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### Paper:

Stark, Z., Lunke, S., Brett, G., Tan, N., Stapleton, R., Kumble, S., Yeung, A., Phelan, D., Chong, B., et. al. (2018).

Meeting the challenges of implementing rapid genomic testing in acute pediatric care. *GENETICS in MEDICINE*

<http://dx.doi.org/10.1038/gim.2018.37>

Data tables.

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**Table 4 Clinical and laboratory barriers to rapid genomic diagnosis identified in the course of the study, and solutions implemented**

<b>Barrier</b>	<b>Solutions implemented</b>	<b>CFIR construct</b>	<b>Impact/ consequence</b>
<b>Clinical</b>			
Delay in referral to clinical genetics	Communicate rWES availability and indications to referrers	Networks & Communications	Earlier referrals to clinical genetics
	Feedback diagnostic and clinical impact of rWES to referrers, e.g. at monthly NICU/Genetics case review meetings	Relative advantage	Increased number of referrals to clinical genetics
Delay in patient assessment by clinical genetics	Increased availability and flexibility of inpatient clinical genetic services to provide same-day consultation when possible	Implementation climate	Earlier WES initiation
Delay in rWES initiation	Feedback of clinical impact of rWES to clinical geneticist group	Relative advantage Reflecting and evaluating	Earlier and faster WES initiation
	Flexibility in patient approval mechanisms including electronic communication	Adaptability	
	Rapid turnaround of SNP microarray	Relative priority	Earlier and faster WES initiation

Parental difficulty processing complex information in a stressful environment	Increased genetic counseling support in the acute setting	Implementation climate	High rate of rWES acceptance by families
<b>Clinical-laboratory interaction (pre-sequencing)</b>			
Absence of an established rWES pathway	Clinical and laboratory champions identified	Formal implementation leaders	Formal rWES operating procedure established
	Mapping end-to-end clinical and laboratory processes	Planning	Formal rWES operating procedure established
	Specifying expected turnaround time for each step	Goals and feedback	Decreased time to result
	Definition of 'rapid team' (clinical and laboratory)	Reflecting and evaluating	Improved team communication and motivation
	Feedback of clinical impact of rWES to laboratory team	Networks and communications	Improved team communication and motivation
Samples not identified as 'rapid'	Centralized electronic team communication to track progress of each sample	Networks and communications	Standardized communication and sample tracking procedure

			established
	Sample labeling system modified	Networks and communications	Decreased time to result
Changes in patient clinical condition not communicated	Centralized electronic team communication used to update 'rapid team' on changes in patient status	Networks and communications	Timely communication of changes in clinical condition. Optimized use of limited resource
<b>Laboratory</b>			
Delays in sample transit between core facilities	Clear identification of rWES samples	Networks and communications	Reduction in time to report
	Centralized communication to track progress	Networks and communications	Reduction in time to report
Sample batching for library preparation	Dedicated staff for single sample library preparation	Available resources	Reduction in time to report
		Relative priority	Increased per sample cost Delay in processing 'standard' samples

Sample batching for sequencing	Dynamic use of HiSeq and NextSeq instruments to allow more flexible batching	Available resources	Reduction in time to report Reduction in time to report Increased per sample cost
Instrumentation breakdown	Additional instruments used to provide redundancy	Available resources	
Limited availability of computational resources	Implementation of prioritization system and dedicated computational infrastructure	Relative priority Available resources	
<b>Clinical-laboratory interaction (post-sequencing)</b>			
Result batching for weekly dedicated MDT meeting	Single case MDT, with minimum quorum defined	Adaptability	Reduction in time to report
	Electronic MDT when required	Adaptability	Reduction in time to report
Concern that time pressure may introduce error in data analysis	Increased input from senior laboratory and medical staff, redundancy in data analysis process	Formal implementation leaders	Reduction in time to report

CFIR, Consolidated Framework for Implementation Research; NICU, neonatal intensive-care unit; rWES, rapid singleton whole-exome sequencing.