Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

SYNGAP1 SNV explains development delays in 10-year-old female after series of 20+ genetic tests

Clinical presentation

A 10-year-old female presented with a history of developmental delays and intellectual disability. Additional symptoms included:

- Seizures
- Generalized hypotonia
- Hypercholesterolemia
- Sleep disturbance

Previous genetic testing

Extensive prior genetic testing was performed with negative results, including:

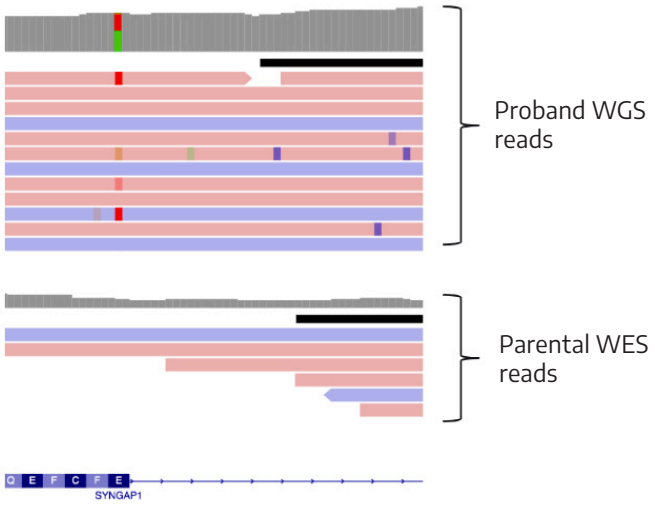
- CMA x2
- FMR1
- PWS/AS
- Mitochondrial genome
- Trio WES, reanalysis x2

Genomic Unity® Whole Genome Analysis was ordered because of its ability to identify all major variant types in a single test.

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a *de novo*, heterozygous, likely pathogenic single nucleotide variant in the *SYNGAP1* gene that results in a p.Glu221Val change.

Diagnosis: *SYNGAP1*-related disorders



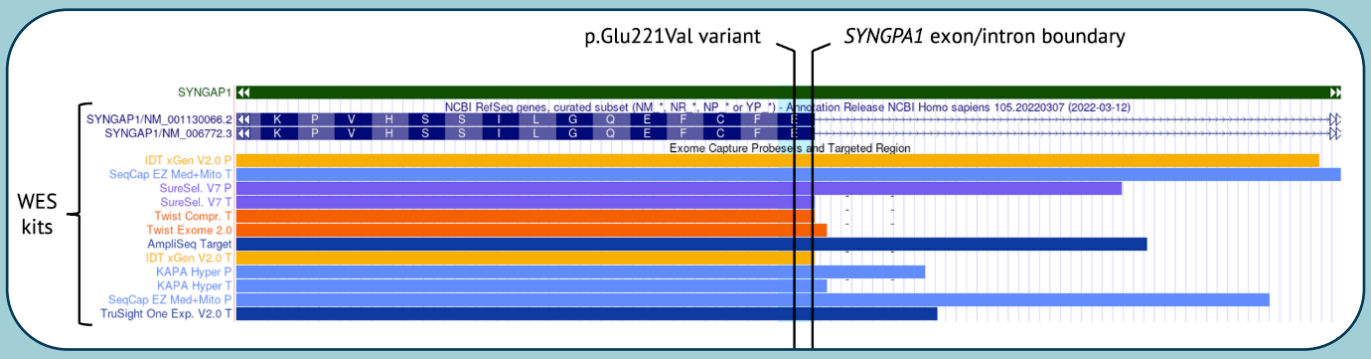
Uniform data from WGS clearly shows a heterozygous variant that falls within a region of poor WES coverage.

Variantyx

The Variantyx difference

Why was this variant detected by Genomic Unity® Whole Genome Analysis, and not detected by prior testing – including exome testing?

- ✓ Small sequence changes can't be detected by CMA.
- ✓ While small sequence changes are usually detectable by exome testing, this variant is located near an exon/intron boundary, in a region that is poorly covered by WES probes. This results in poor read coverage.



Variantyx genome sequencing does not use PCR amplification. Its uniform coverage depth across the entire gene makes it easy to identify this exonic single nucleotide change. Changes in other genes are simultaneously ruled out without the need for gene-specific tests like *FMR1* that extend the diagnostic odyssey.

Variantyx tests that would have identified this variant

Genomic Unity® 2.0 | Genomic Unity® Whole Genome Analysis | Genomic Unity® Lightning Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Exome Analysis | Genomic Unity® Epilepsy Analysis

Bring the power of whole genome sequencing to your practice today.

To get started, contact your Clinical Sales Specialist and visit us online at variantyx.com.

Variantyx

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