Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Deep intronic variant explains progressive myopathy in 19-year-old male

Clinical presentation

A 19-year-old male presented with a history of slowly progressive myopathy with notable humeral and peroneal weakness:

- 7 years old - Myopathy first evident
- 11 years old - Muscle biopsy identified severe dystrophic changes suggestive of dystrophinopathy
- 15 years old - Wheelchair dependent

His brother shares a similar clinical presentation.

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a deep intronic, hemizygous, likely pathogenic variant in the *DMD* gene that causes retention of an intronic segment.

His symptomatic brother shares the same variant.

Diagnosis: Duchenne muscular dystrophy

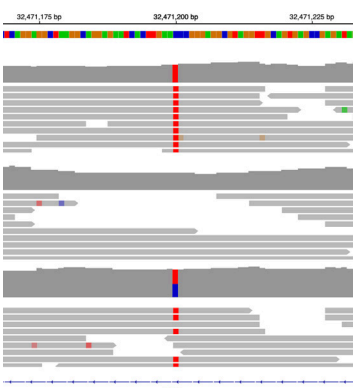
Genomic Unity® Whole Genome Analysis was ordered because of its ability to identify all major variant types in a single test.

Previous genetic testing

Multiple tests were performed including:

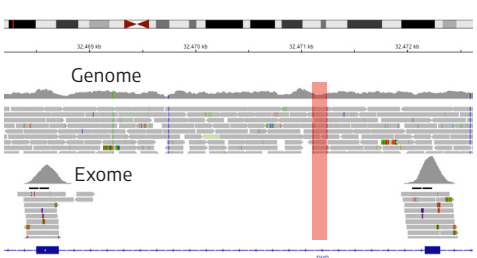
- Limb girdle panel
- SNP microarray
- Whole exome sequencing (WES) analysis

His brother separately had *DMD* and *FSHD* testing performed.



Uniform data from WGS clearly shows the deep intronic variant.

The variant is undetectable by exome testing due to coverage gaps.



The Variantyx difference

Why was this deep intronic variant detected by Genomic Unity® Whole Genome Analysis, and not detected by other tests?

✓ Single nucleotide changes are below the typical limit of detection (50-100 kb) of SNP microarray tests.

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

✓ The variant is deep within an intron, making it undetectable by most available technologies - including panel and exome tests.

Variantyx genome sequencing includes intronic regions, enabling variant detection regardless of location.

Variantyx tests that would have identified this variant

Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Exome Analysis | Genomic Unity® Neuromuscular Disorders Analysis | Genomic Unity® Muscular Dystrophy 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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