# IriSight™ Case Study

Transforming reproductive genetics through whole genome analysis

## Single exon deletion explains multiple congenital anomalies in male fetus

### Clinical presentation

Multiple anomalies were noted on a routine anatomy scan of a young primigravida including:

- Bilateral cleft lip and palate
- Micrognathia
- Hand clenching with overlapping fingers
- Distal arthrogryposis with toe syndactyly
- Ambiguous genitalia

Consanguinity was reported and subsequently confirmed.

### Results and interpretation

Variantyx **IriSight™ Prenatal Analysis** identified an inherited, homozygous, likely pathogenic 318 bp deletion spanning exon 6 of the *RIPK4* gene.

The deletion likely results in termination of the protein prior to the conserved ankyrin repeat domain.

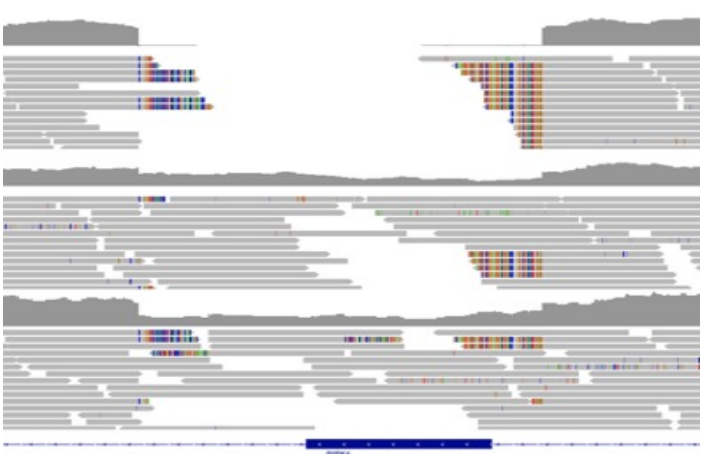
**Diagnosis:** Autosomal recessive Bartsocas-Papas syndrome

### Previous genetic testing

Multiple tests were performed including:

- Carrier screen via capture-based panel
- SNP microarray

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



Uniform data from WGS clearly shows the 318 bp deletion.

# The Variantyx difference

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

- The *RIPK4* gene is not included in the carrier screening test that was performed, nor is it included in similar tests offered by other providers.

  **Variantyx genome sequencing does not exclude any gene.**

- The size of the single exon deletion (318bp) is below the limit of detection of the SNP microarray performed (50kb), as well as similar tests offered by other providers - including exome tests.

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

- The deletion breakpoints are intronic, making the event undetectable by most available technologies - including panel and exome tests.

  **Variantyx genome sequencing includes intronic regions, enabling breakpoint detection regardless of location.**

**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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