Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Compound heterozygous variants in an sncRNA explains multisystemic symptoms in 4-year-old male

Clinical presentation

A 4-year-old male presented with a complex medical history including:

- Mitochondrial complex I deficiency
- Global developmental delay
- Spasticity
- Congenital hypotonia
- Persistent left superior vena cava
- Mild oropharyngeal dysphagia

Results and interpretation

Variantyx Genomic Unity® Whole Genome Analysis identified a likely pathogenic, paternally inherited SNV and a pathogenic, maternally inherited indel, both within the *RNU7-1* gene.

Both variants have been experimentally demonstrated to alter U7 snRNP function.

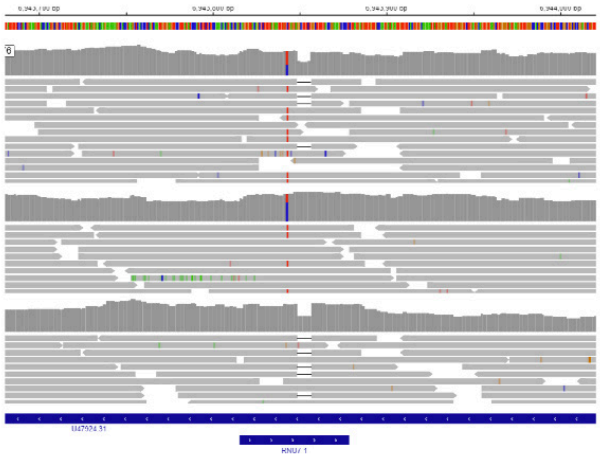
Diagnosis: Aicardi-Goutieres syndrome 9

Previous genetic testing

Multiple tests were performed including:

- Chromosomal microarray (CMA)
- SMN1/2 testing
- Cerebral palsy panel
- Mitochondrial sequencing & del/dup analysis identified non-diagnostic, heteroplasmic variants
- Whole exome sequencing (WES), reanalysis

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 inherited SNV and indel.

Variantyx

The Variantyx difference

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

- The variants fall within an sncRNA gene. Other tests focus predominantly, if not exclusively, on protein-coding genes.
- While exomes have the potential to include sncRNA genes, a survey of commercial exome probe sets shows that only 1 in 7 targets *RNU7-1*.

Variantyx genome analysis provides truly comprehensive mitochondrial and nuclear gene coverage, including analysis of sncRNAs. Ordering Variantyx genome analysis as a first line test would have shortened the diagnostic odyssey while significantly saving healthcare costs by eliminating multiple step-wise tests.

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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