Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

SNV in perinatal lethal gene explains suspected Joubert syndrome diagnosis in 4-year-old male

Clinical presentation

A 4-year-old male presented with a suspected clinical diagnosis of Joubert syndrome. In addition to MRI results showing molar tooth sign and a history of neurodevelopmental (motor and speech) delay and autism, his symptoms included:

- Self injurious behavior
- Muscle hypotonia
- Dysphagia
- Hearing loss
- Constipation
- Coarse appearing face

Previous genetic testing

The patient has an extensive history of prior non-diagnostic genetic testing, including:

- Chromosomal microarray (CMA)
- Whole exome sequencing (WES) (2x)
- Mitochondrial sequencing
- Whole genome sequencing (WGS), reanalysis

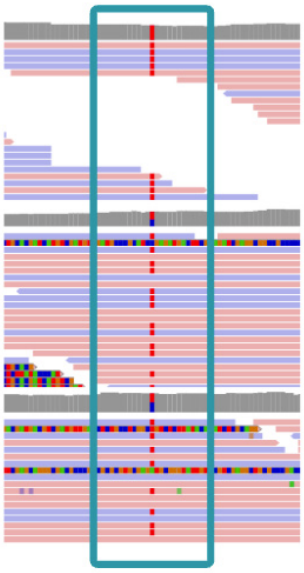
Genomic Unity® Whole Genome Analysis was ordered because of its ability to identify all relevant variant types in a single test

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a homozygous, pathogenic SNV in *HYLS1* shared with his symptomatic brother. The p.Arg61Ter change results in early termination of the 299 amino acid protein.

This case provides additional evidence that stop gain changes result in a milder, non-perinatal lethal phenotype compared to the typical p.Asp211Gly change.

Diagnosis: Joubert syndrome



Uniform data from PCR-free WGS clearly shows the inherited SNV.

Variantyx

The Variantyx difference

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

✓ CMA tests are unable to detect SNVs.

Variantyx genome analysis has a detection range from 1bp to whole chromosomal events, easily detecting this SNV.

✓ Exome and genome tests should have detected the variant. The region looks to be well covered by commercial exome probe kits and is not notable for repetitive sequences likely to interfere with amplification.

Variantyx variant scientists have interpretation capabilities that enable thorough investigation of every variant, oftentimes uncovering atypical presentations.

Variantyx tests that would have identified this variant

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