variantyx

Genomic Unity® Exome Plus Analysis

CP010

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® Exome Plus Analysis takes full advantage of the Genomic Unity® single platform method, providing a full, phenotypically driven analysis of all relevant genes and variant types.

Order directly, or reflex up from one of the available targeted analyses.

Method

PCR free whole genome sequencing (WGS) is used as the underlying NGS technology. Its consistent read depth across >98% of the 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.

Accepted sample types

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

Included analyses

- ✓ Exome analysis, including characterized intronic and regulatory variants
- ✓ Genome wide copy number changes, deletions, duplications, inversions, regions of homozygosity and mobile element insertions
- ✓ Mitochondrial genome analysis with heteroplasmy (≥5%)
- ✓ Short tandem repeat analyses
  - ✓ Adult-onset movement disorder (with or without cognitive involvement) STR analysis: *AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, C9ORF72, CACNA1A, CNBP, CSTB, DMPK, FMR1, FXN, NOP56, NOTCH2NLC, PABPN1, PPP2R2B, TBP*
  - ✓ Early-onset intellectual disability disorder STR analysis: *AFF2, DIP2B, FMR1*
  - ✓ Other disorders STR analysis: *PHOX2B, TCF4*

Test performance

Highly uniform sequencing depth

- 30X 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

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