variantyx

Case Study

10-year-old female with Developmental Delays, Intellectual Disability, & Epilepsy

Case background

A 10-year-old female who presented with a history of developmental and growth delays, intellectual disability, epilepsy, hypotonia, strabismus, and behavioral abnormalities (aggression, tantrums, skin picking).

The family history was negative for neurodevelopmental disorders and her birth was full term and uncomplicated.

Previous genetic testing

She had >20 tests in 10 years, including:

- 2x whole exome sequencing (WES) analysis
- Mitochondrial genome analysis
- 2x WES reanalyses
- 2x chromosomal microarrays
- Lumbar puncture for CSF analysis
- Mitochondrial/metabolic panel
- Fragile X syndrome
- Prader-Willi/Angelman syndrome
- Adrenoleukodystrophy (*ABCD1*) sequencing

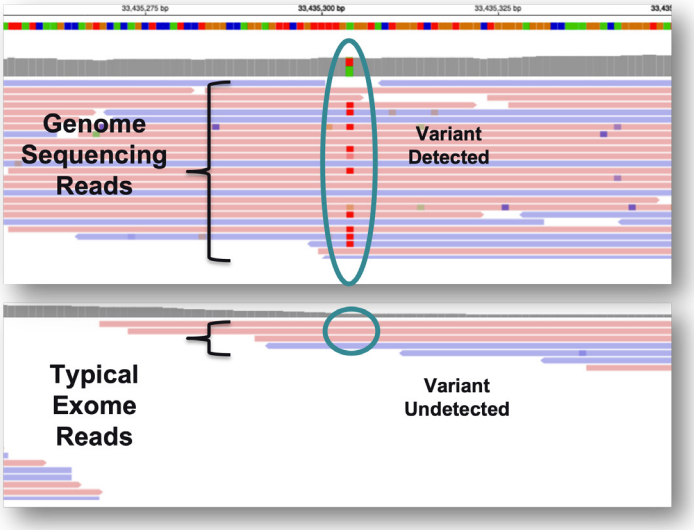
Interpretation and results

Diagnosis: *SYNGAP1*-related non-syndromic intellectual disability

Variantyx Genomic Unity® Whole Genome Analysis identified a *de novo,* likely pathogenic missense variant in the *SYNGAP1* gene (p.Glu221Val). Parental testing confirmed this variant was not inherited.

Multiple exome sequencing and panel tests could not detect this variant due to its location near an intron boundary, leading to poor coverage. Variantyx uses PCR-free whole genome sequencing without an enrichment step, allowing for clear data across ~98% of the genome that can identify all major variants, including this *SYNGAP1* variant.

Uniform data from WGS clearly shows the heterozygous variant.



Starting with a Genomic Unity® test could have:

- Improved time to diagnosis.
- Avoided invasive procedures – including a lumbar puncture.

Variantyx tests that would have identified this variant:

- Genomic Unity® Whole Genome Analysis
- Genomic Unity® Exome Plus Analysis
- Genomic Unity® Exome Analysis
- Genomic Unity® Epilepsy Analysis

See the impact of this result on the patient and her family here:

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