

IriSight™ Case Study

Transforming reproductive genetics through whole genome analysis



De novo indel explains hypoplastic left heart in male fetus

Clinical presentation

Multiple anomalies were noted on a routine ultrasound including:

- Hypoplastic left heart
- Cystic hygroma
- Hydrops fatalis
- Echogenic kidneys and bowel

A typical work-up for hypoplastic left heart syndrome is long, with multiple possible genetic causes including aneuploidies, CNV disorders and single gene disorders.

Results and interpretation

VarianteX **IriSight™ Prenatal Analysis** identified a *de novo*, heterozygous, pathogenic single nucleotide deletion in *KMT2D*.

The nucleotide deletion likely results in a frame shift which causes truncation of the protein.

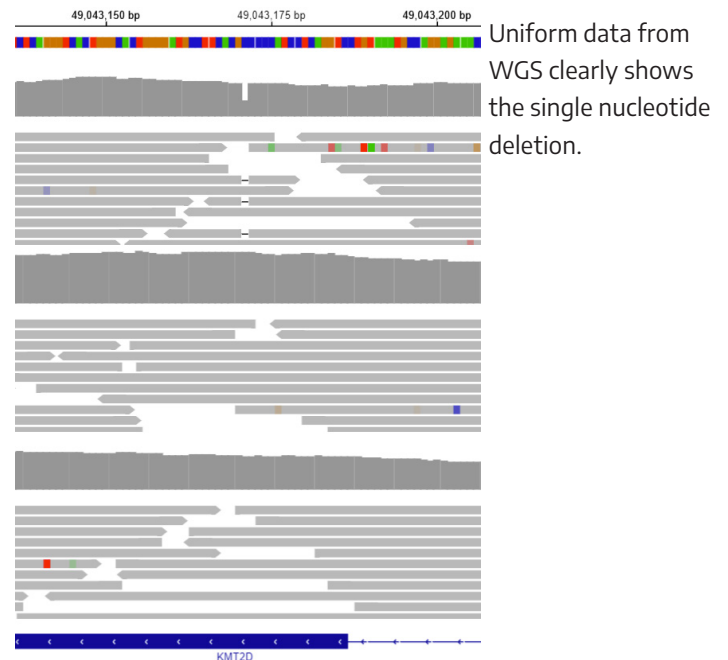
Diagnosis: Kabuki syndrome

Previous genetic testing

Multiple tests were performed including:

- Karyotype
- SNP microarray
- 30 gene NIPS panel

IriSight™ Prenatal Analysis was ordered because of its ability to identify all major variant types in a single test.



The Variantyx difference

Why was this single nucleotide deletion detected by IriSight™ Prenatal Analysis, and not detected by other tests?



Indels are below the limit of detection of karyotype (typically 4-5 Mb) and SNP microarray (typically 50-250 kb in clinically relevant regions) tests.

Variantyx genome sequencing has a detection range from 1bp to whole chromosomal events.



The *KMT2D* gene is not included in the 30 gene NIPS test that was performed, nor is it included in similar tests offered by other providers

Variantyx genome sequencing does not exclude any gene.

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](https://www.variantyx.com).