variantyx

Genomic Unity® Testing

A single method approach to comprehensive genetic testing

Revolutionizing genetic testing for rare disease patients

Traditional genetic tests detect only certain types of changes in an individual's DNA. **Sanger** and **NGS** methodologies are used for small sequence changes. **Southern blots** for detection of large short tandem repeat expansions (STRs) and large deletions. **PCR** and **capillary electrophoresis** for shorter STRs. **qPCR** and **MLPA** for deletions and duplications, and **arrayCGH/microarray**, **FISH** and **karyotype** for gross chromosomal deletions.

At Variantyx, we use a **single method** to detect all of these variant types from a **single sample**, providing our findings in a **single, unified clinical report**.

Variants detected and analyzed by Genomic Unity®

Using multiple different methods to detect different types of variants is a slow process that additionally leaves open the possibility of missing a diagnostic connection in a patient with a combination of changes.

For example, arrayCGH analysis performed by one laboratory may identify a heterozygous deletion. Subsequently, panel or exome testing by another laboratory may identify a heterozygous SNV.

A connection between these two results can easily be overlooked when the data is analyzed independently. It's only when the data is analyzed together the compound heterozygous relationship becomes clear.

SNVs	✓
Small indels (<50bp)	✓
Structural variants (≥50bp)	✓
- Deletions	✓
- Duplications	✓
- Insertions	✓
- Inversions	✓
Mitochondrial variants (≥5% heteroplasmy)	✓
Tandem repeat expansions	✓

The method behind Genomic Unity® testing

PCR free whole genome sequencing

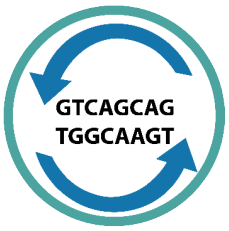
We use PCR free whole genome sequencing (WGS) as the underlying NGS technology. Its consistent read depth across >98% of the genome enables identification of multiple variant types from a single patient sample.

Proprietary algorithms

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

Expert variant interpretation

Our rigorously trained variant scientists interpret all variant types in the context of the patient's phenotype and generate a unified clinical report that is reviewed and signed by our board-certified clinical and molecular geneticists.



Ability to rerun analyses, without resequencing

Because the entire genome has been sequenced and analyses are performed in-silico, the data can be reanalyzed at any time to incorporate new findings from the scientific literature or to reinterpret variants in the context of newly acquired phenotypes.

Flexible ordering options

Start with a targeted analysis and reflex up, or opt for a full analysis from the start.

Genomic Unity® targeted testing options

Order one of the following tests (or from our larger online menu) for a targeted analysis with the option to reflex up to Genomic Unity® Whole Genome Analysis if a positive result is not identified:

- Genomic Unity® Epilepsy Analysis
- Genomic Unity® Intellectual Disability Analysis
- Genomic Unity® Motor Neuron Disorders Analysis
- Genomic Unity® Movement Disorders Analysis
- Genomic Unity® Comprehensive Ataxia Analysis
- Genomic Unity® Neuromuscular Disorders Analysis

For a complete list of available testing options, including a description of the genes and variant types targeted, visit our website:

www.variantyx.com/genomic-unity-analysis

Genomic Unity® Whole Genome Analysis

Order this test for a complete analysis of all variants detected by our algorithms, including:

- Exome sequence analysis, plus characterized intronic and regulatory variants
- Constitutional genome-wide structural variant analysis
- Mitochondrial genome analysis with heteroplasmy (≥5%), includes large deletions
- Early-onset intellectual disability disorder STR analysis: *AFF2, DIP2B, FMR1*
- 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, PP2R2B*
- Other STR analysis: *PHOX2B, TCF4*

Optionally includes:

- Huntington-related STR analysis (requires special consent): *JPH3, HTT*

Flexible, transparent billing

We offer patient pay, institutional billing and insurance billing options. For insurance billing cases, we perform all benefits investigation and prior authorization in-house.

There's a lot more to learn about Genomic Unity® testing

Visit our online resources to take a deeper dive into the data

www.variantyx.com/genomic-unity-resources

Performance statistics

How we detect structural variants

How we detect repeat expansions

Clinical validation study

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