



FulGenome

Whole Genome Sequencing

Introduction

FulGenome is a next-generation whole genome sequencing (WGS) solution designed to provide comprehensive genomic insights through a single, streamlined report. Leveraging advanced sequencing technology, FulGenome delivers enhanced variant detection, including single nucleotide variants (SNVs), copy number variants (CNVs), genome-wide del/dups, mitochondrial genome alterations, and repeat expansions.

The optional addition of RNA analysis (RISE) enables deeper characterization of functional impact, providing a powerful tool for clinicians.

Who is FulGenome for?



Rare Disease Diagnostics

Offers a high diagnostic yield in patients with complex, undiagnosed conditions.



Pediatric Genetics

Early diagnosis of genetic disorders in newborns and children can be critical for timely medical interventions.



Broad Clinical Evaluation

Supports clinical decision-making across a range of specialties by offering a comprehensive view of the genome.

What Sets Us Apart

- **PCR-Free NGS Advantage** - Provides more uniform coverage and reduces amplification bias from traditional PCR-based library preparation, leading to higher accuracy in variant calling.
- **Exceptional Resolution** - Detects CNVs with >2 exon resolution and genome-wide del/dups, surpassing traditional exome sequencing.
- **Enhanced Diagnostic Yield** - Captures non-coding variants beyond +/- 20 intronic junctions, mitochondrial genome variants, and structural changes often missed in exome-based approaches.
- **Integrated RNA Analysis** - Provides functional insights into genetic variants, enabling deeper characterization of pathogenicity. Detects aberrant gene expression, mono-allelic gene expression, and aberrant splicing of expressed target genes.
- **Streamlined Reporting** - Unlike traditional testing requiring multiple separate analyses, FulGenome consolidates key insights into a single, comprehensive report.
- **Flexible Options** - Add on ACMG secondary findings, duo/trio analysis*, and/or complimentary PGx analysis as needed.

*Analysis limited to DNA analysis of SNVs and CNVs.

RNA-Integrated Sequence Evaluation (RISE)

SUPPLEMENTAL TESTING

RISE integrates whole exome or genome sequencing with RNA sequencing to enhance diagnostic accuracy for complex or undiagnosed cases. By evaluating the RNA-level effects of genetic variants, including those in coding, splice site, and regulatory regions, RISE maximizes clinical insights.

Benefits of RISE



Evidence-Based Yields

Up to 36% diagnostic yield in previously undiagnosed WES/WGS cases¹



Clearer Insights

Refines DNA variant classification to increase diagnostics rate



Richer Analysis

Detects gene expression impact of coding and noncoding variants

Single, Integrated Report

Fulgent's FulGenome report integrates DNA sequencing, genome-wide del/dups, mitochondrial genome sequencing, and RNA analysis.

Test Specifications

Turnaround Time 3-5 weeks

Sample Requirements




1 x EDTA TUBE

- 4 mL whole blood
- If submitting family member samples, please include them with the proband's sample to avoid delays. Family member samples and information **must be received within 3 weeks** of the proband's sample receipt to be included in the proband's analysis.

Contact us to request a kit.

Reference

¹ Curry, P.D.K., Broda, K.L. & Carroll, C.J. The Role of RNA-Sequencing as a New Genetic Diagnosis Tool. *Curr Genet Med Rep* 9, 13-21 (2021).



800-350-0537 | 800-454-1667
 info@fulgentgenetics.com
 www.fulgentgenetics.com

PATIENT
 Smith, Jane
 DOB: Jan 15, 1990
 Sex: F
 MRN: 12345
 Internal ID: F123456789

ACCESSION
 FG-000000
 Study ID: FG000000
 Subject ID: FG000000
 Collection Date: 01/15/2025
 Collection Site: FG000000
 Collection Date: 01/15/2025
 Collection Site: FG000000

PHYSICIAN
 John Doe, MD
 M.D. Name: John Doe, MD
 Client Name: John Doe, MD
 Phone: 555-555-5557
 Fax: 555-555-5557
 Report Date: 01/15/2025

LABORATORY
 Fulgent Genomics LLC
 CNA: 00000000
 CNA: 00000000
 Laboratory Director: John Doe, MD
 Report Date: 01/15/2025

FINAL REPORT


FulGenome

Whole Genome Sequencing (WGS) Analysis. Gene sequencing with deletion and duplication, genome-wide copy number, mitochondrial genome, and select repeat expansion analysis. RISE was performed.

CLINICAL INFORMATION

List of major phenotypes and other phenotypes noted in the provided medical and family history.

RESULTS



Positive

Test Interpretation: Multiple variants were identified which are consistent with a molecular diagnosis of a POLR1A-related condition. Clinical correlation of these results is recommended.

Clinically Significant Findings

Gene	Inheritance	Variant(s)	Zygosity	Classification
POLR1A	Autosomal Recessive	c.1052G>T (p.Arg351*)	Homozygous	Pathogenic
NM_000099.3				

Variants of Uncertain Significance

Gene	Inheritance	Variant(s)	Zygosity	Classification
GEN1	Autosomal Recessive	Deletion of Exons 1-20	Heterozygous	Unknown Significance
NM_000272.3				

RNA Analysis

This analysis included integrative analysis of DNA and RNA sequencing. The RNA analysis for the reported gene/variants above did not clarify the potential function of these variants.

Sample Report