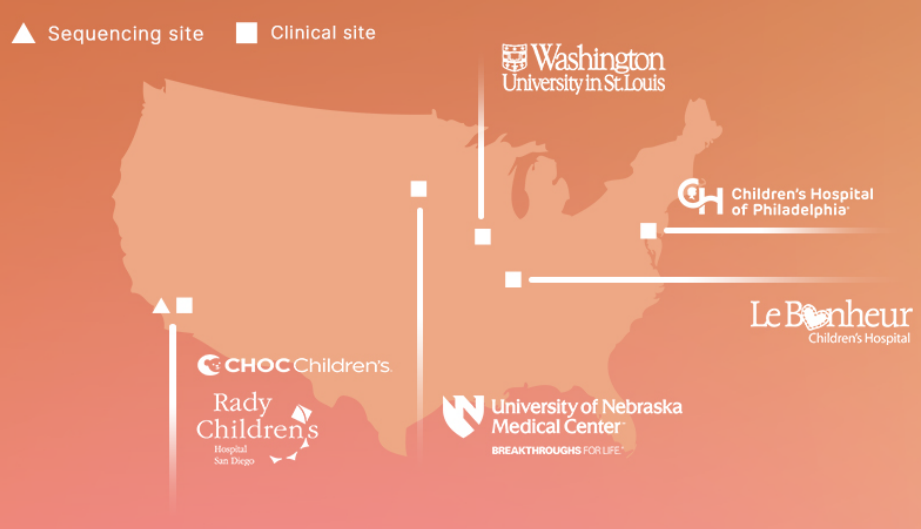
Investigation of the impact of whole-genome sequencing (WGS) on the clinical management of acutely ill newborns with suspected genetic disease



Study Objective

Evaluate the impact of WGS on the management of acutely ill newborns.

Prospective, time-delayed, randomized - control trial.

Multi-site with 5 participating children's hospitals across the US.

Population and Methodology

Primary Objective: Assess if whole-genome sequencing leads to changes in patient management.

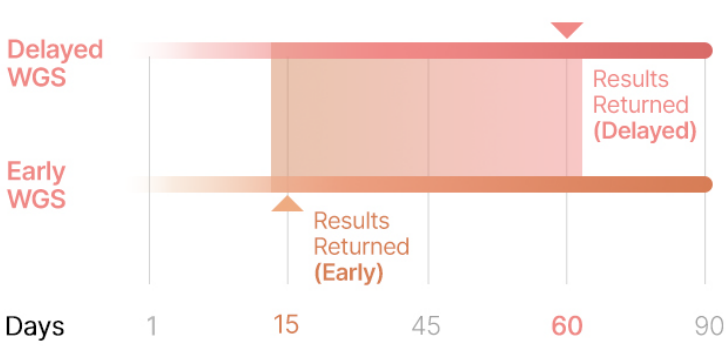
354 acutely ill newborns admitted to an intensive care unit with suspected genetic disease and aged between 0 and 120 days.

Delayed arm group

178 acutely ill newborns received delayed WGS.
Results returned 60 days after study enrollment.

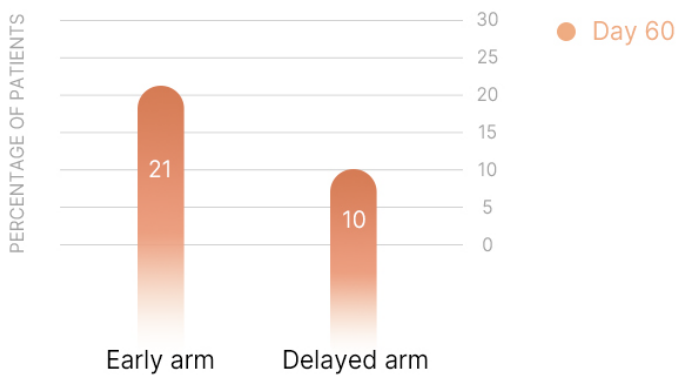
Early arm group

176 acutely ill newborns received early WGS.
Results returned 15 days after study enrollment.



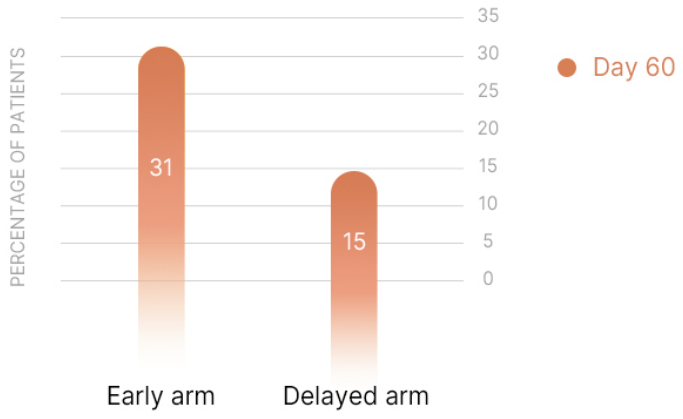
Change of Management

2x **Change of management** when having WGS compared to "usual care*" leading to a more precise care path.



Diagnostic Efficacy

2x **increase in diagnostic yield** when having WGS compared to "usual care."



* "Usual care" varied by site, and included a range of genetic tests including karyotype, chromosomal microarray, single gene testing, panels, biochemical analysis, exome sequencing and in a few cases, genome sequencing.

Types of Change of Management

Systematic Use of WGS

Using WGS as a first-line test in an acute care setting can lead to improved clinical management and higher diagnostic efficacy, and may reduce health care disparities.

NICUSeq Study Group. Effect of whole-genome sequencing on the clinical management of acutely ill infants with suspected genetic disease: a randomized clinical trial.
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