Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Compound heterozygous of mobile element insertion and SNV explains progressive dysphagia in 42-year-old male

Clinical presentation

A 42-year-old male presented with multiple symptoms including:

- Progressive dysphagia
- Memory impairment
- Ambulatory dysfunction
- Body twitches

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified likely compound heterozygous variants in the *VPS13A* gene: a likely pathogenic single nucleotide deletion that results in frameshift and a likely pathogenic AluY mobile element insertion (MEI) that disrupts exon 33.

Diagnosis: Chorea-acanthocytosis

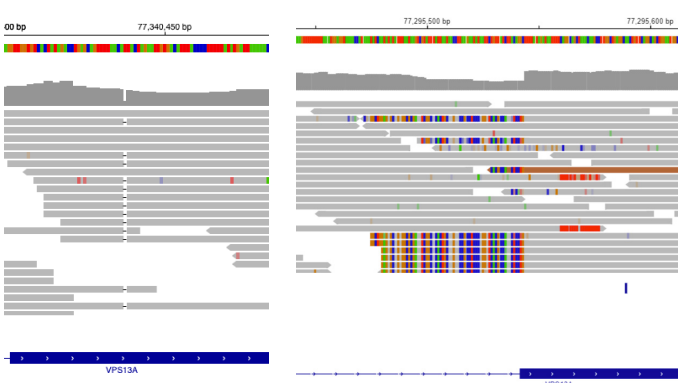
Previous genetic testing

A lysosomal storage disorders enzyme panel test was negative.

Additional targeted genetic tests were performed based on suspected clinical diagnoses including:

- *HTT* testing
- *MERFF* targeted variant testing

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



Uniform data from WGS clearly shows both the single nucleotide deletion (left) and the MEI breakpoint (right).

Variantyx

The Variantyx difference

Why were these likely compound heterozygous variants detected by Genomic Unity® Whole Genome Analysis, and not detected by other tests?

- The *VPS13A* gene was not included in the targeted gene and variant testing performed, and would be unlikely to be selected based on the clinical presentation.

  Variantyx genome analysis does not exclude any gene.

- Mobile element insertions (MEI) are undetectable by most available technologies - including gene sequencing, panel and exome tests.

  Variantyx genome analysis detects many types of structural variants including copy number variants, deletions/duplications, inversions, mobile element insertions, regions of homozygosity and aneuploidy.

Variantyx tests that would have identified this variant

Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Movement Disorders 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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