Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Compound heterozygous *FXN* variants explain progressive gait disturbance in 39-year-old female

Clinical presentation

A 39-year-old female with a clinical diagnosis of Friedreich ataxia of uncertain origin presented with multiple clinical symptoms including:

- Progressive gait and balance difficulty with juvenile onset
- Paraparesis
- Dysarthria
- Difficulty coordinating upper extremities
- Pain and paresthesia over distal lower extremities

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified compound heterozygous variants within the *FXN* gene: a pathogenic repeat expansion and a pathogenic 2772bp deletion.

The deletion partially spans the final exon removing coding plus 3' UTR sequence.

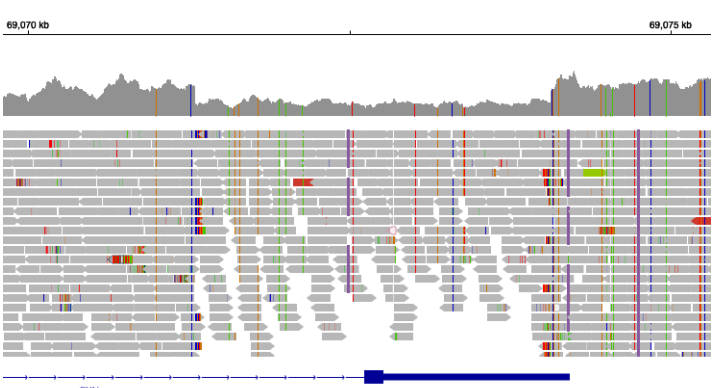
Diagnosis: Friedreich ataxia

Previous genetic testing

Prior *FXN* testing identified a heterozygous expansion of 866 repeats that did not fully explain the clinical presentation.

Follow on testing with comprehensive sequencing of ataxia genes was negative.

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 the deletion.

Variantyx

The Variantyx difference

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

✓ Repeat expansions and deletions can not be simultaneously detected by standard tests including gene sequencing, panels and exomes.

Variantyx genome analysis detects all major variant types in a single test including small sequence changes, structural variants, repeat expansions and mitochondrial variants.

✓ Exon level deletions are not detectable by gene sequencing tests and would be unlikely to be detected by panel or exome tests.

Variantyx genome analysis has a detection range from 1bp to whole chromosomal events.

✓ The deletion breakpoints fall within an intron and the 3' UTR, making the event undetectable by most available technologies - including panel and exome tests.

Variantyx genome analysis includes intronic and untranslated regions, enabling breakpoint detection regardless of location.

Variantyx tests that would have identified this variant

Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Movement Disorders Analysis | Genomic Unity® Comprehensive Ataxia Analysis | Genomic Unity® Ataxia Repeat Expansion 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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