Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Heteroplasmic mitochondrial deletion explains multiorgan dysfunction in 19-year-old female

Clinical presentation

A 19-year-old female presented with a history of short stature and failure to thrive throughout childhood, with multiorgan symptoms present since at least 14 years of age, including:

- Hearing loss
- Retinitis pigmentosa
- Kidney failure, resulting in renal transplant
- Type I diabetes
- Hypertrichosis

Results and interpretation

Variantyx **Genomic Unity® Whole Genome Analysis** identified a heteroplasmic (20%), pathogenic mitochondrial deletion ~2.3kb in size.

The deletion spans the *MT-ND5, MT-TH, MT-TL2* and *MT-TS2* genes.

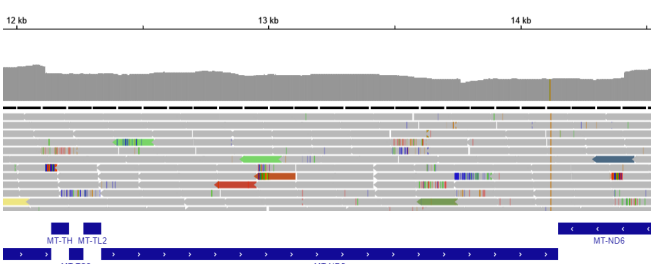
Diagnosis: Mitochondrial DNA deletion syndrome

Previous genetic testing

Multiple tests were performed including:

- Retinal dystrophy panel
- Mitochondrial genome sequencing (2x)
- Mitochondrial genome deletion analysis

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



Uniform data from WGS clearly shows the mitochondrial deletion

Variantyx

The Variantyx difference

Why was this heteroplasmic mitochondrial deletion detected by Genomic Unity® Whole Genome Analysis, and not detected by other tests?

✓ Mitochondrial genes are not typically included in retinal dystrophy panels.

Variantyx genome analysis does not exclude any gene, including mitochondrial genes.

✓ The relatively low level of heteroplasmy (20%) and relatively small size of the deletion (~2.3kb) fall near the lower end of the detection limits of the mitochondrial deletion test ordered.

Variantyx genome analysis includes >2000x coverage of the mitochondrial genome, with detection of heteroplasmy ≥5%

Variantyx tests that would have identified this variant

Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Mitochondrial Genome Analysis | Genomic Unity® Comprehensive Mitochondrial Disorders 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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