IriSight™ Case Study

Transforming reproductive genetics through whole genome analysis

Compound heterozygous sequence variants explain hydrops fatalis in 2nd pregnancy loss

Clinical presentation

A couple presented with their second pregnancy with nonimmune hydrops, without other anomalies, in midgestation.

Hydrops was identified in the first pregnancy therefore nuchal translucency was tested in this pregnancy and found to be normal (1.7mm).

Previous genetic testing

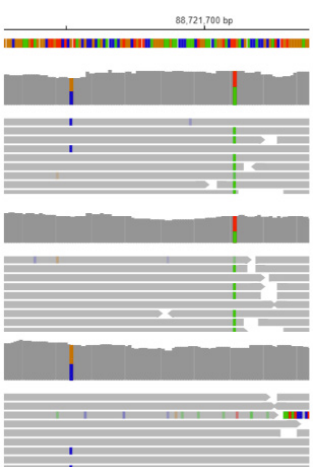
Multiple tests were performed during the first pregnancy including: Karyotype, Chromosomal microarray (CMA) and PWS/AS methylation.

NIPS was performed during the second pregnancy. CMA was subsequently performed on the product of conception and identified a duplication of uncertain significance with no associated disorder.

Results and interpretation

IriSight™ testing identified maternally and paternally inherited compound heterozygous, pathogenic variants in *PIEZO1*, both resulting in early termination of the protein.

Diagnosis: Autosomal recessive lymphatic malformation type 6



Uniform data from WGS clearly shows both variants and the pattern of inheritance.

IriSight™ for Pregnancy Loss was ordered because of its ability to identify all major variant types in a single test.

The Variantyx difference

Why were these compound heterozygous sequence variants detected by IriSight™ for Pregnancy Loss, and not detected by other tests?

✓ Single nucleotide variants (SNVs) are not detectable by karyotype, chromosomal microarray or NIPS technologies which represent the majority of POC offerings.

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

✓ POC samples are rarely accepted for targeted panel testing and exome testing is typically not made available until after aneuploidy has been ruled out, extending the diagnostic odyssey.

Variantyx genome sequencing provides comprehensive coverage of all major clinically-relevant variant types and all genes within a single test, eliminating the need for sequential, often inaccessible, follow on testing.

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

1671 Worcester Road, Ste 300, Framingham, MA 01701 | variantyx.com | info@variantyx.com

617-209-2090 | | VYX-678-0223