

Rapid FulGenome

Whole Genome Sequencing for the NICU/PICU

Overview

Rapid FulGenome is an advanced whole genome sequencing (WGS) solution designed to provide time-sensitive, comprehensive genomic insights for critically ill patients.

Clinical Use/Indications

Early diagnosis of genetic disorders in newborns and children can be critical for timely medical interventions. FulGenome's comprehensive analysis can aid in these diagnoses, enabling informed medical management.



With a preliminary report delivered in just 2 days, Rapid FulGenome offers unparalleled speed and accuracy, empowering clinicians to make informed decisions for urgent medical or surgical interventions.

Features



2 days - Preliminary Report
7 days - Final Report



Phenotype-driven,
multi-variant analysis



Singleton, Duo, or Trio analysis

Time-Sensitive Reports



Preliminary Report in 2 Days

A focused, early analysis highlighting pathogenic or likely pathogenic variants in genes with strong clinical relevance to the patient's presentation.

This rapid insight is designed to guide immediate medical management decisions while a more comprehensive review is underway.



Final Report within 7 days[†]

A complete, detailed genomic analysis that incorporates additional testing and confirmatory studies.

The final report:

- Includes pathogenic, likely pathogenic, and uncertain (VUS) variants
- Provides gene-level details with expert clinical interpretation
- Delivers results that are both actionable and comprehensive within the first week of testing

[†]When ordered, RISE RNA-seq results will be provided in an updated final report within 3-4 weeks.



Expanded Insights with RNA-Integrated Sequence Evaluation (RISE)

Scan QR code to learn more.

Facts About Rapid Fulgenome



Broad Testing Boosts Diagnoses

Testing infants whose condition isn't explained by trauma, infection, or prematurity results in more diagnoses.



First-Line Testing Improves Outcomes

Implementing rapid genome sequencing as a first-line test in NICU workflows is proven to:

- Change medical management for up to 87% of babies.¹
- Reduce healthcare costs up to \$15,786 per child.²



Clinical WGS Adoption Remains Low

Clinical rapid whole genome sequencing is performed in <5% of eligible patients.³

¹ PMID 39999847 - Analysis limited to DNA analysis of SNVs and CNVs; 2 PMID 34089648; 3 PMID 384136394

² PMID 34089648

³ PMID 38413639

Enhanced Variant Detection



Single nucleotide variants (SNVs)



Regions of homozygosity (ROH)



Copy number variants (CNVs)



Repeat expansions (20 genes covered) including:

ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS/ATXN8, BEAN1, C9orf72, CACNA1A, CNBP, DMPK, FGF14, FMR1, FXN, HTT, NOP56, PNRP, PPP2R2B, TBP



Genome-wide del/dups



Mitochondrial genome alterations

Sample Requirements

Proband Sample

1 x EDTA TUBE



Minimum 1 mL whole blood

*Sample receipt within 48 hours of sample collection

Duo/Trio Samples

1 x EDTA TUBE OR SALIVA SWAB



Minimum 1 mL whole blood (strongly preferred)



Saliva/buccal swabs will be accepted for family member testing (Duo/Trio); **however, they are not recommended** and will result in testing delays (~1 day).

Contact us to request a kit.