Genomic Unity® Intellectual Disability Analysis

NR001

Overview

Genomic Unity® Intellectual Disability Analysis is an effective and innovative test for revealing the genetic cause of both syndromic and non-syndromic intellectual disabilities.

It is an effective test for X-linked disorders including Fragile X syndrome, Fragile XE syndrome and more. As well as autosomal causes including: ADNP syndrome, GRIN2B related neurodevelopmental disorder, Rett syndrome, Phelan-McDermid syndrome and more.

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.

Included analyses

- ✓ Constitutional Genome-Wide Copy Number Variant Analysis
- ✓ Early-onset intellectual disability disorder STR analysis: *AFF2, FMR1*
- ✓ Full sequencing and duplications/deletions analysis of intellectual disability associated genes: *ADNP, CHD2, FOXG1, FOXP1, FOXP2, GRIN2A, GRIN2B, NLGN3, NLGN4X, MECP2, SHANK3 and PTEN.*

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

- 99.7% sensitivity
- 99.6% positive predictive value

Highly sensitive detection of structural variants

- Higher than 99% clinical sensitivity

Accepted sample types

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

Turnaround time

6-8 weeks after sample receipt

Reflex

If the Genomic Unity® Intellectual Disability Analysis does not identify causal variants, the option is given to reflex up to Genomic Unity® Whole Genome Analysis which looks more broadly for causal variants across all genes.

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