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 cases[1]

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.

FINAL REPORT

FulGenome

CLINICAL INFORMATION

RESULTS

Positive

Clinically Significant Findings

Gene	Inheritance	Variant(s)	Zygosity	Classification
POLR3A NM_000059.3	Autosomal Recessive	c.1051C>T (p.Arg351*)	Homozygous	Pathogenic

Variants of Uncertain Significance

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

RNA Analysis

Sample Report

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).

4399 Santa Anita Ave, El Monte, CA 91731 | www.FulgentGenetics.com | info@fulgentgenetics.com | **P** +1 626.350.0537 | **F** +1 626.454.1667

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