variantyx

Genome methodology provides better testing outcomes for patients

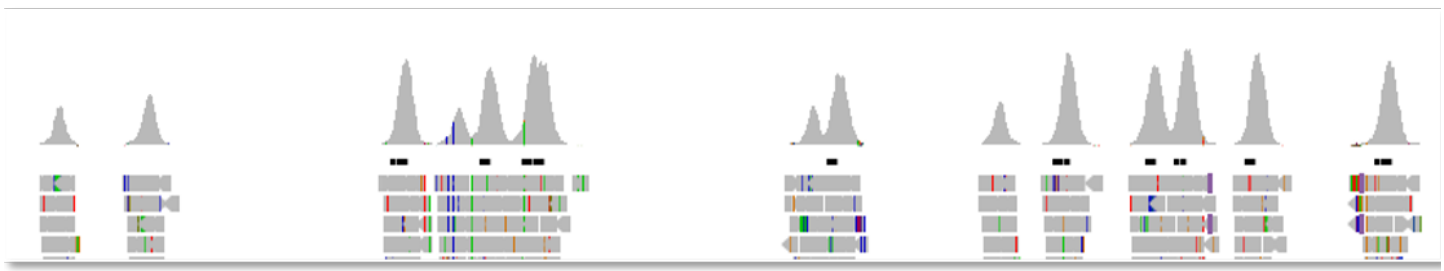
Genomes provide more uniform coverage

Panel, exome and genome tests begin with the same fragmented DNA.

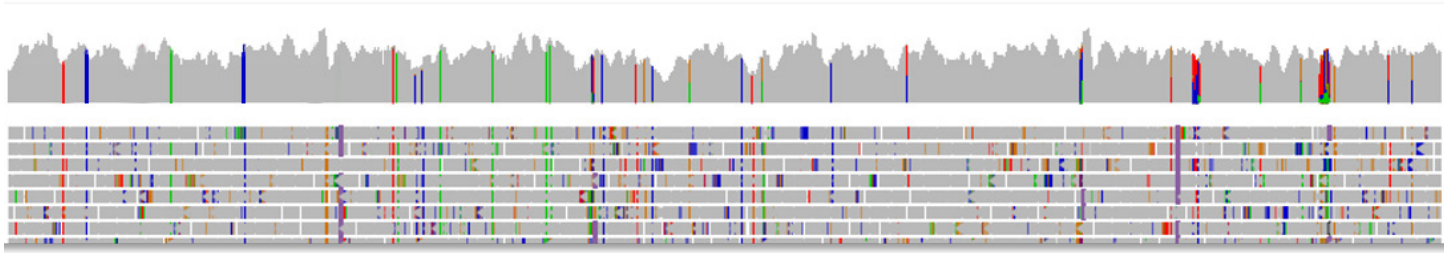
Panels and exomes mechanically isolate small sections of the DNA and then amplify the sections using PCR before sequencing. These steps both remove and skew the data. As a result, even with exomes, only 1-2% of your patient's DNA is covered - with uneven peaks and valleys and lots of holes.

Compare this to genomes which do not use PCR amplification and sequence the DNA directly. As a result, >98% of your patient's DNA is covered - evenly and with no holes.

Panel & Exome



Genome



Genomic Unity® specifications

Sequencing depth
- 30X mean mappable coverage
- 98.6% of nucleotides covered at ⩾8x
- 99.7% of HGMD and ClinVar variants covered at ⩾8x

SNV performance
- 99.9% sensitivity
- >99.9% specificity
- 99.8% PPV

Indel sensitivity
- 1-4bp: 98.6%
- 5-15bp: 98.4%
- 16-50bp: 97.1%

Structural variant performance
- Detects variants ⩾ 300bp
- >99.9% clinical sensitivity
- Exact coordinates determined in most cases

Mitochondrial variant performance
- Detection down to 5% heteroplasmy

Tandem repeat expansion performance
- Sensitive detection of >20 loci

Turnaround time
- 6-8 weeks

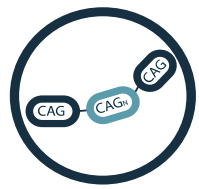
Accepted samples
- Blood
- Saliva
- gDNA

Genomes detect more variant types

It is the comprehensive and uniform coverage of genome data that enables identification and definitive reporting of all major types of phenotypically relevant genetic variants.

Small sequence changes

SNVs, Indels



Tandem repeat expansions

>20 loci covered

Structural variants

CNVs, inversions, aneuploidies, UPD, LOH, mobile element insertions

Mitochondrial variants

SNVs, Indels, microdeletions ⩾5% heteroplasmy

Genomes solve more cases

Detection of 1-2 exon deletions, tandem repeat expansions and non-exonic variants, among others, are a limitation of panel and exome tests. Genome testing has solved many such cases, including:

Case summary	Causal variant(s)
8 mos old with lissencephaly and developmental delay; Positive for *de novo* 1 exon deletion in *PAFAH1B1* for Miller-Dieker lissencephaly syndrome	
5 yr old with clinical symptoms of Lennox-Gastaut syndrome; Positive for *de novo* 2 exon deletion in *MECP2* for Rett syndrome	
5 yr old with seizures and developmental delay; Positive for *de novo* 2 exon deletion in *KIAA2022* for X-linked intellectual disability 98	
17 yr old with clinical symptoms of neuropathy; Positive for compound heterozygous SNV and repeat expansion in *FXN* for Friedreich's ataxia	
25 yr old with clinical symptoms of ataxia; Positive for compound heterozygous intronic SNV and 8 exon deletion in *POLR3A* for POLR3-related leukodystrophy	
44 yr old with clinical symptoms of dystonia; Positive for 2 repeat expansions in *ATXN8OS* for Spinocerebellar ataxia 8	

Variantyx Inc.
1671 Worcester Road, Suite 300
Framingham MA 01701

617 209 2090
info@variantyx.com
www.variantyx.com

Controlled document #: DCVXXCCN4001B
Updated 02-11-2021