Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Trans splice variants explain suspected skeletal dysplasia in newborn female

Clinical presentation

A newborn female presented with congenital osteopetrosis.

Prenatal findings included severe IUGR suspected for skeletal dysplasia.

Postnatal findings included diffuse severe sclerosis consistent with osteopetrosis and bilateral 5th fingers clinodactyly.

Previous genetic testing

Multiple prenatal tests were performed:

- NIPS
- Chromosomal microarray
- Skeletal dysplasia panel, non-diagnostic

Followed by additional postnatal testing, including:

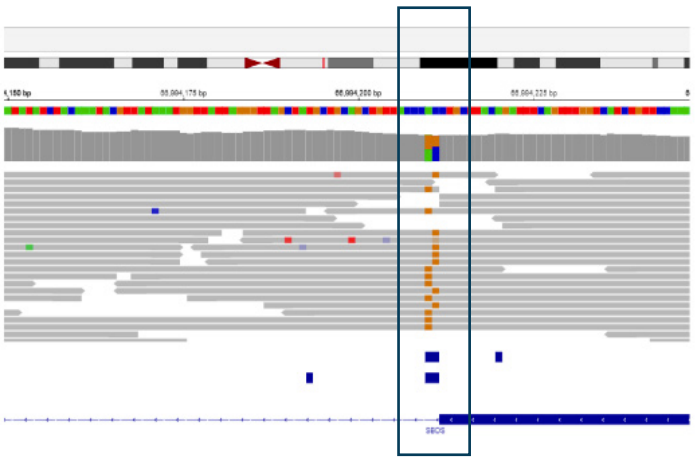
- Chromosomal microarray
- Expanded skeletal dysplasia panel, non-diagnostic

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

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified compound heterozygous, pathogenic splice variants in the *SBDS* gene. The findings provided guidance for clinical management and treatment including avoiding the need for an invasive liver biopsy, demonstrating the importance of a timely diagnosis.

Diagnosis: Shwachman-Diamond syndrome



Uniform data from WGS clearly shows the two splice variants.

Variantyx

The Variantyx difference

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

✓ The *SBDS* gene is hard to sequence and interpret due to pseudogene interference (*SBDSP1*).

Variantyx genome analysis uses PCR-free sequencing, avoiding issues with probe design and performance in low diversity and repetitive regions which can be an issue for both panel tests performed as well as exome tests.

✓ SNVs are not detectable by chromosomal microarray.

With a detection range from 1bp to whole chromosomal events, Variantyx genome sequencing is able to detect CNVs and small sequence changes within a single test.

Variantyx tests that would have identified this variant

Genomic Unity® Lightning Genome Analysis | 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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