. This theme discussed the actions people with OI needed to take in order to get the treatment or referrals that they needed. The first sub-theme was ‘Advocating for care’. Here participants discussed the ways in which they had to advocate for themselves and for their children. The lengths they needed to go to and the fervent nature of those efforts was often described as a “fight” and reflected the difficulty and that ‘fight’. The second sub-theme was ‘Being active in research’ and here the participants discussed how important they felt research was and how they felt that engaging in research for OI was important not only for the wider OI community but also for themselves

as it led to them having more screening or treatments that they would otherwise not have access to. The final sub-theme of this theme was 'OI community', here they discussed how beneficial the OI community had been to them. They stated that the use of social media, the internet and the BBS contributed to their efforts to educate themselves in OI and become better equipped to advocate for themselves and their children.

The final theme identified was 'Health care needs', this theme discussed both the current and future needs of individuals. The first sub-theme was 'Support and communication', here participants described the impact that a lack of support and communication had on them. They described how this poor communication and lack of support contributed to the difficulty in managing their condition as they had to take on the responsibility of passing information between HCPs. Here they also discussed the lack of transition services and how this left them without someone to turn to when they needed help. The second sub-theme was 'Referrals to specialists and regular appointments'. This included discussions around the frequency of appointments in adult services compared with paediatric services and how this shift was a shock to them and was difficult to adjust to. The importance of regular appointments to them was also discussed with participants advocating for regular check ups not only for medical interventions but also for sources of support and for providing them with reassurance. The final sub-theme was 'Multidisciplinary team management'. The participants shared how they had not experienced MDT management and that they felt this was important for providing holistic care.

The findings of this phase of the study were consistent with the literature and supported both the need for further investigation in OI and the need for resources to provide both individuals and HCPs with information about OI. These findings were used to develop a number of research questions which would be used to guide the exploration of the online semi-structured survey findings.

6.3 Quantitative phase

The second phase of data collection was the quantitative phase which consisted of an online semi-structured survey. This survey was developed using the data gathered in the quantitative phase and the literature review. The goal of this survey was to broaden the

understanding of the issues raised in the qualitative phase and to ascertain whether the experiences discussed was shared across the wider OI community. The results of the survey were explored using the research questions, each of these questions was then explored using a number of questions from the survey.

The first question posed following the qualitative phase was, 'Do people who have a smooth transition report a better experience in adult services?'. This was explored using the results from four of the questions in the survey. The response to the question regarding support from HCPs during their transition was crosstabulated with the question which asked whether care was better in paediatric or adult services. This showed that 22 out of 43 respondents stated that they did not receive support during their transition and that they felt care was better as a child. The question regarding support during transition was then crosstabulated with the question which asked participants to rate their overall experience in adult services. The most common response here was having no support during transition and a rating of poor for adult services, this selection was made by 14 out of 36 participants. The final analysis made for this research question compared the results of the support during transition with the responses to OI type. This was done in order to ascertain whether there was a connection between severity and level of support. The results for this cross-tabulation did not show a connection as almost all of the OI types reported not having support more frequently. There were no OI types which stated they did receive support at a higher rate than those that did not receive support. This suggests that the differences in treatment are not determined by severity of OI but are instead the result of other factors. This could be due to service availability, and this would negatively affect those who live in remote areas or those without the ability to travel for appointments.

The second research question was, 'Is there a connection between continuity of care and how people rate their care in adult services?'. To address this question, two sets of results from the survey were used. Here a crosstabulation was run between the question asking whether participants had experienced continuity in their care and the question which asked participants to rate their overall experience in adult services. The most common response combination for these questions was that they did not experience continuity and that their overall experience in adult services was poor. This suggests that there is a connection between continuity and having a positive experience in adult services. The lack of continuity

was also discussed in the free-text responses where participants expressed that paediatric care showed more consistency and offered better care that adult services.

The third research question, 'How does knowledge of HCPs vary between child and adult services?', was explored using two sets of results independently and then a crosstabulation was run. Firstly, the responses to the question regarding their paediatric consultant's knowledge was reviewed. Here 14 out of the 45 respondents rated their paediatric consultant's knowledge as excellent. This question was then asked about the participants consultant in adult services. This saw the number of consultants receive a knowledge rating of excellent drop to five out of 41 respondents. Although some participants dropped out of the survey between these questions, the percentage of consultants who received a rating of excellent dropped from 31% in paediatric services to 12% in adult services. These two questions were then crosstabulated, and the most frequent selection was good for adult service consultants and excellent for paediatric service consultants. This shows a shift in the rating and a drop in the level of knowledge when the participants transitioned to adult services. The importance of being treated by a specialist was also mentioned in the free-text responses, it was noted that many HCPs don't know what to do when it comes to treating people with OI. This also highlights the benefits of MDT management for people with OI.

The fourth research question, 'Is there a connection between poor care for adults and a lack of adult specialist services?'. In order to address this question, the question which asked whether participants had an adult consultant who is an OI specialist was reviewed. This showed that the majority of participants, 32 out of 42, did not have a specialist consultant. This finding was then compared to the results of the question regarding overall experience in adult services. This showed that the most frequent response in this crosstabulation was that overall experience in adult services was poor and that they did not have a specialist consultant, this combination of responses occurred 14 out of 36 times. This would suggest that having a consultant who is an OI specialist plays a significant role in the overall experience in adult services for people with OI.

The fifth research question was, 'Is there a link between being dismissed by HCPs and the occurrence of missed fractures?'. This was explored using two questions and a crosstabulation between these questions. Firstly, the results to the question which asked whether participants

had been dismissed or doubted by HCPs was used, this showed that 36 out of 39 respondents had experienced this in adult services. Secondly, the results regarding the incidence of fractures being missed was reviewed which showed that 32 out of 39 had experienced this in adult services. Finally, these results were crosstabulated and this showed that 29 out of 39 respondents had been both dismissed by HCPs and had fractures missed by HCPs. This was also reflected in the free-text responses where participants stated they had been doubted and disregarded in the past by HCPs. These findings suggest that there is a connection between these results and that in order for fewer fractures to be missed by HCPs, there is a need for a HCPs to listen to individuals with OI and work collaboratively with them when deciding on treatment plans.

The sixth research question was, 'Is there a connection between missed fractures and poor HCP knowledge?', this links with the previous question. The exploration of the previous question showed that missed fractures was a common occurrence in the sample population. This finding was then crosstabulated with the question which asked participants to rate the knowledge of their consultant in adult services. This showed that the most common response to these questions was that they had experienced fractures being missed and that they felt their consultant's knowledge of OI was good. This suggests that the issue might not solely be with educating HCPs in OI but that the issues of the previous question might be more influential. This also reflects the importance of educating HCPs not only in OI as a condition, but how to manage individuals with chronic conditions and the importance of respecting their expertise as someone who lives with the condition.

The seventh research question was, 'Are people who are members of the BBS more likely to have more knowledge and/ or to advocate for themselves?'. The participants were asked whether they were members of the BBS, the responses to this question were then crosstabulated to three other questions. Firstly, the BBS membership results were compared to the question which asked participants to rate their own knowledge of OI. This showed that 35 out of 39 respondents are members of the BBS and of those 35, 30 rated their knowledge as either excellent or good. This suggests that being involved in the OI community would help individuals to gain more knowledge in their condition. Secondly, the BBS membership results were crosstabulated with the question which asked whether participants share their opinion with their HCP. Here the most common combination of responses was that they are members

of the BBS but that they only shared their opinion sometimes, this combination saw 14 out of 39 responses. If this result is compared with the four respondents who are not members of the BBS, one selected that they would never share their opinion, one selected rarely and the remaining two selected sometimes. Thirdly, the BBS membership question was compared with the question which asked whether they would advocate for themselves in the healthcare setting. This showed that 16 out of 39 respondents stated that they are members of the BBS and would often advocate for themselves. When this is compared with the four respondents who are not members of the BBS, two selected that they would often advocate for themselves and the other two would never advocate for themselves. The need for advocacy was also seen in the free-text responses where participants describe this as a fight which mirrors the statements of the participants in the qualitative phase. These findings when taken together support the argument that the OI community is an important part of enabling individuals with OI to engage in self-management and to have the necessary skills and knowledge to advocate for themselves.

The final research question was, 'Is there a connection between GPs not giving referrals when requested and whether adults have a consultant and/or regular appointments?'. Two crosstabulations were conducted in order to address this question. The first crosstabulation used the question which asked whether participants had been able to get referrals from their GP and compared this to the question which asked whether they had regular appointments in adult services. The results to the question regarding regular appointments was split fairly evenly with 17 selecting 'yes' and 19 selecting 'no'. When these are compared to the GP referral question the results are less equal. Of the 17 who stated that had regular appointments, 15 of those were able to get referrals from the GP either always, often or sometimes. For those who stated they did not have regular appointments, only two stated they were always able to get referrals from their GP when needed. The second cross-tabulation for this research question compared the results regarding GP referrals and the question asking about general referrals to needed services. In this crosstabulation the most common selected was sometimes to both of the questions. This indicates that there may be other issues in obtaining referrals to services and that GPs are not the only ones who are inhibiting the referral process for adults with OI. Although these findings do not strongly show a connection, the difficulty in access referrals from GPs was seen in the qualitative phase and

again in the free-text responses of the survey where participants stated that GPs will not give them the referrals they need.

The online semi-structured survey findings were consistent with both the literature and the qualitative phase. These findings further support the need for a resource to fill the knowledge for both individuals and HCPs. These findings in combination with the findings of the qualitative phase, and the literature review were used to design a patient guide. The aim of which was to fill the gap recognised in this research.

6.4 Resource development phase

The final phase of this research involved the development of a patient guide for adults with OI. Feedback was then gathered on the guide from a number of different stakeholders, and this was the third and final data collection point of this research. Following each round of feedback collection, amendments were made to the guide to ensure it met the needs of both the OI community and included the best information for HCPs to allow for best practice to occur.

The feedback from the OI community reflected the importance of co-production as the participants were able to provide suggestions on additional content for the guide. This showed that the people within the OI community do have a great deal of knowledge about the condition and have ability to function in the role of the expert patient. Participants were able to recognise aspects of the condition that were missing based on their own experience. This would benefit not only themselves but the rest of the community too who may not otherwise known where to access information. This difficult in accessing information has been discussed throughout this research and shows that there is recognition of the lack of knowledge among individuals with OI but that they do not have the necessary support to find information and educate themselves.

The feedback collected from all stakeholders supported the use of the guide as a means of educating individuals but could also be utilised by parents of children with OI, HCPs, and any other people who need to have an understanding of OI. This guide provides not only information about the condition but also signposting and explanation of jargon they might encounter in appointments with HCPs.

6.5 Theoretical implications

A number of models and theories were utilised throughout this research. The elements of these models and their suitability for this research was considered throughout and this led to a number of strengths and limitations being identified within the models and theories. This section will explore these models and will suggest amendments to these models which may improve their utility in future research of a similar nature to this research study.

6.5.1 Movin' on up model

The were many aspects of this model which aligned with the findings of this research. Firstly, the need for education of individuals with chronic conditions. This is necessary to enable people to be proactive and look after themselves which would be a significant shift in their experience as paediatric care is heavily managed by HCPs and parents. Secondly, this model emphasizes the need for treatments to be maintained. A risk facing people during their transition to adult services is that treatments offered by HCPs in paediatric services are not guaranteed to be offered by HCPs in adult services. This model highlights that treatments need to be considered during the transition.

Despite this there were some topics that were missing from this model that did appear in the findings. Based on this, the movin' on up model was updated to include some elements that appeared repeatedly during the qualitative phase of the research. While most of the aspects of this model aligned well with the findings, there were some discussed in the focus group and the interviews which were not covered by the model. These changes can be seen in figure 5.

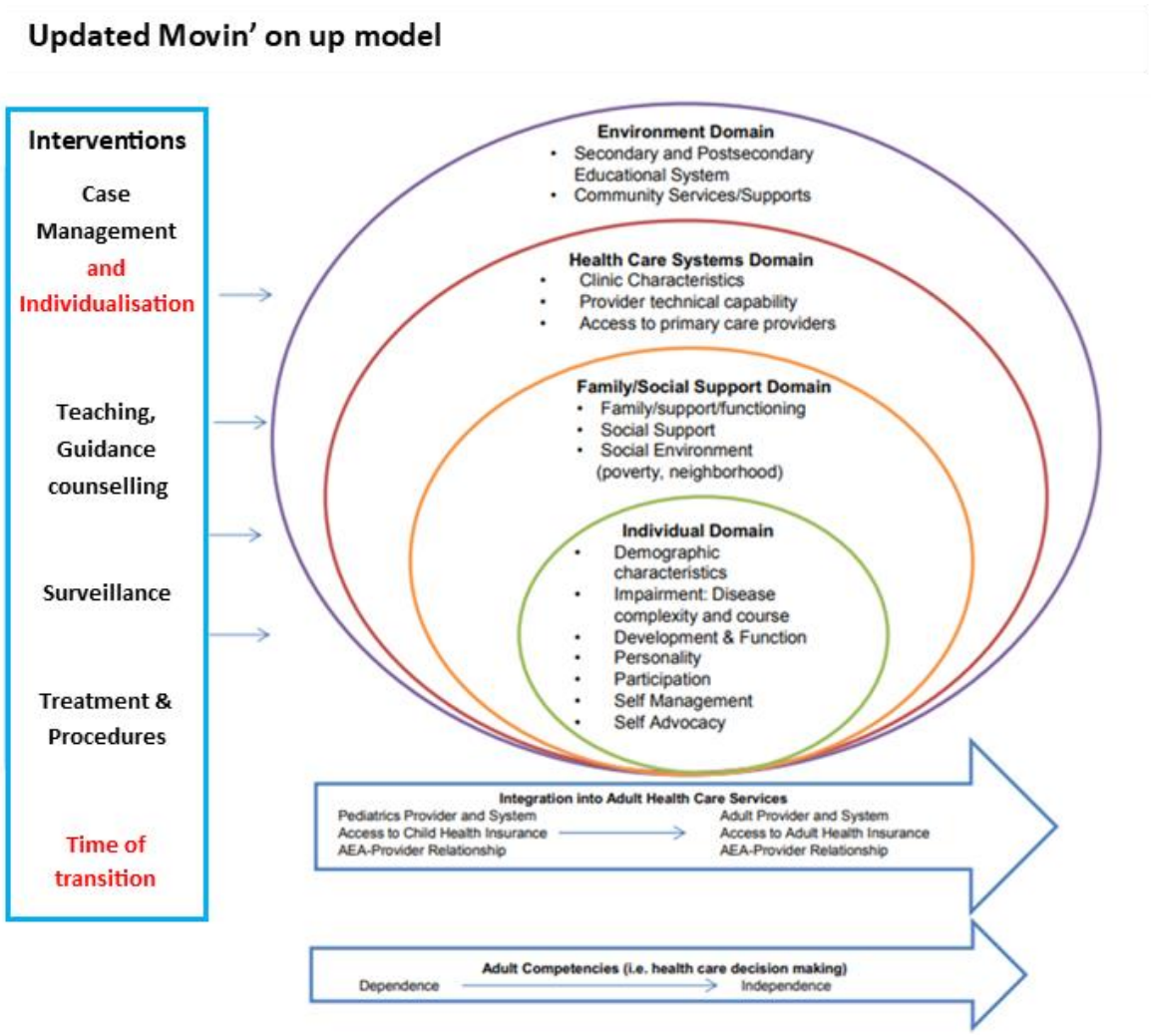

Figure 5: Updated movin' on up model

The first element which was added to the model was individualisation. While it can be argued that the case management aspect of the model would allow for individualisation, it was felt that the need for individualised care was vital and needed to be included. This came through in the results of the qualitative phase where participants expressed that they felt HCPs should have altered treatment plans in order to meet their specific needs. This was also seen throughout the literature and individualisation has been described as the best way of treating not only OI but chronic conditions in general.

The second element which was added to the model was time of transition. This was included as the time of transition can vary greatly depending on where the person is being treated. The current guidance states that transitions to adult services should occur at age 16, however some participants in the focus group stated that they were retained by paediatric services

until their early 20's. Therefore, the time of transition will be dependent on individual circumstances, and it should not be assumed that all individuals will transition to adult services at the same age. This was also seen in the qualitative phase where the age at which participants transitioned to adult services varied and was dependent on their current treatment needs.

These changes to the model would align with the findings of this research but would need to be reviewed in further research in order to validate this model as a useable tool for transition for people with OI and potentially other chronic conditions to adult services.

6.5.2 Minimally disruptive medicine care model

The MDM was utilised throughout all phases of this research. This model was selected because a number of the elements within the model aligned with the findings, however there were some elements which did not align. The first element which aligned with the findings was the capacity coaching element. This describes the measures which should be taken to assist individuals in managing the burden of their condition. The burden of illness is described throughout this research as the lack of services and knowledge of the condition causes undue stress on individuals. In the absence of adequate services to support the individuals they are forced to take on the burden of managing their condition. This presents an additional challenge to them, as the aforementioned lack of education and training for individuals results in them lacking the requisite skills and knowledge to take on this role.

The second element which aligns with this research is shared decision making. This has been shown to be an important part of managing chronic conditions in general and specifically for individuals with OI due to their complex needs. The ability for individuals to share their opinion with their HCP is a vital part of achieving empowerment and allowing them to have autonomy over their health care. This research showed that a lack of participation from individuals with OI was often the result of dismissal by HCPs.

The MDM is split into two sections, while the first section 'Tools to identify the right care', aligned with the findings of the qualitative and quantitative phases, the second section 'Tools to make the right care happen' did not. This was no longer the case following the resource development phase. This section of the model guided the development of the patient guide

with a number of the elements being incorporated into the design of the patient guide. These elements were; community navigators, wisdom leadership, and choosing wisely. Community navigators were an important aspect as the role of the OI community was seen throughout the research, with this community often being utilised as a source of information and support. Wisdom leadership refers to care being led by HCPs who have an understanding of the condition and are able to treat it effectively. This was lacking in the results of this research where participants reported poor HCP knowledge. Choosing wisely was essential as a means of signposting to services, which would enable individuals to seek care from the appropriate source, thereby preventing incorrect usage of services.

The main drawback of this model is reflected in the absence of a self-management element. Self-management has been a recurring theme throughout this research and this reflects the importance of it to adults with OI. Without self-management in the model, it is felt that this model does not fully align with this research and presents a limitation of the model which could be remedied with future research where elements could undergo feasibility testing.

6.5.3 Morgan self-management model

The findings of this research and the comparison of those findings with the existing model revealed that there were a number of limitations which meant the existing models were not wholly compatible with this research. As a result of this, a model was designed which would better suit the findings of this research and would provide a better framework for similar research studies in the future. When designing this model, it was clear that self-management was central to maintenance of the health of adults with OI. The other major aspects identified from the existing models and from the research all feed into self-management and are necessary to create self-actualised and autonomous patients. Although this research is focused on OI, this model is OI specific. The model covers issues which would be relevant to many chronic conditions, and it would be possible for this model to be used in the research of other chronic conditions and not solely for OI research.

The Morgan Self-Management Model (Figure 6) is centred around the individual rather than HCPs or the health system. This was done to enable individuals to take control of their health and have the skills needed to manage their condition successfully. It was clear from the

research that there is a high level of demand placed on the individual with a lot of responsibility placed on them. Without the requisite skills, they would struggle to advocate for themselves. This model will allow for resources to be developed with the individual in mind and ensure they are prepared for the challenges facing them in adult services.

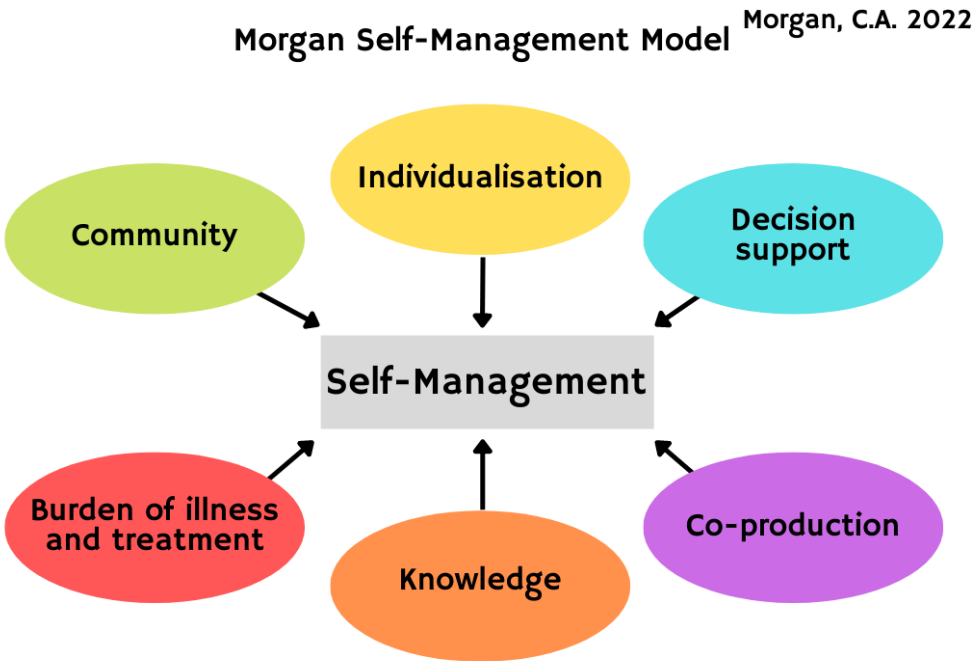

Figure 6: Morgan self-management model

The model is based around the Chronic care model (CCM) and the minimally disruptive care model (MDM) models, both of these models included vital aspects which have been carried into this model, but both also included a number of aspects which did not apply to individuals and would be beyond their control.

Although it can be argued that this model includes aspects which are reliant on cooperative HCPs, it is vital that individuals know what they need from their HCPs, and by providing them with the tools and the knowledge, they will be able to advocate for themselves and get what they need from their appointments and from their HCPs.

The community arm of the model refers to groups or organisations which support the individual. For this research, this consists of OI Facebook groups and the brittle bone society. In this research the impact of the OI community was seen in the qualitative phase with many participants expressing their utilisation of the BBS and Facebook groups to find information and to get support. One participant expressed that they *"got more support out of the OI community generally, than [they] did specifically from health care or social care professionals"*

(M, 26yo, T4). This shows that the OI community, and communities for individuals with chronic conditions generally, play a huge part in the support of people with chronic conditions and should not be overlooked as a resource in the development of self-management protocols or guidelines.

The individualisation arm refers to the need for treatment plans to be personalised to meet the needs and wants of the individual. This can be applied to this research as the needs of the individuals varied greatly and the differing types of OI result in vastly different symptoms and complications. The need for this element of the model was seen in this research as it was stated by many participants in the qualitative phase that they had not received treatments that were best for them and that their HCPs base treatments *"on the treatment plan for an osteoporosis person"* (F, 33yo, T3). This would be especially important for people with conditions like OI as their severity can vary greatly and as a result the treatment needs for individuals can also be very diverse, this could also be the case for other complex chronic conditions. This need for individualisation would be dependent on both the HCP and the individual having knowledge of the condition and the treatment options. In addition, the ability to make decisions regarding treatment would also require the individual to be supported by HCPs, to ensure their voice was being heard, this links to the decision support arm of the model.

The decision support arm, which was also present in the CMM, involves the need for individuals to be supported when making decisions regarding treatment. The need for support was expressed numerous times in the data. In the focus group, one participant stated that they were asked to make decisions about their care as a child, stating that their parent decided that *"at the age of eight I could make up my own mind"* (F, 33yo, T3), and that with hindsight they felt that they might have made a different decision. This supports that individuals with chronic conditions need to be supported throughout their life and this also highlights the need for individuals to be educated in their condition and its treatments.

The co-production arm of the model describes the need for treatment plans to be designed and agreed upon by both HCPs and the individual. The need for co-production was seen in the MDM, here the principle of co-production was highlighted in two elements, shared decision making and patient partnership. Both of these emphasize the need for the values of

HCPs and individuals to be respected whilst they are working together to ensure the treatments chosen are suitable. These principles are essential in the management of chronic conditions as the goal for treatment is not to cure but is instead to achieve the best possible quality of life for the individual. These principles link back to the need for individualisation and highlight that this process of individualisation is not only the responsibility of the HCP as it also requires cooperation from the individual.

The knowledge arm highlights the need for individuals and HCPs to be knowledgeable of the condition the individual has. In this research, knowledge was a major theme which the need for OI knowledge for individuals and for HCPs. In the qualitative phase, it was expressed by several participants that accessing information was difficult and that they have not been educated about their condition by HCPs and as a result they "*end up having to learn a lot of things for [themselves], which was quite a lot to take on*" (M, 26yo, T4). The pressure of having to find information for themselves also links to the next arm of this model which is burden of illness. Without sufficient levels of knowledge, people with chronic conditions such as OI cannot properly advocate for themselves, and it can be argued that without full knowledge of their condition they would be unable to give their fully informed consent. This presents an issue with regard to patient safety and introduces an ethical dilemma as HCPs have a duty of care and are required to ensure they obtain informed consent but in the case of complex chronic conditions, obtaining fully informed consent may not be achievable without first educating the patient. In addition, attempting to obtain consent for treatments or procedures from a person without full knowledge of their condition would impede their right to self-determination.

The burden of illness and treatment arm, which was also included in the MDM, refers to the demands placed upon the individual, including the physical demands resulting from their condition and the demands placed on them by the health system, such as arranging appointments or facilitating communication between HCPs. For this research, an example of the burden of illness and treatment was seen when individuals described the need for them to relay information between HCPs.

All six of these arms feed into and enable self-management. All of these elements are essential for the individuals to manage their condition.

6.6 Limitations and reflection

Despite best efforts being made to ensure this research was thorough, methodologically sound and achieved the aim and objectives set out in the introduction chapter, there were some limitations to this research study, and these will be explored and discussed in this section. This section will also reflect on the study and discuss changes that would have been made to the methodology with the benefit of hindsight.

One aspect of the study which must be reflected on is the changes made to the methodology of the qualitative phase of the research. The original methodology selected for this phase of data collection was focus groups. However, following significant challenges during the recruitment, it was decided that instead of multiple focus groups, there would be a single focus group and multiple one-to-one interviews in order to gather a sufficient amount of data. This change presented a number of challenges, and the changes must bring into question whether the data that was gathered might have been different had multiple focus groups been conducted. One of the challenges that the one-to-one interviews presented was the difficulty in getting the participants to discuss the topics when they did not have other participants to bounce ideas off of. In the first one-to-one interview, this proved to be very difficult. At the beginning of the interview, the participant needed a lot of prompting in order to go into any level of detail. This improved throughout the interview as they became more comfortable, but this does bring into question what they might have been willing to say had they been part of a group. The second one-to-one interview did not present this issue. The participant was more than happy to discuss the topics, however, as they did not have others to talk about issues with, they did have a tendency to go off topic and this meant it was necessary to remind the participant of the questions and offer prompts in order to ensure their discussions were relevant to the research.

It could be argued that these difficulties suggest that further recruitment should have been conducted in order to allow for another focus group to be conducted, however, after multiple attempts at recruitment, there was no further interest in participation, and it became clear that the research needed to move on to the next phase of the study despite these challenges.

A limitation which was seen throughout the research was the small sample size. In the qualitative phase the sample size was only six and this fell below both the target sample size and the recommended sample size laid out in the literature. The quantitative phase also fell below both the target sample size and the recommended size based on literature and power calculations. As a result, the data gathered did not have sufficient power to allow the null hypothesis to be rejected. Although the results regarding the overall experience in adult services were statistically significant, the other questions raised in this research could not be addressed adequately. This suggests that further research is required in this field in order to allow for these issues to be fully explored and the research questions at hand to be answered more effectively.

Numerous efforts were made to increase recruitment throughout the research. The recruitment post was advertised on Facebook multiple times and the BBS shared the recruitment advertisement on their social media accounts and on their website. The reasoning for the small sample size has been discussed in previous chapters and the explanation for this was that the OI population in the UK is small, meaning the pool of potential participants was small to begin with. In addition, not all of the OI community would be engaged in OI groups on Facebook and may not be members of the BBS. This means that there would be a proportion of the OI population which was inaccessible and therefore it would have been extremely difficult and time consuming to both find them and recruit them into the study.

Despite the small sample size, the findings of this research will be useful and can further the discussion into the issue of transition to adult services and the ongoing management of adults with OI.

Another limitation which follows on from the small sample size, is the use of quantitative methods for this research. When the research study was designed it was felt that the research needed to include a mixed of methods and that the sequential design would allow for the issues affecting people with OI to be identified and explored more broadly.

This phase of the research involved a detailed online semi-structured survey with the hope that this would allow for further understanding of the research questions which were

developed following the qualitative phase. Although the findings of this phase did offer some support to the findings of the qualitative phase, they did not have statistical significance and offered little new information. Based on this it is possible that this phase was not necessary and if this study were to be conducted again, in lieu of a quantitative phase, a larger qualitative phase may be more appropriate.

6.7 Implications for future research

The implications for this research are quite varied and there are wide ranging implications for both people with OI and people with other chronic conditions. Firstly, this research has shown that there is a need for further research into transition services in the UK. Although some hospitals do offer these services as discussed in the first chapter, these services are limited and not all people with OI would be able to attend them due to their geographical distribution across the UK. In addition, due to the lack of OI specific guidance regarding transition, the various centres will use different methods and consequently there will not be consistency in the transition experiences. Further research into transition services would provide HCPs with information regarding the best practice for transition and could allow for the development of guidelines for transition to adult services for people with OI which at the time of writing (August 2023), do not exist.

This research has shown that self-management is highly important for people with chronic conditions but is not easily achieved unless those with chronic conditions are both supported by HCPs and educated in their condition. The methods of supporting people with chronic conditions have been discussed in the existing literature and by participants in this research. These suggestions from participants ranged from having dedicated HCPs to offer them support and guidance in adult services to the provision of information packs. These methods are vastly different, and the effectiveness would need to be explored in order to determine which methods are the most effective and whether this would differ depending on the chronic condition. Future research exploring these methods and how they could be best introduced would need to be conducted. This could include the use of the Morgan self-management model which aims to enable self-management. In addition, the use of resources such as the patient guide developed in this research should also be explored as a method of enabling self-management.

The Morgan self-management model should also be researched as this model could be utilised in research for many different chronic conditions and the uses of the model should be fully explored. This could include a feasibility study testing the model, this would involve assessing each of the elements of the model using a deductive methodology to allow them to be tested. The model could be explored across a number of different chronic conditions and could utilise more powerful statistical approaches. Further research into the model could also allow for more elements to be added into the model and allow it to become a more useful tool for self-management.

The research implications for the guide that was developed for adults with OI are not limited to research involving OI. The methods used for the development of the guide could be used for many other chronic conditions and this would allow similar resources to be developed for a great number of other chronic conditions. Further guides would need to be researched and this would include ensuring that they are reliable and beneficial to individuals with the chronic conditions. Research with the support of charities relevant to those specific chronic conditions could be conducted in a similar manner to this research whereby the BBS provided a method of ratification through their MAB and SAB.

6.8 Implications for individuals and HCPs

This research has wide reaching implications to not only individuals with OI but also to HCPs and family members of people with OI. OI is a complex condition and the findings throughout this research have shown that the effects of the condition on people are profound. The condition affects all aspects of their lives and results in a litany of issues which the individuals are forced to deal with. The lack of services for adults with OI results in the isolation of adults with OI and the requirement for them to self-manage their condition. However, as was seen throughout this research, there is a lack of preparation for adults with OI and they are not equipped with the knowledge, or the tools needed to manage their condition successfully. One of the key findings of this research has been the identification of knowledge of OI as a gateway to successful management.

Ensuring people with OI have knowledge of their condition is not only important for ensuring they can ask for the correct treatments from HCPs, it is also important as it will ensure they

are able to give informed consent to any treatments or interventions offered by HCPs. Informed consent preserves the autonomy of the individual and ensures they have all the necessary information to decide whether a treatment is right for them (Man, 2013). Having an understanding of OI and the treatments would greatly aid in the provision of informed consent and would enable the individual to be more empowered by making the decision for themselves.

This provision of information will not only benefit people with OI but will also be beneficial for family members of people with OI. Although this study was not focused on parents of children with OI, there were discussions on the impact of having a child with OI in the qualitative phase. It was shown that having a child with OI allowed the participants to learn more about the condition and that they were motivated to learn about the condition to prevent their child from facing the same barriers they faced. Hill et al. (2019) also discussed how the parents of children with OI who want their children to be independent and know that the best way to achieve that is to equip them with knowledge and skills, but this cannot occur until they themselves have been educated and trained. Hill et al. (2019) discussed a study by Bozkurt et al. (2014) which stated that educating parents of children with OI had wider ranging benefits including reducing emotional burnout, reducing anxiety, and improving coping skills. They stated that a barrier to this was the lack of available information. This is consistent with the findings of this research and further supports the guide as a measure of improving access to information about OI.

Giving individuals the ability to be engaged in their care and allowing for a partnership to develop between the HCP and the patient is key for coproduction. Coproduction allows for treatment plans to be discussed and agreed upon by both HCPs and people living with the condition (Filipe et al., 2017). While this would be beneficial as it would take the wants and needs of the individual into account, it is dependent on the HCP being willing to listen to them and compromise to accommodate their needs. Within medicine this has not always been the norm with the “doctor knows best” attitude having been the norm for many years and often the decisions made by those HCPs was not questioned (Margolis, 1992). It is now understood that people have the right to say in their treatments and should be able to decide how to manage their condition. The attitude of HCPs has a significant role to play in the

successful management of OI. The ability to respect the wishes of the person with OI is essential and when this is not given, the mental health of the person with OI is put at risk.

If the HCPs are not able to respect the wishes of the individual, it is likely to affect the trust that person has in the health system. If they are frequently being dismissed by HCPs or their wishes are not being respected, it is possible that they will not want to seek help when they need it. This could result in comorbidities not being diagnosed or worsening unnecessarily. This would have a detrimental impact on their health and by extension, their quality of life. It was also seen in this research that access to expertise was dependent on a number of factors. These included where they lived, their participation in research studies and where their children were receiving treatment among others. This resulted in inequities in care, and this would result in some receiving far greater care than others. This would contribute to poorer health outcomes for some whilst enabling others to receive care which would be considered best practice for OI. Inequity could also be seen where individuals had been denied referrals to services they needed and were subsequently told that they would need to pay for those services privately instead of receiving them for free from the NHS. Although some people may be able to pay for services privately either to receive them more quickly or to have them provided at all, it is not possible for everyone. Many people from low-income households would not be able to pay for services themselves and as a result they would be left without the services altogether. This would result in people from disadvantaged backgrounds experiencing worse health outcomes than those who were able to afford the care privately.

The lack of knowledge among HCPs of OI and the benefits of utilising the expertise of expert patients, presents a significant barrier to care for people with chronic conditions. As has been discussed, this research has shown that people with OI report a high level of negative interactions with HCPs, not only being dismissed but also being the victims of the misconceptions held by their HCPs. As there is a lack of services for adults with OI and there are a limited number of specialists working with adults, this provision of education will enable more HCPs to fill their knowledge gaps and could allow more HCPs to provide the best standard of care for adults with OI.

Further to this, the results in this research showed there is a lack of services such as physiotherapy for adults with OI. The lack of such services for adults with OI following

fractures, surgeries, or other such problems, is likely to hinder their recovery or result in incorrect healing which can have long term issues. Not only can it lead to health-related issues such as reduced mobility, but this can also have an impact on the person's ability to work or care for children (Magaziner et al., 2015). Ensuring people with OI have sufficient access to rehabilitation services such as physiotherapy is not only important for their health outcomes but also their quality of life in general. Although this guide cannot directly address this lack of services, it provides people with OI with information regarding the different services they may need and provides signposting on how to access these services. One of the barriers described in the results was that referrals were refused by HCPs as a result of misconceptions regarding the condition and the associated comorbidities. This could also be addressed by educating HCPs and allow for more people to obtain the referrals they need.

Further misconceptions that appeared in the results were related to chronic pain and the lack of treatment or understanding of it in OI. The impacts of this are not limited to physical health but also to mental health. Chronic pain is known to have a detrimental impact on mental health (Turk et al., 2016). It affects sleep, which can affect the ability to do everyday tasks, including work, and will affect and compromise the person's ability to function and will reduce their quality of life (Hadi et al., 2019). If this chronic pain is not treated, the life of the individual will be severely affected, the impact would also be felt by the family of the person, as it is likely that they would turn to their family for help and support (Ojeda et al., 2014). Pain is included in the guide and this presents another potential source of education for both people with OI and for HCPs.

Issues such as pain, if not addressed can contribute to the burden of managing OI, which would have an effect on the life of an individual. This burden is not limited to coping with pain, but also includes booking and attending appointments and the communication between multiple HCPs. This burden would be universally faced by all individuals with OI, but for anyone with OI who is in education or is working, the burden of managing their work and their condition would be very exhausting and would likely have a detrimental effect on their mental health. For those with children with OI, the task of managing their care in addition to their own would be exceptionally difficult. Although there are many burdens associated with OI which cannot be resolved by changing treatments, there are other ways of easing the

burden that chronic conditions place on individuals, this could include altering the way appointments are conducted.

Advancing technology should be embraced by health service, the use of technology to enable health services to be provided has become essential since 2020. There are many benefits to their use, people with mobility issues do not need to travel to see their HCP and if individuals have chronic pain, they can still attend their appointment without putting themselves through more pain (Iacobucci, 2021) (Kolovos et al., 2021). These methods would also make multidisciplinary appointments much easier and would benefit the individual in many ways. This would allow for treatments to be discussed and agreed upon by all specialities, it would also reduce the number of appointments necessary which would save both time and money for the NHS and it would mean fewer appointments for the individual which would ease some of the burden of managing the condition. The use of online or telephone appointments would also help to ease the inequities resulting from the lack of specialists as individuals could have appointments with HCPs from anywhere and would increase the number of people who were able to access specialist OI care. This could improve their quality of life which is one of the main objectives in the management of chronic conditions like OI.

Although these burdens also cannot be directly addressed through the introduction of the guide, the guide presents an opportunity to education HCPs not only on the condition's symptoms, treatments, and complications, but also on the wider impacts that the condition has on an individual. This increased awareness could then encourage the adoption of other methods of appointments that would ease some of the burden on the individual.

6.9 Conclusions

The aim of this research was to investigate the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta. What this research has shown is that there are significant gaps in the knowledge of OI. These knowledge gaps create fundamental flaws in the management of the condition which commence at the moment of transition. The absence of sufficient services leaves them at the mercy of their OI and results in the need for them to take an active role in the management of their condition.

Without sufficient resources to provide them with the knowledge they need, they would be unsuccessful in their attempts to manage their condition. The patient guide provides the vital tool needed to fill this service gap and ensure adults with OI can be empowered, expert patients who are capable of taking ownership of their health and wellbeing. Future research should explore the applications of the patient guide for OI and the possibility of similar guides for other chronic conditions.

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Appendices

Appendix 1: Literature Search Results

Search	Databases	Search Terms	Inclusion dates	Total results	Duplicates	Off topic	Remaining
1	CINAHL Plus Full Text, MEDLINE, PsycArticles, PsycInfo	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "management" OR "managing" OR "manage"	2010-2020	226	45	7	174
2	CINAHL Plus Full Text, MEDLINE, PsycArticles, PsycInfo	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "transition"	2010-2020	21	2	4	15
3	ASSIA	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "management" OR "managing" OR "manage"	2010-2020	3	0	0	3
4	ASSIA	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "transition"	2010-2020	1	0	0	1
5	British Nursing Index	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "management" OR "managing" OR "manage"	2010-2020	21	0	3	18
6	British Nursing Index	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "transition"	2010-2020	5	0	0	5
7	Web of Science Core Collection	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "management" OR "managing" OR "manage"	2010-2020	194	22	12	160
8	Web of Science Core Collection	"osteogenesis imperfecta" OR "OI" OR "brittle bone disease" AND "transition"	2010-2020	23	0	11	12
				494	69	37	388

Appendix 2: Changes to Ethics application

Section 4 – As the research will not now be able to run on site of the University where will it now take place? How will confidentiality be assured?;

The research will now be run from my home, using a room which can be isolated and on a password protected computer.

Sections 6 and 18 – Please provide me with permissions from the relevant FB sites prior to posting your research;

Requests for permission have been included in Appendix 2 and screenshots of the conversations in which permission was given is included in Appendix 5.

Section 7 – So you are aiming for a maximum of 16 participants in total? What if more wish to participate, how will you select? What if you do not get sufficient interest, how many would make the research viable? How will the interview be recorded on GoToMeeting and what are the implications of GDPR for this type of recording? What will it record, just audio or visual as well?;

All responses to the recruitment post will be reviewed and if more than 16 respond, the 16 will be selected by myself in order to ensure a diverse group in the online focus groups. It is felt that a minimum of 10 participants would be needed.

GoToMeeting cannot be used as it is not available off campus, Zoom will now be used as the university has a licence for it and it can be used from my home. When recording it records both audio and visual but participants have the option of not showing their face if they do not want to be video recorded.

Participants will be able to see when they are being recorded as it is displayed on their screen. Security will be protected with this software as Zoom meetings can be password protected. Participants will need to have the meeting identification number and password in order to join.

Section 8 – Is it appropriate to offer peer support be provided during the research data collection FG?;

Participants will be able to support one another, providing peer support as the researcher will be a last resort if the other participants are not able to ease the distress of other participants.

Appendix 2 – Is the link to the PIS something that you have prepared in advance? Can you please include you and your supervisors contact details (name and e-mail addresses) on this advertisement? Can you also please include that the research has been approved by the Research Ethics committee, College of Human and Health Sciences, Swansea University? ;

The link was created for the PIS and is an active link. Supervisor details have been added as well as the statement.

Appendix 4 – Can you just change the word “patient” to “participant” on the bottom line please?;

This has been changed.

Appendix 5 – Can you please include supervisor details?

Supervisor details have been added.

As a final point, in the light of the ongoing situation, can you and your supervisors please take note of the attached instructions.

These instructions were reviewed and this research does not involve any face-to-face interactions.

Appendix 3: Focus Group Topic Guide

Investigating the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta

Topic Guide

19/03/20

Opening remarks

The researcher will;

Introduce themselves and thank the participants for agreeing to take part in the focus group

Go over the purpose of the focus group and answer any questions

Discuss the video and audio recording of the focus group and confirm they are happy with the recording

Discuss confidentiality and set ground rules and discuss how their data will be used and stored

Advise participants that they are free to leave the focus group at any time

Go through the consent form

Aim of focus group

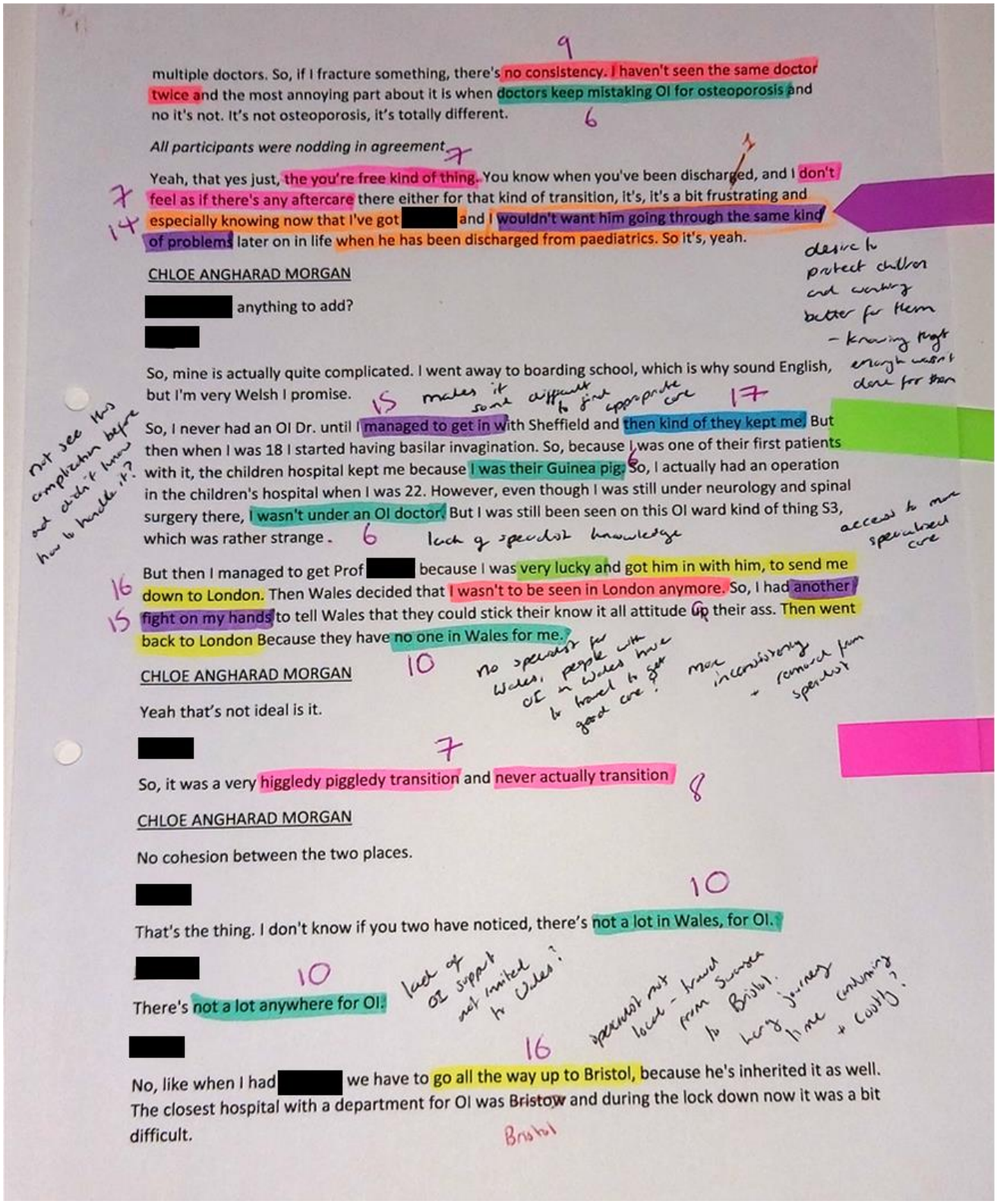
The aim of these focus groups is to develop an understanding of the experiences of people with Osteogenesis imperfecta relating to the transition from paediatric to adult healthcare services and the management of their care as adults.

Topics for discussion

The following have been identified as areas of interest from the literature and will be discussed in the focus group. There will be scope for participants to discuss issues they feel are pertinent and relate to the main topics for discussion.

1) Transition from paediatric to adult services
    a) Preparation for the move
        i) How long in advance of being discharged from paediatric services were you told that you would be discharged?
        ii) Looking back – what would have helped you move to adult services?
        iii) Which services were your referred to after being discharged if any?
    b) Education of people with osteogenesis imperfecta
        i) What information did you have before leaving paediatric services?
        ii) What information do you have now you are in adult services?
        iii) What additional information would you find helpful?
2) Management of care as adults
    a) The use of multidisciplinary teams
        i) Can you recall a time when your care has been overseen by multiple specialties?
        ii) What obstacles if any have you faced when trying to access the care or specialists you think you need?
    b) Are treatment plans custom-made or one size fits all?
    c) Expert-patient
        i) How have healthcare professionals made you feel regarding the management of your condition?
        ii) What have you done to ensure you have your say regarding treatment?

Appendix 4: Manual coding of transcripts



multiple doctors. So, if I fracture something, there's no consistency. I haven't seen the same doctor twice and the most annoying part about it is when doctors keep mistaking OI for osteoporosis and no it's not. It's not osteoporosis, it's totally different.

All participants were nodding in agreement

Yeah, that yes just, the you're free kind of thing. You know when you've been discharged, and I don't feel as if there's any aftercare there either for that kind of transition, it's, it's a bit frustrating and especially knowing now that I've got [redacted] and I wouldn't want him going through the same kind of problems later on in life when he has been discharged from paediatrics. So it's, yeah.

CHLOE ANGHARAD MORGAN

[redacted] anything to add?

[redacted]

So, mine is actually quite complicated. I went away to boarding school, which is why sound English, but I'm very Welsh I promise.

So, I never had an OI Dr. until I managed to get in with Sheffield and then kind of they kept me. But then when I was 18 I started having basilar invagination. So, because I was one of their first patients with it, the children hospital kept me because I was their Guinea pig. So, I actually had an operation in the children's hospital when I was 22. However, even though I was still under neurology and spinal surgery there, I wasn't under an OI doctor. But I was still been seen on this OI ward kind of thing S3, which was rather strange.

But then I managed to get Prof [redacted] because I was very lucky and got him in with him, to send me down to London. Then Wales decided that I wasn't to be seen in London anymore. So, I had another fight on my hands to tell Wales that they could stick their know it all attitude up their ass. Then went back to London Because they have no one in Wales for me.

CHLOE ANGHARAD MORGAN

Yeah that's not ideal is it.

[redacted]

So, it was a very higgledy piggledy transition and never actually transition

CHLOE ANGHARAD MORGAN

No cohesion between the two places.

[redacted]

That's the thing. I don't know if you two have noticed, there's not a lot in Wales, for OI.

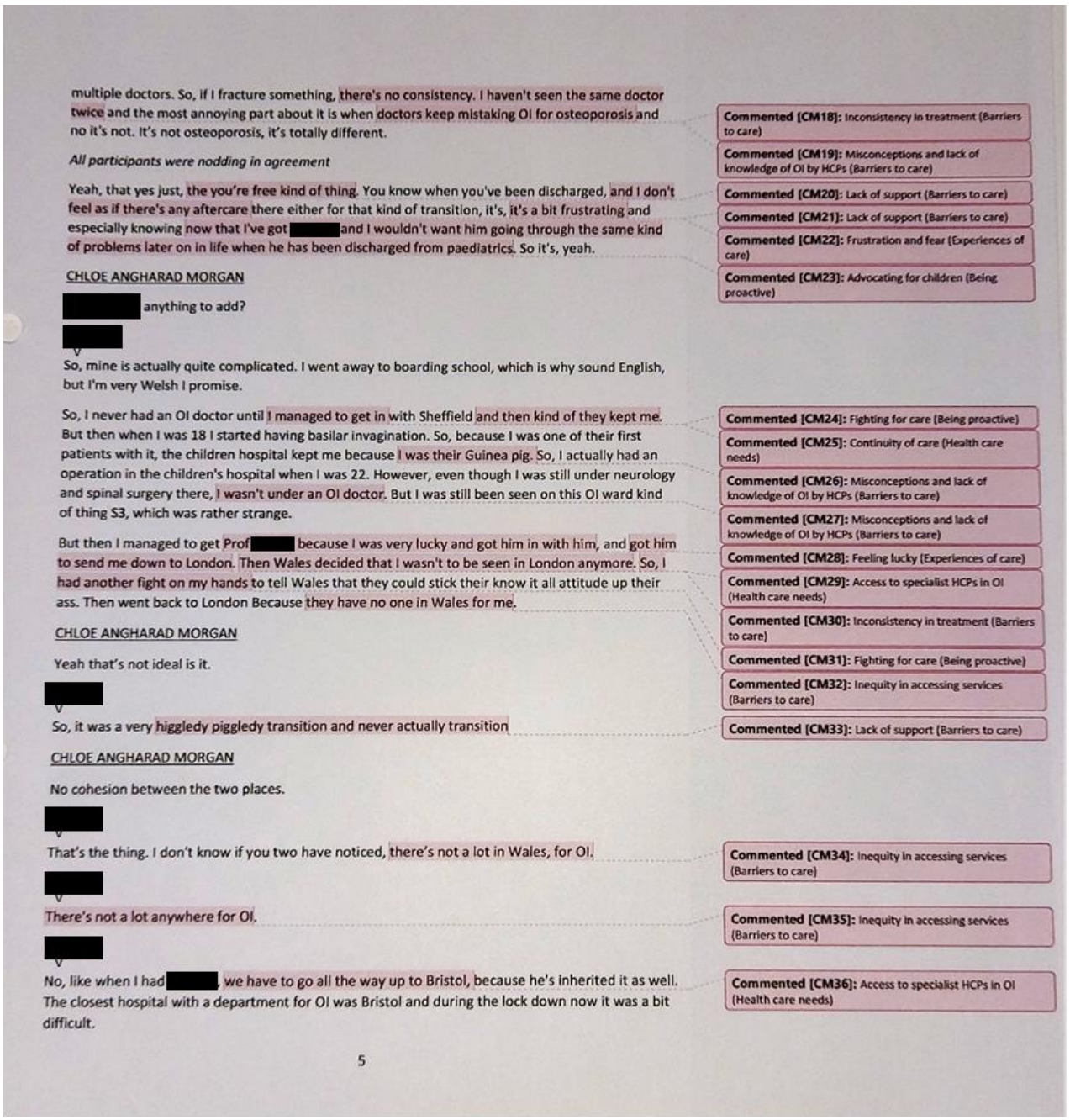
[redacted]

There's not a lot anywhere for OI.

[redacted]

No, like when I had [redacted] we have to go all the way up to Bristol, because he's inherited it as well. The closest hospital with a department for OI was Bristow and during the lock down now it was a bit difficult.

Appendix 5: Transcripts with themes and sub-themes



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[redacted]

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[redacted]

There's not a lot anywhere for OI.

[redacted]

No, like when I had [redacted] we have to go all the way up to Bristol, because he's inherited it as well. The closest hospital with a department for OI was Bristol and during the lock down now it was a bit difficult.

5

Commented [CM18]: Inconsistency in treatment (Barriers to care)

Commented [CM19]: Misconceptions and lack of knowledge of OI by HCPs (Barriers to care)

Commented [CM20]: Lack of support (Barriers to care)

Commented [CM21]: Lack of support (Barriers to care)

Commented [CM22]: Frustration and fear (Experiences of care)

Commented [CM23]: Advocating for children (Being proactive)

Commented [CM24]: Fighting for care (Being proactive)

Commented [CM25]: Continuity of care (Health care needs)

Commented [CM26]: Misconceptions and lack of knowledge of OI by HCPs (Barriers to care)

Commented [CM27]: Misconceptions and lack of knowledge of OI by HCPs (Barriers to care)

Commented [CM28]: Feeling lucky (Experiences of care)

Commented [CM29]: Access to specialist HCPs in OI (Health care needs)

Commented [CM30]: Inconsistency in treatment (Barriers to care)

Commented [CM31]: Fighting for care (Being proactive)

Commented [CM32]: Inequity in accessing services (Barriers to care)

Commented [CM33]: Lack of support (Barriers to care)

Commented [CM34]: Inequity in accessing services (Barriers to care)

Commented [CM35]: Inequity in accessing services (Barriers to care)

Commented [CM36]: Access to specialist HCPs in OI (Health care needs)

Appendix 6: Quantitative phase questionnaire

Investigating the transition from paediatric to adult health services and the management of ongoing care of adults with osteogenesis imperfecta

Questionnaire

Please complete all of the questions in this questionnaire.

Section 1 – About your OI

All of the questions in this section is about your Osteogenesis imperfecta (OI).

1) What OI Type do you have?

- ☐ Type I
- ☐ Type III
- ☐ Type IV
- ☐ Other
- ☐ Unknown Severe
- ☐ Unknown Moderate
- ☐ Unknown Mild

2) At what age was your OI diagnosed?

Years ____

Months ____

3) How was your OI diagnosed?

- ☐ With genetic testing
- ☐ Without genetic testing

4) Do you have any family members with OI? (Please tick all that apply)

- ☐ No- First in family to have OI
- ☐ Yes- Parent/s have OI
- ☐ Yes- One or more siblings have OI
- ☐ Yes- Another family member has OI

Section 2 – Paediatric experiences of care

All of the questions in this section should be completed based on your experiences in paediatric services and your transition to adult services.

5) Did you have a consultant who was an OI specialist as a child?

- ☐ Yes
- ☐ No
- ☐ Don't know

a) If yes, were you seen in a specialist OI centre?

- ☐ Yes
- ☐ No

6) How would you rate your paediatric consultant's knowledge of OI?

- ☐ Excellent
- ☐ Good
- ☐ Average
- ☐ Fair
- ☐ Poor

7) Do you feel the care you received for your OI was better as a child or not?

- ☐ Yes- it was better
- ☐ No- it was not better
- ☐ No difference

a) If yes, please explain in what ways was the care you received better?

_____

_____

8) Did you have support from healthcare professionals when being discharged from paediatric services?

- ☐ Yes
- ☐ No

a) If yes, please explain in what ways they supported your transition to adult services?

_____

_____

9) When discharged from paediatric services, were you referred to a consultant in adult services?

☐ Yes ☐ Don't know

☐ No

a) If yes, was there a delay in accessing a consultant in adult services and if so, how long were you without a consultant?

☐ Yes ☐ No

☐ 0-6 months ☐ 2-4 years

☐ 6-12 months ☐ 4+ years

☐ 1-2 years

Section 3 – Adult experiences of care

All of the questions in this section should be completed based on your experiences in adult services prior to the coronavirus pandemic.

10) Do you have a consultant who is an OI specialist?

☐ Yes ☐ No

a) If no, what kind of doctor is your consultant? (Tick all that apply)

- ☐ Rheumatologist
- ☐ Endocrinologist
- ☐ Orthopaedic
- ☐ Other (Please state)

11) How would you rate your consultant's knowledge of OI?

- ☐ Excellent
- ☐ Fair
- ☐ Good
- ☐ Poor
- ☐ Average

a. Please explain how your consultants level of OI knowledge has affected your care.

_____

_____

12) How would you rate your knowledge of OI?

- ☐ Excellent
- ☐ Fair
- ☐ Good
- ☐ Poor
- ☐ Average

13) What kind of information would you like to have access to or feel you would benefit from? (Tick all that apply)

- ☐ Fracture treatments
- ☐ Alternative therapies
- ☐ Pharmacological Treatments
- ☐ Hearing problems in OI
- ☐ Pain management
- ☐ Dentistry for OI/ dentinogenesis imperfecta
- ☐ Spinal problems in OI
- ☐ OI Genetics

- ☐ Heart problems in OI
- ☐ Breathing problems in OI
- ☐ OI Research
- ☐ OI Pregnancy
- ☐ Specialists
- ☐ Other (Please state)

_____

14) How would you want to receive information regarding OI? (Tick all that apply)

- ☐ Web-based
- ☐ Leaflet
- ☐ Poster
- ☐ Social Media
- ☐ Other (Please state)

_____

15) Have healthcare professionals given clear explanations of your condition, treatment options and support options to you?

- ☐ Yes
- ☐ No

a. If no, from where did you get information regarding OI, treatment options and support options? (Tick all that apply)

- ☐ Brittle Bone Society
- ☐ Osteogenesis imperfecta foundation
- ☐ Other websites (Please state)_____
- ☐ GP
- ☐ Specialist/ Consultant
- ☐ Other Healthcare professional
- ☐ Social media
- ☐ Friends/ Family
- ☐ Other (Please state)

_____

b. Of the options you selected, which would you go to first to get information regarding OI, treatment options and support options?

_____

_____

c. What prompted you to look for information regarding OI?

_____

_____

16) Do you actively research treatments offered to you?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

a. Do you share your opinion on these treatments with your healthcare professionals?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

17) Are you a member of the brittle bone society or social media groups for OI?

- ☐ Yes
- ☐ No (Go to Q18)

a. If yes, please explain in what ways being a member of these groups has helped you if at all?

_____

_____

18) Do you feel healthcare professionals respect you, your knowledge, and experience of the condition?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

19) Have you been dismissed or doubted when raising concerns to healthcare professionals?

- ☐ Yes
- ☐ No

20) Have you had fractures missed by healthcare professionals?

- ☐ Yes
- ☐ No (Go to Q21)

a. If yes, please explain what led to these fractures being missed.

_____

_____

21) How would you rate your confidence regarding advocating for your care?

- ☐ Excellent
- ☐ Good
- ☐ Average
- ☐ Fair
- ☐ Poor

22) Have you ever had to advocate with healthcare professionals for treatments/ tests to be provided?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

a. If yes, was advocating for your treatments/ tests from healthcare professionals successful?

☐ Yes ☐ No

23) Have you ever moved to a different area and moved to a different hospital or health board?

☐ Yes ☐ No (Go to Q24)

a. If yes, did care available to you differ between the hospitals or health boards?

☐ Yes ☐ Don't know

☐ No

24) Have you had healthcare professionals offer conflicting advice at the same time?

☐ Yes ☐ No (Go to Q25)

a. If yes, please explain what you do to manage the conflicting advice?

_____

_____

25) As an adult, have you experienced continuity in your care?

☐ Yes ☐ Don't know

☐ No

a. If no, how has this affected your care if at all?

_____

_____

26) Have you been able to get referrals to appropriate services when you have needed or asked for them?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

a. If no, what has prevented you from getting these referrals?

_____

_____

27) Has your GP referred you to appropriate services when needed?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

28) Have you had regular appointments with a consultant under adult services or not?

- ☐ Yes (Go to Q29)
- ☐ No

a. If no, would you like to have regular appointments?

- ☐ Yes
- ☐ No

29) At any point under adult services, have you been seen by a multidisciplinary team or not?

- ☐ Always
- ☐ Often
- ☐ Sometimes
- ☐ Rarely
- ☐ Never

a. If yes, please explain how this was facilitated and whether it was beneficial or not?

_____

_____

30) Do you have any children with OI?

☐ Yes ☐ No (Go to Q31)

a. If yes, please explain in what ways have you advocated for their care?

_____

_____

31) How would you rate your overall experience in adult services?

☐ Excellent ☐ Fair

☐ Good ☐ Poor

☐ Average

32) Is there anything else which you think is important regarding your transition from paediatric to adult health services and your ongoing care as an adult which you think is important and has not been addressed in the previous questions?

_____

_____

_____

Section 5 – About you

All of the questions in this section are about you and will help to give an idea of the experiences of people with OI and will allow for comparison between groups of people.

33) Age

_____

34) Gender

- Male
- Female
- Trans* male
- Trans* female
- Gender non-binary
- Self-Defined (please state)

35) Ethnicity

White

- English, Welsh, Scottish, Northern Irish or British
- Irish
- Gypsy or Irish Traveller
- Any other White background

Mixed or Multiple ethnic groups

- White and Black Caribbean
- White and Black African
- White and Asian
- Any other Mixed or Multiple ethnic background

Asian or Asian British

- Indian
- Pakistani
- Bangladeshi
- Chinese
- Any other Asian background

Black, African, Caribbean, or Black British

- African
- Caribbean
- Any other Black, African, or Caribbean background

Other ethnic group

- Arab
- Any other ethnic group

36) Country of residence

- ☐ England
- ☐ Scotland
- ☐ Northern Ireland
- ☐ Wales

37) Household income per annum

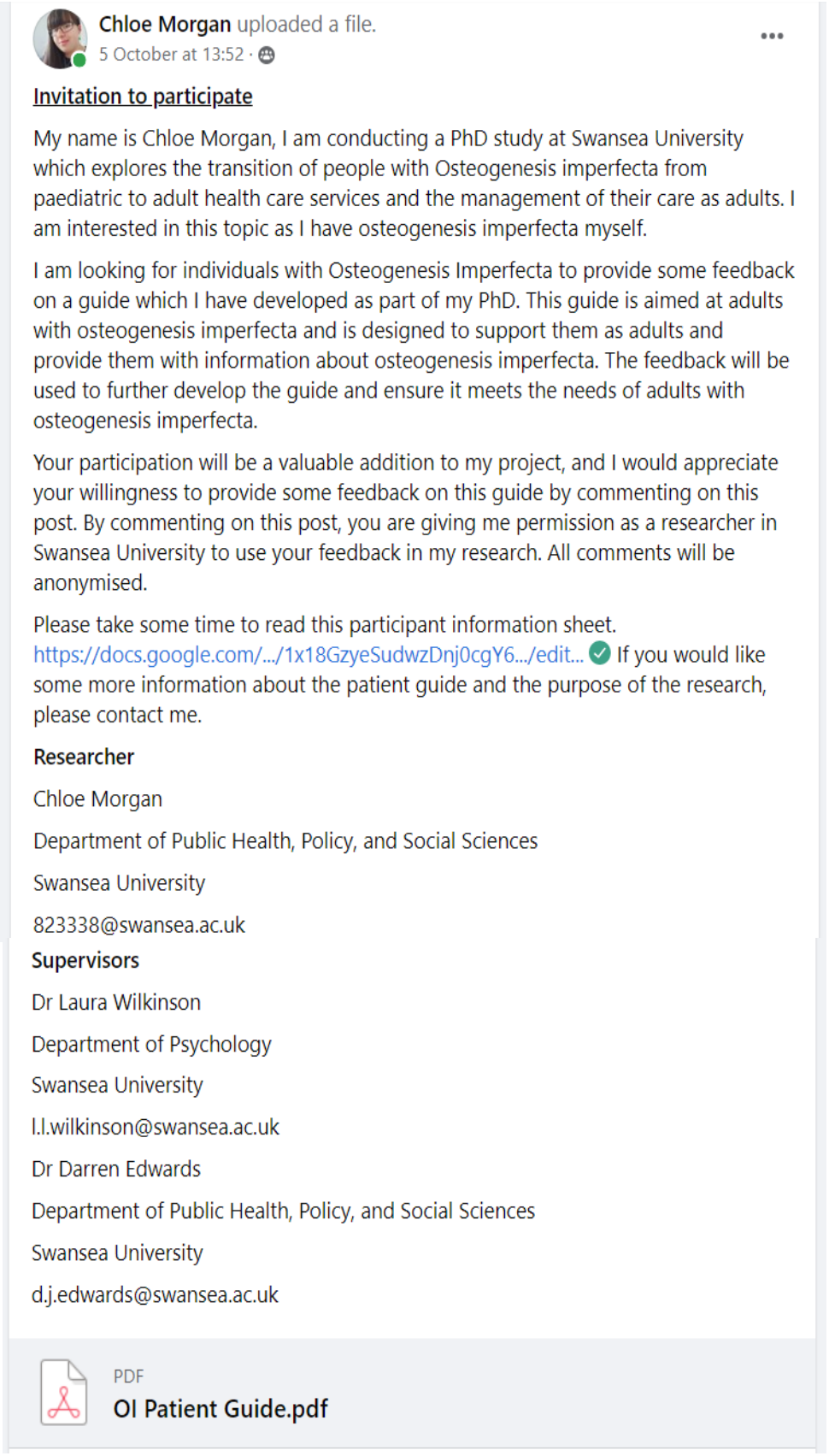
- ☐ £0-20,000
- ☐ £60,001-80,000
- ☐ £20,001-40,000
- ☐ £80,001+
- ☐ £40,001-60,000
- ☐ Prefer not to say

38) Education level

- ☐ No educational qualifications
- ☐ GCSE (or similar)
- ☐ AS Levels (or similar)
- ☐ A Level (or similar)
- ☐ Undergraduate degree (or similar)
- ☐ Postgraduate degree (or similar)

Thank you for participating in this study

Appendix 7: Facebook recruitment post



Chloe Morgan uploaded a file.
5 October at 13:52 ·

Invitation to participate

My name is Chloe Morgan, I am conducting a PhD study at Swansea University which explores the transition of people with Osteogenesis imperfecta from paediatric to adult health care services and the management of their care as adults. I am interested in this topic as I have osteogenesis imperfecta myself.

I am looking for individuals with Osteogenesis Imperfecta to provide some feedback on a guide which I have developed as part of my PhD. This guide is aimed at adults with osteogenesis imperfecta and is designed to support them as adults and provide them with information about osteogenesis imperfecta. The feedback will be used to further develop the guide and ensure it meets the needs of adults with osteogenesis imperfecta.

Your participation will be a valuable addition to my project, and I would appreciate your willingness to provide some feedback on this guide by commenting on this post. By commenting on this post, you are giving me permission as a researcher in Swansea University to use your feedback in my research. All comments will be anonymised.

Please take some time to read this participant information sheet. https://docs.google.com/.../1x18GzyeSudwzDnj0cgY6.../edit... If you would like some more information about the patient guide and the purpose of the research, please contact me.

Researcher

Chloe Morgan

Department of Public Health, Policy, and Social Sciences

Swansea University

823338@swansea.ac.uk

Supervisors

Dr Laura Wilkinson

Department of Psychology

Swansea University

l.l.wilkinson@swansea.ac.uk

Dr Darren Edwards

Department of Public Health, Policy, and Social Sciences

Swansea University

d.j.edwards@swansea.ac.uk

PDF
OI Patient Guide.pdf

Appendix 8: Participant information sheet

Investigating the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta

Participant Information Sheet

Dated: 08/03/21

You are being invited to take part in some research. Before you decide whether or not to participate, it is important for you to understand why the research is being conducted and what it will involve. Please read the following information carefully.

What is the purpose of the research?

I am conducting research on the transition from paediatric health services to adult health services for people with Osteogenesis imperfecta (OI) also known as Brittle Bone Disease. The purpose of the study is to understand how people with osteogenesis imperfecta manage moving to adult services and how this move affects the care they receive. Your participation in this study will take approximately forty-five minutes.

Who is carrying out the research?

The data is being collected by Chloe Morgan a PhD student in the Department of Public Health, Policy, and Social Sciences, Swansea University, under the supervision of Professor Joy Merrell and Dr Darren Edwards.

The research has been approved by the College of Human and Health Sciences Research Ethics Committee.

What happens if I agree to take part?

I will invite you to complete an online questionnaire. The questions will ask about your experiences regarding transitioning from paediatric to adult services and experiences of managing osteogenesis imperfecta as an adult.

You will be asked for some background information including your age, sex, type of osteogenesis imperfecta and what region of the UK you live in. You are not required to give your name. All data will be anonymous.

Consent will be required and recorded through a tick box before commencing the questionnaire.

Are there any risks associated with taking part?

The research has been approved by the College of Human and Health Sciences Research Ethics Committee. There are a few risks associated with your participation.

Firstly, becoming fatigued while completing the questionnaire as it will take around forty-five minutes to complete. If this is the case, please complete the questionnaire at a time when you feel able to concentrate for around forty-five minutes.

Secondly, it is possible that some questions may be distressing for some if they have had difficult experiences. Information on where to find information and support will be provided at the end of the questionnaire.

Finally, you will not directly benefit from being involved in the study. However, I plan to develop a guide for people with osteogenesis imperfecta to aid transition to adult services. It is hoped that this guide will be beneficial for other people with OI in the UK.

Data Protection and Confidentiality

Your data will be processed in accordance with the Data Protection Act 2018 and the General Data Protection Regulation 2016 (GDPR). All information collected about you will be kept strictly confidential. Your data will only be viewed by the researcher/research team.

All electronic data will be stored on a password-protected computer file in Swansea University. All paper records will be stored in a locked filing cabinet. Your consent information will be kept separately from your responses to minimise risk in the event of a data breach.

Please note that the data I will collect for this study will be anonymous.

What will happen to the information I provide?

An analysis of the information will form part of my report at the end of the study and may be presented to interested parties and published in scientific journals and related media.

Please note that all information presented in any reports or publications will be anonymous and unidentifiable.

Is participation voluntary and what if I wish to later withdraw?

Your participation is entirely voluntary – you do not have to participate if you do not want to. If you decide to participate, but later wish to withdraw from the study, then you are free to withdraw, without giving a reason and without penalty.

Data Protection Privacy Notice

The data controller for this project will be Swansea University. The University Data Protection Officer provides oversight of university activities involving the processing of personal data, and can be contacted at the Vice Chancellors Office.

Your personal data will be processed for the purposes outlined in this information sheet.

Standard ethical procedures will involve you providing your consent to participate in this study by completing the consent form that has been provided to you.

The legal basis that we will rely on to process your personal data will be processing is necessary for the performance of a task carried out in the public interest. This public interest justification is approved by the College of Human and Health Sciences Research Ethics Committee, Swansea University.

The legal basis that we will rely on to process special categories of data will be processing is necessary for archiving purposes in the public interest, scientific or historical research purposes or statistical purposes.

How long will your information be held?

The research data will be preserved and accessible for a minimum of 10 years after completion of the research.

What are your rights?

You have a right to access your personal information, to object to the processing of your personal information, to rectify, to erase, to restrict and to port your personal information. Please visit the University Data Protection webpages for further information in relation to your rights.

Any requests or objections should be made in writing to the University Data Protection Officer:-

University Compliance Officer (FOI/DP)

Vice-Chancellor's Office

Swansea University

Singleton Park

Swansea

SA2 8PP

Email: dataprotection@swansea.ac.uk

How to make a complaint

If you are unhappy with the way in which your personal data has been processed you may in the first instance contact the University Data Protection Officer using the contact details above.

If you remain dissatisfied, then you have the right to apply directly to the Information Commissioner for a decision. The Information Commissioner can be contacted at: -

Information Commissioner's Office,

Wycliffe House,

Water Lane,

Wilmslow,

Cheshire,

SK9 5AF

www.ico.org.uk

What if I have other questions?

If you have further questions about this study, please do not hesitate to contact us:

Researcher

Chloe Morgan

Department of Public Health, Policy, and Social Sciences, Swansea University

[redacted]@swansea.ac.uk

Supervisors

Professor Joy Merrell

Department of Public Health, Policy, and Social Sciences

Swansea University

[redacted]@swansea.ac.uk

Dr Darren Edwards

Department of Public Health, Policy, and Social Sciences

Swansea University

[redacted]@swansea.ac.uk

Appendix 9: Debrief sheet

Investigating the transition from paediatric to adult services and the management of ongoing care of adults with osteogenesis imperfecta

Debrief Sheet

Dated: 08/03/21

Thank you for completing an online questionnaire for the above project.

If you are seeking more information about Osteogenesis imperfecta, the Brittle Bone Society website has lots of information freely available at https://brittlebone.org/

If you would like to receive a summary of the research findings, then please let me know and I can email you when the study is completed. Findings of the study may be published in academic and professional journals.

If you have any further questions about your involvement with the study, please do not hesitate to contact me:

Researcher

Chloe Morgan

Department of Public Health, Policy, and Social Sciences, Swansea University

[redacted]@swansea.ac.uk

Supervisors

Professor Joy Merrell

Department of Public Health, Policy, and Social Sciences

Swansea University

[redacted]@swansea.ac.uk

Dr Darren Edwards

Department of Public Health, Policy, and Social Sciences

Swansea University

[redacted]@swansea.ac.uk

Appendix 10: Screenshots of feedback for patient guide

[redacted] my son was first diagnosed at age 10 he would wake in mornings feeling a bit dizzy initially thought it was ear issues but he had a series of tests at Gosh were they diagnosed BI but was deemed to well to undergo surgery as surgery was so risky in 2001,he was grand until he was 17 , he woke one morning unable to swallow, he was fitted with a peg for feeding which caused a perforated bowel which caused aspirated pneumonia from vomiting, by then he was too Ill to be considered for neck surgery. He kept slipping into comas for days at a time, wake up, for a few days, then go again, it was all very bizarre,until he finally didn't wake up at all. I believe nowadays the policy is to do surgery as soon as there are any symptoms so there is hope now if caught early enough.

Like Reply 3 w

[redacted] i am so sorry for your loss, your son and i were diagnosed around the same time, 2000/01. I also experienced a lot of dizziness, still do. I had my first surgery in 2004 and yeah it was very experimental.

Like Reply 2 w

[redacted] am so glad you are doing well,even though it's been a bumpy ride for you x

Like Reply 2 w

Reply to [redacted]

[redacted]
First glance love the colour and layout but lot of typos x

Like Reply 4 w

Chloe Morgan Author
[redacted] Thank you [redacted] The typos will definitely be resolved before this is shared further.
Any feedback on the type of content and the information included? Are there enough details etc?

Like Reply 4 w

[redacted]
Chloe Morgan still reading dont worry will give more later x

Like Reply 4 w

Chloe Morgan Author
[redacted] thank you 😊

Like Reply 4 w

3 [redacted]
I'd be happy to give feedback.
Im 26 from the Midlands, was under the care of Birminghams children's hospital until 18.
Like Reply 4 w

Chloe Morgan Author
[redacted] That's great thank you. Please leave any feedback as a comment on this post.
Like Reply 4 w

4 [redacted]
Loving the mention of Bone anchored hearing aids Chloe 😍 you could also mention Cochlear implants for profound hearing loss(rarer in OI) also MIXED hearing loss is more common in older OI people ie conductive and sensorineural.
Like Reply 4 w

Chloe Morgan Author
[redacted] Thank you for the suggestions. I will definitely try to add more to the hearing section.
Like Reply 4 w

4 [redacted]
You could also mention stapedectomy surgery which has been successful in some OI patients (usually milder types)
Like Reply 4 w

Chloe Morgan Author
[redacted] Thank you. I haven't read anything about that for OI. I will be sure to do some more research and add it in.
Like Reply 4 w

4 [redacted]
Chloe Morgan could I share your guide to other OI groups? You would need to make public from your own profile or send to me via messenger.
Like Reply 4 w

Chloe Morgan Author
[redacted] I only received approval from my ethics committee to share in this group for now. Thank you though
Like Reply 4 w

Reply to [redacted]

5 [redacted]

I found this an interesting read and very informative. I have a fair few points hope this is helpful. Thank you for putting this together to help people with OI.

Page 3, Black on dark purple and other colours hard to read, coloured background should be lighter more like the purple on next page. (I am dyslexic so might be why i struggled)

green fractures are more common in kids

WHATIS RODDING? needs a space pg 6

Page 8, u dnt explain wht the risks are just use medical terms
Maybe Bisphosphonates before pain management, and Cam follow on better from pain management for better flow of information

hydrotherapy more thn just swimming more like physio in the water so less weight/ stress on joints.

Page 11, dental implants, (depending on jaw densety and biophsphinate use)

Pregnancy page very interesting, as hard to find information about it.
Pg 15. HAEMORRHAGE/ BLEEDING ?? Oi related risk of general

Like the last page with picture of you and information why it was made

On page 17 you mention the bbs but don't say who they are (charity for oi, in uk)

Could suggest for more information on ou go to bbs website

Glossary is pg 18 not 19

Glossary could do with being in abc order so easy to find word, also not sure you mentioned all the words on this page

Further reading (ur references) is hard to read because of background

Images could be related to bones or people with oi rather than just shapes and stock images

Like Reply 1 w Edited

Chloe Morgan Author

[redacted] Thank you so much for all of your feedback. This will definitely be taken forward in the guide.

Like Reply 1 w

6 [redacted]

This is a great guide Chloe, well done, good luck with the PhD.

Couple of points I found going through.

Since the guide is aimed at adults but also has parts about genetics and having children, I have read this from a viewpoint of an adult who has OI who also has a child with OI. But I would perhaps consider that it could be read by a parent without OI who has a child with OI.

Page 5
Where should I go for treatment, perhaps look at other sources of information such as NICE guidelines as I would also recommend calling 999 for open fractures, femoral fractures, pelvic fractures, unstable tib/fib fractures, amongst others not just limited to fractures of the spine.

Page 6

Telescopic rods, although they grow with the child they may need changing if the child outgrows the rod, some of our children have them inserted at a young age and they need changing out before the child has finished growing in addition to the reasons you mentioned.

Page 8
Bisphosphonates, not all individuals have a DEXA scan before starting treatment, my son started on treatment before he was old enough to have a DEXA scan, i think it was because they don't have the reference values for the age group below 5 years old to work out the Z score.

Page 10
Building muscle mass also helps to reduce fractures.

Physiotherapy/Occupational therapy.

A lot of the specialist OI Centres have PTs/OTs on their OI team and they would be my first port of call rather than community PT/OT through GP as they are already familiar with OI and will have a greater understanding of the effects of the condition. (Also couple of typos on the hydrotherapy bit "muss" instead of mass and "fot" instead of for.

Page 11
DI
I had a referral to a specialist dental hospital by my adult OI team, this was very useful as they had the understanding beyond a normal dentist of how to treat DI. They did say dental implants were a no go in OI due to the risks of it failing due to OI in the jaw bones but i don't know if this is blanket rule or not.

6

Page 13
Atrial Fibrillation also puts people at increase of blood clots that can cause strokes/heart attacks and people are usually medicated with an anticoagulant to reduce the risk of the blood clots.

Page 16

Genetics
It may be worthwhile mentioning that for those with OI who are planning to have children a referral can be made by GP/OI team for genetic counselling. This was useful for me and my wife as they helped explain to my wife (who does not have OI) how the genetics work and also were able to advise on what was available in terms of IVF to select embryos that don't carry the OI gene (we weren't eligible for this for reasons I can explain if you wish) Throughout the pregnancy we were under consultant obstetrician led care and this was also important for us with extra scans and a birth plan. We were also offered amniocentesis which we declined.

Hope this feedback is helpful/of use and good luck with the PhD.

7

[redacted]
Really interesting Chloe well done. I have a much delayed appointment at the hospital on 8th November with Dr [redacted] now have lots of questions to add to my usual list. Feel very neglected at the moment at the lack of interest and help in the NHS.

Like Reply 6 d

Chloe Morgan Author
[redacted] thank you for your feedback. I hope your appointment goes well

Like Reply 6 d

8

[redacted]
The guide is amazing. The colours and layout are lovely and makes it easy to read. Well done!!

There is however, something I would love to finally see written or spoken about is OI and the impact of mental health and the help available. 🙂

Like Reply 9 w

Chloe Morgan Author
[redacted] Thank you for your feedback. Mental health and support for mental health is definitely something I would like to incorporate in the guide in the future.

Like Reply 9 w

Appendix 11: Changes made to guide from Facebook feedback

Page 4	Colour of text boxes changes. Typos corrected.
Page 5	Clarified that greenstick fractures are more common in children. Added that 999 should also be phoned for "open fractures, pelvic or femur fractures or unstable tibia fractures." Typos corrected.
Page 6	Added that "children can still outgrow these rods as they have a maximum length they can extend to." Typos corrected.
Page 7	Typos corrected.
Page 8	Added explanation of osteoporosis and osteopenia. Added more common side effects of bisphosphonates and explained the rarer side effects. Typos corrected.
Page 10	Additional explanation of hydrotherapy. Typos corrected.
Page 11	Added that dental implants are not always suitable for people with OI. Typos corrected.
Page 12	Added section on mixed hearing loss. Added stapedectomy and cochlear implants to treatment section.
Page 13	Added risk of blood clots and treatment of this to atrial fibrillation section. Typos corrected.
Page 14	Expanded section on basilar invagination by adding symptoms. Typos corrected.
Page 15	Explained that "You will be seen by obstetrics who will help you to decide on a birth plan. You and the baby will be monitored closely."
Page 16	Simplified the explanation of recessive inheritance. Added that genetic counselling is available.
Page 17	Added information on the Brittle Bone Society

Page 18	References were split into groups according to the pages they were on to make finding the references easier.
Page 19	Glossary was put in alphabetical order. Typos corrected. Pages 18 and 19 put into correct order.
Page 20	References split across two pages to make reading easier

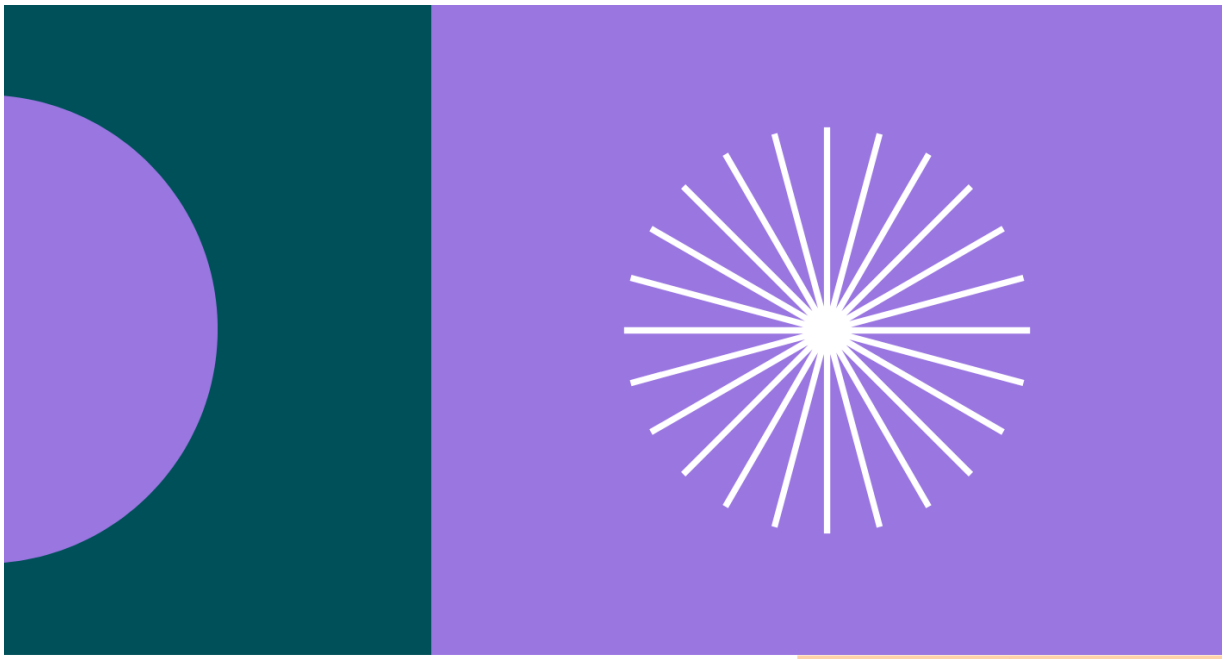
Appendix 12: Changes made to guide from supervisor feedback

Additional page	A page was added explaining the different types of OI
Move page	Genetics page was moved forward to come after the "OI types" page
Page 2	Make contents page clearer
Page 3	Added explanation of the references in the guide and how to use them
Pages 8, 9, 17	Typos corrected
Page 19	Made the web addresses in the reference list hyperlinks to make the references more accessible
Page 20	Paragraph reworded, "This guide was informed by research studies, NHS choices website and guidance from governing bodies such as NICE"

Appendix 13: Changes made to guide from BBS feedback

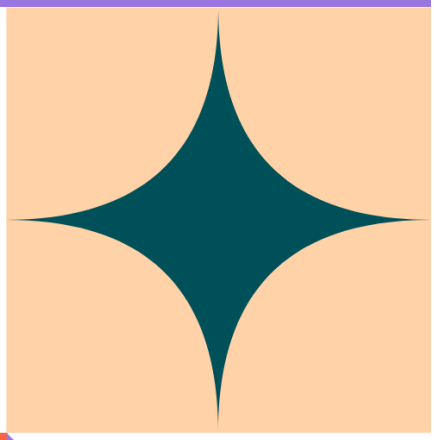
Page 6	Typo in gene name SERPINFI should be SERPINF1
Page 7	Mention vertebral fractures in fracture section
Page 8	Telescopic rods, express these are not for adults and cannot be left in permanently
Page 10	Reword the explanation of how bisphosphonates work Clarify that most side effects occur within 48 hours and usually only with first treatment
Page 12	Explain more of what physiotherapists can do OTs involved in wheelchair provision but not other mobility aids
Page 15	OI was not affected by covid-19
Page 16	Scoliosis and kyphosis would not usually be treated with braces in people with OI
Page 17	OI babies are more likely to present in the breech position and are more likely to need a caesarean
Page 18	Difference between HSS centres and the regional centres

Appendix 14: Final version of patient guide



OSTEOGENESIS IMPERFECTA

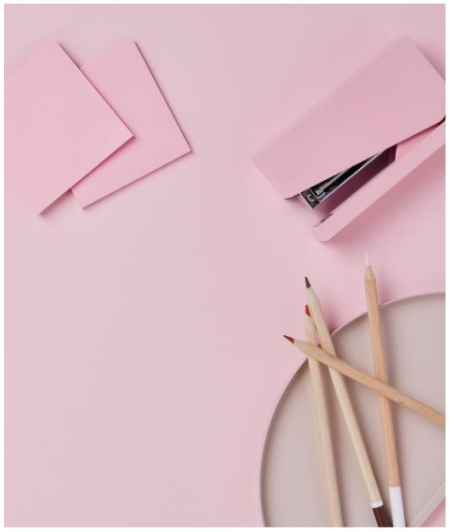
The pocket guide for adults living with OI



Chloe Morgan
Dr Laura Wilkinson
Dr Darren Edwards
Professor Joy Merrell

Version 4
January 2023

TABLE OF CONTENTS



WHAT IS THIS GUIDE FOR?

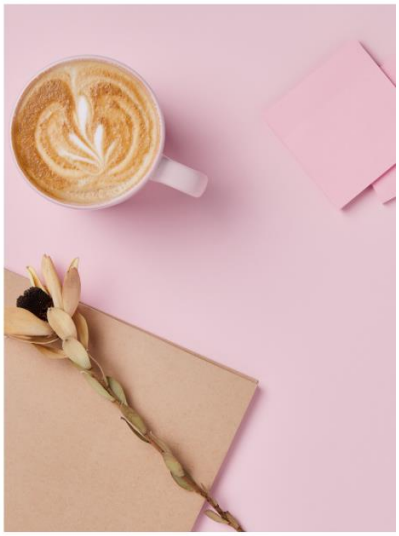
This guide was made to ensure people with OI have access to information about their condition.

The aim of the guide is to allow people with OI to make informed decisions about their care and manage their condition.

WHO IS THIS GUIDE FOR?

Although this is mainly aimed at adults with OI this could be used by anyone who wants to learn more about the condition.

You could refer to this in appointments with your healthcare professionals.



HOW TO USE THIS GUIDE?

Throughout this guide you will see numbers inside brackets like this (1), these numbers correspond to the hyperlinks included on the References pages. You can use these to read more into topics that are interesting and / or relevant to you.

GENERAL OI INFORMATION

WHAT IS OI?

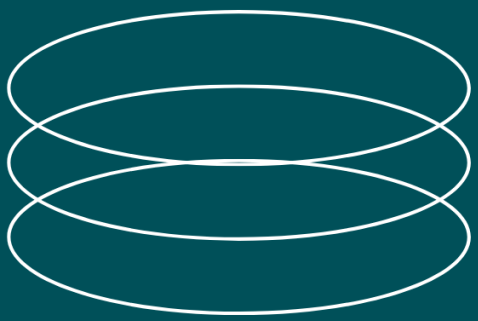
Osteogenesis imperfecta / Brittle Bone Disease is a genetic condition which results in your body making weaker collagen or not producing enough collagen. (1)

WHAT DOES COLLAGEN DO?

Collagen provides the structure to many of the tissues in your body. When it is weak or there is not enough, those tissues are weaker than they should be. (2)

HOW COMMON IS OI?

OI is esitmated to affect one in 15,000 people in the UK. This means almost 5,000 people in the UK have OI. (3)



IS THERE A CURE?

There is no cure for OI but with treatment people with OI can live very productive lives and be successful. (4)

OI TYPES

Thanks to genetic testing, more types of OI have been identified in recent years. There are currently 21 identified OI types (5), these range in severity, some cases are very mild and others can be very severe and sometimes fatal. Types I, II, III and IV are the most common and account for 85-90% cases of OI.

This table shows the known types of OI as of January 2023. The severity for each type is shown but this varies for everyone and should be used as a guide only. This also shows the inheritance pattern for each of the types. These patterns of inheritance are explained on the following page of the guide (Genetics - page 6).

Type	Severity	Inheritance	Gene
I	Mild	Dominant	COL1A1/2
II	Fatal	Dominant	COL1A1/2
III	Severe	Dominant	COL1A1/2
IV	Moderate	Dominant	COL1A1/2
V	Moderate	Dominant	IFITM5
VI	Moderate	Recessive	SERPINF1
VII	Severe	Recessive	CRTAP
VIII	Severe	Recessive	P3H1
IX	Severe	Recessive	PPIB
X	Varies	Recessive	SERPINH1
XI	Varies	Recessive	FKBP10
XII	Moderate	Recessive	SP7
XIII	Severe	Recessive	BMP1
XIV	Varies	Recessive	TMEM38B
XV	Moderate	Recessive	WNT1
XVI	Severe	Recessive	CREB3L1
XVII	Varies	Recessive	SPARC
XVIII	Severe	Recessive	TENT5A
XIX	Severe	X-Linked	MBTPS2
XX	Varies	Recessive	MESD
XXI	Varies	Recessive	KDELR2

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GENETICS

Many discoveries have been made in genetics in the last few decades. More is known about OI now as a result of developments in the field of genetics. This brief summary includes some of the key information regarding genetics and OI.

CAN OI BE PASSED ON TO CHILDREN?

OI can be passed on to children. The likelihood of it being passed on depends on the type of gene which it affects.

If you have OI and are pregnant or are considering whether you want to have children, you can be referred for genetic counselling by your GP or by the OI team managing. They will be able to advise you on the options and explain the chances of your child having OI.

DOMINANT GENES

Most cases, 85-90% occur in dominant genes. (6)
Dominant genes are more common in OI. Only one copy of the gene is needed in order for the person to have OI. Therefore, there is a 50% chance of inheriting it from a parent who has OI. (7)

RECESSIVE GENES

Only 10-15% of OI mutations occur in recessive genes. Two copies of the faulty gene are needed to have a recessive form of OI. This means it would need to be passed on by both parents in order for the person to have OI. If both parents have one recessive OI gene, there is a 25% chance that the child will have OI. (8)

However, people with only one copy of the recessive gene for OI may never know as they would not have OI. This is because they have a healthy version of the gene which cancels out the negative effects of the faulty gene.

X-LINKED GENES

X-linked means the gene is found on the X chromosome. The X and Y chromosomes determine the sex of the person, women have two X chromosomes and men have an X and a Y. Having a mutation on an X chromosome is more likely to affect men than women. Women will likely have a healthy version of the gene on one of the X chromosomes which will prevent the mutated gene from causing disease.
As men only have one X chromosome, they will not have a healthy version of the gene and will therefore have the condition.

FRACTURES

Fractures/ Breaks are common for people with OI and often occur without trauma. Around 25% of lifetime fractures occur during adulthood for people with OI. (9)

TYPES OF FRACTURES

These are some of the types of fractures you might be diagnosed with and what they mean. (10)

Non-displaced	The bone is still in the correct position
Displaced	The bone has moved and is not in the correct position
Closed	The skin is not broken
Open/ Compound	The bone has come through the skin
Comminuted	The bone has broken into small fragments
Hairline/ Stress	A small fracture, may be called a crack in the bone
Greenstick	The bone is broken but not all the way through the bone and may be bent. These are more common in children.
Avulsion	A ligament or tendon has pulled a piece a bone out of the correct position
Pathological	Caused by a disease which weakens the bones

Fractures in people with OI are often missed due to the low bone density and fractures may not be seen until healing begins.

HOW ARE FRACTURES TREATED?

How your fracture is treated will depend on what type of fracture it is. Some fractures won't need any treatment and will be able to heal on their own, some may require the use of a plaster cast or splint to keep the fracture stable. Some severe fractures may need surgery. (11) (12)

Vertebral (spine) fractures are common in OI. More information on how these can be managed and treated can be found on Page 16.

WHERE SHOULD I GO FOR TREATMENT?

NHS advises that;

Minor injury units or urgent care centres deal with fractures in fingers or toes

A&E departments should deal with fractured arms or legs

Call 999 for fractures in the spine or neck, open fractures, pelvic or femur fractures or unstable tibia fractures

SURGERY

Surgery is often part of life for people with OI. Either to resolve fractures, or to correct deformities of the limbs such as bowing. Surgery can improve mobility and prevent fractures. (13)

WHAT IS RODDING?

Rodding surgery involves placing metal rods, often titanium or steel, in the middle of the bone (the medulla) this surgery can also be referred to as intramedullary rodding.

There are many different kinds of rods which can be used, and this will depend on the size and shape of the bone. Most rods will fit into one of two categories: stable rods or telescopic rods. (14)

STABLE RODS

The rods are a fixed length.
Used more in adults as they are no longer growing.

TELESCOPIC RODS

These rods will extend as the bone grows.
Good for children who are still growing but are not used in adults.
Children can still outgrow these rods as they have a maximum length they can extend to.
These rods are susceptible to failure and cannot be left in permanently.

HOW LONG DO RODS LAST?

Unlike replacement joints, rods are not under mechanical stress and do not degrade over time. They can be left in for as long as is needed unless there are complications such as fractures or infections.

There is also a risk rods migrating out of the bone and they can also bend or break.

OTHER METALWORK

Metal plates and screws may also be used in the event of fracturing. There is a risk of stress fracturing around screws in OI bones.

OSTEOTOMIES

An osteotomy involves cutting and reshaping a bone. This can be done to straighten a bone and correct bowing.

Osteotomies may need to be performed before metalwork can be placed if there is bowing in the bone.

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PAIN MANAGEMENT

Pain can be either acute or chronic and both types of pain are common for people with OI and there are many ways to deal with it. Acute pain is short term pain which might be from an injury. Chronic pain is when pain lasts for more than six months. Your GP can offer help and advice for chronic pain and there are a number of things they can offer.

PAIN MEDICATIONS

There a number of different medications that are used to treat pain. This table shows the different categories of medications and some examples. This is not exhaustive and not all medications are suitable for everyone. (15)

Non-opioid	Paracetamol	
NSAID's	Ibuprofen	Naproxen
Opioids	Codeine	Morphine
Adjuvant analgesics	Lidocaine	Benzodiazepine

WHAT IS A PAIN CLINIC?

Pain clinics are specialist services which deal with chronic pain. They can offer different medications, injections and therapies such as physical therapies and/ or complementary therapies.

Some people who attend pain clinics can be offered a place on a pain management programme. (16)

PAIN MANAGEMENT PROGRAMMES

Pain management programmes teach you how to live with chronic pain. These would be useful for people who do not benefit from the use of pain medications.

The function of these programmes is to teach you coping strategies and how to prevent pain flare ups by better understanding your pain and it's triggers.

The aim of these programmes is to improve quality of life and allow you to take control of your situation and get the most out of life. (17)

BISPHOSPHONATES

Bisphosphonates increase bone mineral density (BMD) and make bones stronger. Possible outcomes of bisphosphonate therapy are reduced pain, fewer fractures and increased mobility but these are not guaranteed. (18)

HOW DO BISPHOSPHONATES WORK?

Bisphosphonates work by reducing how quickly bone is broken down, the reduction in bone breakdown increases bone density and reduces bone fragility.

WHO CAN HAVE BISPHOSPHONATES?

Bisphosphonates are given when bone mineral density is lower than it should be. People with OI often have lower bone density and if it is found that the bones have osteopenia or osteoporosis in addition to osteogenesis imperfecta, then there is an increased risk of fracturing, and you might be offered bisphosphonates.
Not everyone with OI can be given bisphosphonates and you will need to have your bone density checked with a DEXA scan before it can be prescribed. Your bone density will need to be monitored while you are having bisphosphonates.

ARE THERE DIFFERENT BISPHOSPHONATES?

There are a number of different bisphosphonates that you might be prescribed. Some are given orally (tablet form) and some are given intravenously as infusions (directly into veins through a drip).

This table shows some examples of bisphosphonates. This is not exhaustive and not all medications are suitable for everyone. (19)

Oral	Risedronate	Alendronate
Intravenous	Pamidronate	Zoledronic Acid

ARE THERE RISKS?

There are some risks with the use of bisphosphonates, especially in adults. (20) In adults, the risk of side effects increase overtime and often if bisphosphonate therapy is given, it is for a limited time (often 5 years).
Flu like symptoms are common 48 hours after beginning treatment, such as muscle aches, fever or an upset stomach, these symptoms do not typically recur.

Some of the less common and more serious side effects include;
Osteonecrosis of the jaw - this is when the cells in the jaw bone die, which can lead to problems with healing. The risk of this increases over time and the risk is higher for intravenous bisphosphonates than oral bisphosphonates
Atypical femoral fractures - the risk of this increases over time

COMPLEMENATRY AND ALTERNATIVE MEDICINE (CAM)

WHAT IS CAM?

Complementary and alternative medicine (CAM) falls outside mainstream medicine. Some CAM treatments are based on principles and an evidence base that are not recognised by the majority of independent scientists. (21)

EXAMPLES OF CAM?

These are just some CAM therapies with some evidence to support their suggested uses, this list is not exhaustive.

Acupuncture - may help with migraines and back pain (22)
Chiropractic - found to improve acute back pain and acute neck pain (23)
Aromatherapy - may improve sleep quality and help with stress (24)
Meditation - can improve chronic pain (25)

These are more CAM therapies without strong evidence to support their suggested uses, this list is not exhaustive.

Naturopathy	Homeopathy
Reiki	Herbal Medicines

CAN I TRY CAM?

Always speak to your doctor before changing your treatment plans, they will be able to advise you on which ones are best for you. They will also need to ensure any CAM treatments you want to try will not interact with existing prescriptions.

The NHS does not offer most CAM treatments.

WHERE CAN I FIND A CAM PRACTIONER?

All Chiropractors must register with;
Chiropractor - General Chiropractic Council https://www.gcc-uk.org/

The following have accreditation from Professional Standards Authority for Health and Social Care (PSA);
Acupuncture - British Acupuncture Council https://acupuncture.org.uk/
Aromatherapist - International Federation of Aromatherapists
https://ifaroma.org/en_GB/home
Complementary and Natural Healthcare Council
https://www.cnhc.org.uk/

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PHYSICAL THERAPIES

Physical therapies include physiotherapy, hydrotherapy and occupational therapy. These therapies are important to build muscle mass and bone mass. Both of which can allow for more mobility and can decrease the risk of fractures.

Therapies should be chosen based on the needs and abilities of each person. OI can vary greatly and so can the abilities.

Joint laxity and hypermobility are common in OI and may affect what physical therapies can be undertaken.

PHYSIOTHERAPY

Physiotherapists consider the body as a whole and do not just focus an injury or illness. They can offer advice on everyday activities or actions such as posture, pacing and lifting or moving things. They can recommend tailored exercises designed to build muscle and improve mobility. Physiotherapists can also give manual therapy to relieve stiffness, pain and encourage better movement.

Physiotherapy may also be used for rehabilitation after a fracture or surgery. For OI, it is beneficial as a way to offset the effects of immobilization such as decreased muscle mass, weakness and fear of movement. (26)

Physiotherapy referrals can be made by GPs and many health boards offer self-referral to physiotherapy services.

HYDROTHERAPY

Also known as water therapy, usually takes place in a heated pool. Hydrotherapy is similar to physiotherapy as it involves performing exercises under the supervision of a physiotherapist who will support and guide the session.

Hydrotherapy can build muscle mass and possibly allow for the person to learn to swim can allow for them to take up swimming on a more regular basis (not just in hydrotherapy pools), swimming is very low impact and is a very effective form of cardiovascular exercise. (27)

OCCUPATIONAL THERAPY

Occupational therapists can offer advice and guidance to allow people to live more independently. They are involved in the provision of wheelchairs and may be able to assist in referrals to wheelchair services. (28)

Occupational therapists often work in the community, you can contact your local social services or your GP to get a referral to an occupational therapist.

TEETH

There are a number of differences that may be seen in the teeth of people with OI. The teeth are likely to be smaller and the oral cavity is also likely to be smaller which may result in overcrowding of the teeth.

DENTINOGENESIS IMPERFECTA

Dentinogenesis imperfecta (DI) results in weak teeth which are at risk of breaking. There are different types of DI, the type associated with OI is type I (Syndrome-Associated).

DI is more common in OI types III and IV but can occur in any type. (29)

SYMPTOMS

There are a number of symptoms with DI, these are some of the symptoms but this list is not exhaustive. (30)

- Discoloured or translucent teeth
- Brittle teeth which wear and break more easily
- Small holes (pitting) in enamel
- Tooth loss
- Speech difficulties (due to teeth being in incorrect positions)

DIAGNOSIS

Usually by a dentist during a clinical exam and may be confirmed using a dental x-ray but this is not always necessary in people with OI as DI is common in people with OI.

TREATMENT

Treatment for teeth with DI would depend on the severity and treatment plans would be tailored to meet the individual's needs. (31)

Treatments could include;

- Crowns, or caps
- Fillings (including preventative fillings to strengthen the teeth)
- Dental implants (this will depend on whether the bone is strong enough to support the implant and may not be possible for some people with OI)
- Dentures

Have regular check-ups with a dentist are very important for DI.

HEARING

Hearing loss is common in people with OI.
It is estimated to affect up to 50% of people with OI but most cases reported are mild. (32)

WHY IS HEARING LOSS COMMON IN OI?

There are two types of hearing loss and both can occur in all different types of OI.

CONDUCTIVE HEARING LOSS

This type is the result of physical problems in the middle ear or externally. Some causes include, fracturing of the bones in the ear, infections or blockages. (33)

SENSORINEURAL HEARING LOSS

This type occurs when the nerves from the ear are no longer able to transmit signals to the brain. (34)

MIXED HEARING LOSS

A combination of conductive hearing loss and sensorineural hearing loss where there is damage to the outer and middle ear's ability to conduct sound and damage to the nerves.

WHEN DOES IT HAPPEN?

It can start at any age but it has been suggested that the risk of hearing loss reduces after the age of 40. (35)

Conductive hearing loss will often occur between ages 20 and 30 but can happen at any age.

WHAT TO DO?

Hearing tests should be sought if you are worried about your hearing.

WHAT IS THE TREATMENT?

Standard in-ear hearing aids would be used first. If these were not working for you or you had complications from using them, such as recurrent ear infections, bone anchored hearing aids or cochlear implants might be an option. (36)

A stapedectomy may also be considered. This involves the removal of part or all of the original stapes bone and replacing it with an artificial device. This procedure restores the transmission of sound waves to the inner ear for hearing.

HEART AND LUNGS

HEART

People with OI have a higher risk of developing cardiovascular (heart) disease than people without OI. (37)
These are some of the heart problems which have been associated with OI through research but this is not an exhaustive list. If you have concerns or a family history of heart disease, you should speak to your GP.

VALVE REGURGITATION

This is when the valves in the heart are weakened and begin to leak.
Some symptoms of valve regurgitation are dizziness, breathlessness, tiredness and chest pain. Valve regurgitation may be treated with medications or in serious cases, with valve repair or replacement surgery. (38)

ATRIAL FIBRILLIATION

Atrial fibrillation is when your heart has an irregular or abnormally fast heartbeat. It is not usually life threatening but can be uncomfortable and may require treatment. Some symptoms include dizziness, shortness of breath and tiredness. This increases the risk of blood clots which can cause heart attacks or strokes. People with atrial fibrillation would likely be given anti-coagulant medications to prevent blood clots.
It may be treated with medications to control the heart rhythm, catheter ablation to destroy the area causing the abnormal rhythm or cardioversion which gives a controlled shock to the heart to restore the normal heart rhythm. (39)

LUNGS

People with OI are more likely to have reduced lung function. (40)
COVID-19 : There was no reported issue for those with OI during the covid pandemic.
The following are some of the issues people with OI can experience, this list is not exhaustive. If you experience difficulty breathing or you are concerned, you should see your GP.

BRONCHIAL THICKENING

The walls of the airways become thicker which limits the airflow in the lungs, this also means mucus builds up in the lungs and can make chest infections more likely.
Symptoms include persistent cough and shortness of breath. Treatment may include exercises to clear the mucus, medications to improve airflow and antibiotics to treat infections. (41)

EMPHYSEMA

Emphysema is a type of chronic obstructive pulmonary disease (COPD). Symptoms include breathlessness, persistent cough or frequent chest infections. Diagnosis would be made using spirometry tests, chest x-rays or blood tests. You may be offered an inhaler or other medications, or lung rehabilitation. Surgery is only required in very serious cases. (42)

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SPINE

There are a number of common issues with the spine in people with OI. These are some of the more common complications and how they can be managed. However, this is not an exhaustive list and management of these issues will vary depending on OI severity and whether there are other health issues which have to be taken into consideration.

VERTEBRAL COMPRESSION FRACTURES

Fractures in the spine are common and the vertebrae can become compressed over time. (43) Compression fractures can cause significant pain, reduced mobility, decreased height, poor posture and weakness/ tingling sensation in the legs. They can be diagnosed with x-rays or CTs. Treatment includes pain medications, physical therapy or surgery to fuse the affected vertebrae in severe cases.

SCOLIOSIS

Curvature of the spine, often seen as an 'S' shape on an x-ray. This can cause back pain. If untreated, scoliosis can also compromise pulmonary function and affect balance.

Treatment will depend on the severity of the curve. Some treatments include the use of a back brace or surgical correction of the curve. Bracing or surgery may not always be possible in OI as the bones may not tolerate these treatments. Some may be offered steroid injections to control pain caused by scoliosis. (44)
Treatment will be individualised, and your consultant will be able to suggest the best treatments for you.

KYPHOSIS

Kyphosis is the abnormal curvature at the top of the spine. This can cause back pain, stiffness and tenderness in the spine.
Mild cases may not require treatment. Surgery may be used for severe cases; this would involve spinal fusion. This involves straightening the spine and joining together the vertebrae responsible for the curve. However, this may not be possible in OI. (45)

BASILAR INVAGINATION

An uncommon but serious complication of OI, where the vertebrae intrude into the skull and put pressure on the brain. Some of the symptoms include headache or pain in the back of the head, feeling dizzy or lightheaded, confusion, or tingling or numbness in hands or feet. You should seek advice from your GP and a referral to neurology if you are experiencing these symptoms.

Treatment could involve physical therapy, the use of a neck brace or surgery to stabilise the spine and prevent the spine from compressing the brain. (46)

PREGNANCY

Pregnancy is possible for people with OI, OI is not known to affect fertility. However, there are many risks associated with pregnancy in general. For those with OI there are some increased risks. (47) You will be seen by obstetrics who will help you to decide on a birth plan. You and the baby will be monitored closely.

NATURAL BIRTH VS CAESAREAN SECTION

Method of birth for women with OI will be dependent on the type of OI the mother has. It will also depend on whether the baby has OI. OI babies are more likely to present in the breech position which will make a caesarean more likely. More severe types of OI make a caesarean section more likely.

PRE AND POST NATAL COMPLICATIONS

These are some of the possible complications, this list is not exhaustive and not everyone will experience these complications.

FRACTURES

There is a risk of fractures during in pregnancy and during birth. (48)

GESTATIONAL DIABETES

Gestational diabetes is a form of diabetes which develops during pregnancy and usually disappears after birth. This is found to be more common in women with OI.

It can cause increased thirst, a dry mouth and tiredness. It may be controlled by changing diet and exercise or through medication or insulin injections. (49)

PRE-ECLAMPSIA

Pre-eclampsia causes high blood pressure, headaches, disturbs vision, vomiting, swelling of feet and ankles and can cause pain below the ribs. The cause is not known and it is more common in women with OI.

Pre-eclampsia can only be cured by delivering the baby. Once it is diagnosed you will be monitored closely and the baby will be delivered when it is safe. You may need to be induced. (50)

HAEMORRHAGE/ BLEEDING

There is a higher risk of bleeding during birth. This may result in the need for a blood transfusion. (51)

PREMATURE DELIVERY AND LOW BIRTH WEIGHT

Women with OI are more likely to deliver early, and the babies have a higher risk of having a low weight at birth. (52)

SPECIALISTS

OI CHARITIES

There are a number of charities worldwide who support people with OI. In the UK, the charity is the Brittle Bone Society (BBS). They support people with OI and their families. They offer lots of information, run events and share OI research studies.

CHILDREN'S SERVICES

There are four highly specialised services (HSS) for children with OI in the UK. These centres provide multi-disciplinary care where you may see a variety of specialists during your visit. They are also able to offer specialist advice and information to your local hospital team.

There are also a number of regional centres throughout the UK and Ireland.

Highly Specialised Services (HSS)

Birmingham Children's Hospital
Bristol Royal Hospital for Children
Great Ormond Street Hospital, London
Sheffield Children's Hospital

Regional Centres

Royal Manchester Children's Hospital
Royal Hospital for Children, Glasgow
Royal Hospital for Children and Young People, Edinburgh
Musgrave Park Hospital, Belfast
Temple Street Children's University Hospital, Dublin
Noah's Ark Childrens's Hospital for Wales, Cardiff

ADULT'S SERVICES

There are currently no specialist centres for adults with OI in the UK.
Many people with OI are seen by consultants who have a special interest in rare conditions such as OI. These may be consultants in Rheumatology, Metabolic Bone Diseases or Orthopaedics.

The Brittle Bone Society has close links with many centres and clinicians in the UK and lists the following centres on their website.
To be seen at any of these centres, you must be referred by your GP.

England

Addenbrooke's Hospital, Cambridge
Bristol Royal Infirmary
Manchester Royal Infirmary
Newcastle upon Tyne Hospitals
Northern General Hospital, Sheffield
Nuffield Orthopaedic Centre, Oxford
Royal National Orthopaedic Hospital, London
Queen Elizabeth Hospital Birmingham

Northern Ireland

Belfast Health and Social Care Trust

The Republic of Ireland

St Vincent's University Hospital, Dublin

Wales

University Hospital Llandough (UHL)

Scotland

Queen Elizabeth University Hospital, Glasgow
Western General Hospital, Edinburgh

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GLOSSARY

BREAK THROUGH THE JARGON

ANALGESICS - Medications used to control pain, more commonly known as pain killers

BISPHOSPHONATES - A type of medication used to increase bone mineral density

DENTINOGENESIS IMPERFECTA - Extremely brittle and weakened teeth

HYPERLORDOSIS - More prominent forward curvature of the lower spine

INTRAMEDULLARY NAIL / ROD - A rod/ nail which is placed in the medulla of the bone

KYPHOSIS - Front or backward curvature of the spine

KYPHOSCOLIOSIS - Combination of scoliosis and kyphosis where the spine curves in both sideways and front/backways directions

MEDULLA - Cavity in the centre of bones

NSAIDs - Non Steroidal Anti Inflammatory Drugs

OSTEOGENESIS IMPERFECTA - Brittle Bone Disease

OSTEOPENIA - Bone mineral density which is lower than normal

OSTEOPOROSIS - Very low bone mineral density

OSTEOTOMY / OSTEOTOMIES - A surgical cut made in the bone in order to correct a deformity

SCOLIOSIS - Sideways curvature of the spine

SPIROMETRY - test used to measure the lung function and to diagnose lung conditions

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This guide was produced at Swansea University as part of a PhD research project.

This guide was informed by research studies, NHS choices website and guidance from governing bodies such as NICE.