



Genomic Unity[®] Testing

Technical specifications

Genomic Unity[®] testing is intended to help physicians diagnose the molecular cause of single gene as well as complex, oftentimes multifactorial, diseases in patients that present with unclear or heterogeneous phenotypes that cannot readily be linked to a clinical diagnosis. Genetic testing can be performed on the patient alone or as a trio where the patient's genomic variation is compared to his or her parents, enabling use of more powerful data analysis methods.

Accepted sample types

Genomic Unity[®] requires high quality DNA. Sample requirements are:

- Blood - optimally 5ml
- gDNA - 5µg minimum
- Saliva - contact us at info@variantyx.com for a kit

Test performance

Clinical-grade whole genome sequencing is conducted on an Illumina platform using the Illumina TruSeq DNA PCR-Free Library Preparation Kit.

Highly uniform sequencing depth

- 30X mean mappable coverage
- 98.6% of nucleotides covered at $\geq 8x$
- 99.7% of HGMD and ClinVar annotated variants covered at $\geq 8x$

Highly sensitive and specific detection of SNVs and indels up to 50 nucleotides

- 99.9% sensitivity, 100% specificity and 99.8% positive predictive value for SNVs
- 98.6%, 98.4% and 97.1% sensitivity for indels of 1-4, 5-15 and 16-50 nucleotides respectively

Detection of mitochondrial variants down to 5% heteroplasmy

Highly sensitive detection of structural variants

- 96% clinical sensitivity for structural variants
- In most cases, the exact genomic coordinates of the structural variant can be determined

Sensitive detection of tandem repeat allele counts for >20 known pathogenic loci