variantyx

Genomic Unity® Mitochondrial Genome Analysis

CP003

Overview

The diagnostic odyssey for unexplained genetic disorders is a frustrating and costly process for patients and their families. Unnecessary delays in identifying the molecular cause of the symptoms result in potentially missed opportunities for changes in treatment for the patient as well as missed screening opportunities for family members.

Genomic Unity® Mitochondrial Genome Analysis provides a full, phenotypically driven analysis of the mitochondrial genome.

Method

PCR free whole genome sequencing (WGS) is used as the underlying NGS technology. Its consistent read depth across the mitochondrial genome enables identification of multiple variant types from a single patient sample.

Proprietary algorithms optimized for each variant type are used to perform discrete in-silico analyses of the data which are brought together for collective interpretation, providing a complete genetic picture.

Rigorously trained variant scientists interpret all variant types in the context of the patient's phenotype and generate a unified clinical report.

Included analyses

- ✓ Mitochondrial genome analysis, including heteroplasmy (≥ 5%)
- ✓ Mitochondrial genome large deletion analysis

Test performance

Highly uniform sequencing depth

- 2000X mean mappable coverage
- >98% of nucleotides covered at ≥8x
- >99% of HGMD and ClinVar annotated variants covered at ≥8x

Highly sensitive detection of SNVs and indels up to 50 bp

- 99.1% sensitivity
- 99.2% positive predictive value

Highly sensitive detection of structural variants

- 96% clinical sensitivity
- In most cases, the exact genomic coordinates (the breakpoints) of the structural variant can be determined

Accepted sample types

- Blood - optimally 5ml
- gDNA - 5µg minimum
- Saliva

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