IriSight™ Case Study

Transforming reproductive genetic testing through whole genome analysis

Dual diagnosis explains phenotypes in male fetus

Clinical presentation

Multiple anomalies were noted during ultrasounds at 17 and 22 weeks, including cystic hygroma, hydrops fatalis, severe fetal growth restriction and polydactyly.

Previous genetic testing

A chromosomal microarray (CMA) test was negative.

IriSight™, a whole genome sequencing (WGS)-based amniocentesis test, was ordered because of its ability to identify all major variant types in a single test.

Results and interpretation

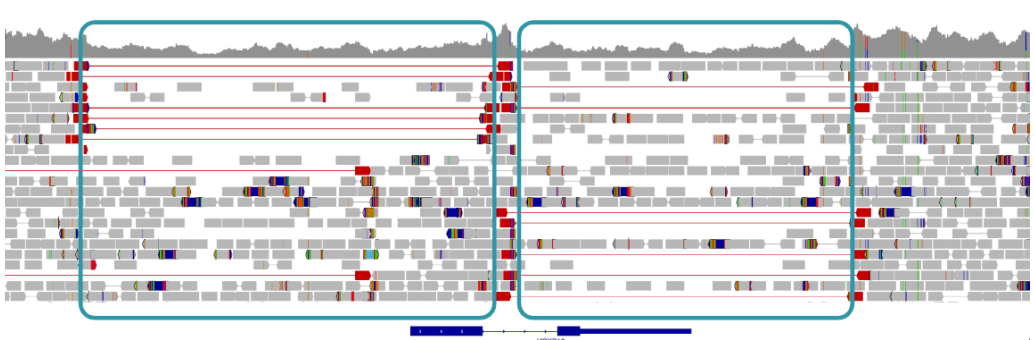
Variantyx **IriSight™ Comprehensive Analysis - Prenatal** identified pathogenic, compound heterozygous variants in *CHRND:* a maternally inherited single nucleotide deletion and a paternally inherited SNV.

Uniform data from WGS clearly shows both inherited variants: a single nucleotide deletion and SNV in *CHRND*.

Diagnosis: Congenital myasthenic syndrome

Variantyx **IriSight™ Comprehensive Analysis - Prenatal** also identified two heterozygous, paternally inherited, likely pathogenic single exon deletions in *HOXD13*.

Diagnosis: Autosomal dominant skeletal abnormalities of the hands and feet



Uniform data from WGS clearly shows the two, single exon deletions, both with breakpoints within the first intron of *HOXD13*.

Variantyx

The Variantyx difference

Why was this dual diagnosis uniquely possible with IriSight™ Comprehensive Analysis - Prenatal, and not possible with other tests?

- ✓ Both the single exon deletions and the small sequence changes are below the limit of detection for CMA. Both diagnoses are missed.
- ✓ If it had been ordered, exome testing could have detected the small sequence changes, potentially making the congenital myasthenic syndrome diagnosis. However, the AD skeletal abnormalities diagnosis would have been missed as single exon deletions are not detectable by exome testing.
- ✓ **Variantyx genome sequencing has a detection range from 1bp to whole chromosomal events. It is the only test that is capable of identifying all of the variants, making the dual diagnosis possible. In our experience, ~6% of prenatal cases result in a dual diagnosis.**

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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