Genomic Unity® Lightning Genome Analysis

We See More

Genomic Unity® Lightning Genome Analysis is a rapid diagnostic test for acutely ill patients designed to identify genetic variants that correlate with the patient's phenotype.

Test Description

Genomic Unity® Lightning Genome Analysis is a comprehensive test that uses a whole genome platform to detect all major clinically relevant variant types from a single sample. It analyzes patient DNA jointly with parental DNA when family samples are included.

Genomic Unity® Lightning Genome Analysis provides a single, unified clinical report that replaces a battery of tests including: whole exome sequencing (WES), chromosomal microarray (CMA), multiplex ligation dependent probe amplification (MLPA), and single gene or targeted gene panel testing, as well as PCR and southern blot tests for short tandem repeat expansions.

When to Order

Order this test for acutely ill patients in need of time-sensitive medical or surgical management, such as newborns or young children in intensive care units with a suspected genetic disorder. This test is most appropriate when clinical symptoms may be caused by a variety of genes or genetic mechanisms, are suggestive of multiple conditions, or are atypical for a certain condition.

Included Analyses

- Genome-wide sequence analysis including: single nucleotide variants, deletions, insertions, intronic, regulatory, and intergenic variants.
- Genome-wide structural variant analysis including: copy number variants (CNVs), duplications/deletions, regions of homozygosity (ROH), uniparental disomy (UPD)*, mobile element insertions, inversions, and aneuploidy.
- Mitochondrial genome analysis including: single nucleotide variants, deletions and insertions with heteroplasmy (≥5%), and large deletions.
- Short tandem repeat (STR) analysis.

Optionally includes:

- ACMG secondary findings.

** Available for trio only.*

Variantyx

Methods and Limitations

Genomic Unity® Lightning Genome Analysis uses a PCR-free whole genome sequencing (WGS) platform paired with our Genomic Intelligence® analytical software.

SNVs and insertions/deletions up to 50bp:

>99.9% sensitivity
>99.9% specificity
>99.8% positive predictive value

Structural variants:

96% clinical sensitivity

Short tandem repeats:

>99% clinical sensitivity

Sample Types

- Blood - 1ml

For trios:

- Parental blood - 5ml

Turnaround Time

A report will be issued within 5 days from receipt of required samples.

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