Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Inversion explains developmental delay and physical anomalies in 11-year-old male

Clinical presentation

An 11-year-old male suspected of having Angelman syndrome presented with a history of global developmental delay and multiple clinical symptoms including:

- Absent speech
- Autism spectrum disorder, behavioral abnormalities
- Dental anomalies
- Hypotonia
- Abnormal gait
- Astigmatism
- Megameatus

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a *de novo*, heterozygous, pathogenic 2.9Mb inversion of 2q31.3 to 2q33.1.

The inversion includes exons 5-11 of the *SATB2* gene and is predicted to result in loss-of-function of the protein.

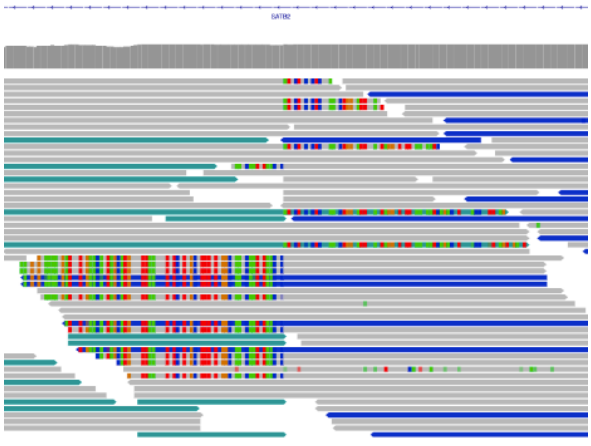
Diagnosis: Glass syndrome

Previous genetic testing

Multiple tests were performed including:

- Chromosomal microarray (CMA)
- *FMR1* testing
- *UBE3A* sequencing and methylation
- Whole exome sequencing (WES)

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 inversion breakpoint.

Variantyx

The Variantyx difference

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

Balanced rearrangements like inversions are undetectable by most available technologies - including the CMA and exome tests performed.

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

While inversions can be detected by karyotype, the size (2.9Mb) is below the limit of detection.

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

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® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Constitutional Genome-Wide Copy Number Variant Analysis | Genomic Unity® Genome-Wide CNV and FMR1 Analysis | Genomic Unity® Epilepsy Analysis | Genomic Unity® Intellectual Disability 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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