Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

DMD inversion provides genetic diagnosis, enabling access to treatment options for 16-year-old male

Clinical presentation

A 16-year-old male presented with a clinical diagnosis of Duchenne muscular dystrophy based on muscle biopsy results and hallmark symptoms, including:

- Motor developmental delays, large calves, toe walking, frequent falls, loss of independent ambulation at age 10
- Elevated CK levels
- Restrictive lung disease

He has been excluded from DMD trials and vamorolone treatment was not covered due to lack of a molecular diagnosis.

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a hemizygous pathogenic 713kb inversion encompassing exons 45-57 of the *DMD* gene. It is expected to result in loss of function which is consistent with prior RNA analysis identifying an intronal inclusion that leads to an inability to produce dystrophin.

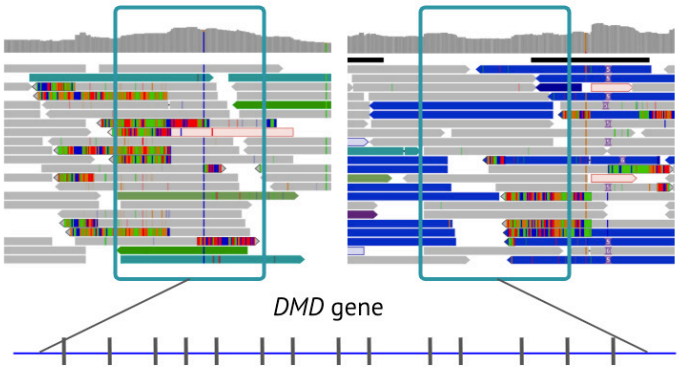
Diagnosis: Duchenne muscular dystrophy

Previous genetic testing

Multiple tests were performed including:

- *DMD* gene sequencing - x2
- *DMD* del/dup analysis - x2
- Dystroglycan-related congenital muscular dystrophy panel

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 identifies the breakpoints of the 713kb inversion spanning *DMD* exons 45-57.

Variantyx

The Variantyx difference

Why was this inversion detected by Genomic Unity® Whole Genome Analysis, and not detected by other targeted *DMD* tests?

✓ Balanced rearrangements like inversions are undetectable by most available technologies - including MLPA and targeted gene sequencing.

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

✓ Both inversion breakpoints are intronic, adding to the complexity of detection.

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

Variantyx tests that would have identified this variant

Genomic Unity® 2.0 | Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Exome Analysis | Genomic Unity® Neuromuscular Disorders Analysis | Genomic Unity® Muscular Dystrophy Analysis | Genomic Unity® DMD 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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