CENTOGENE
THE RARE DISEASE COMPANY

NGS Panel Genomic: the fastest way to diagnose a patient

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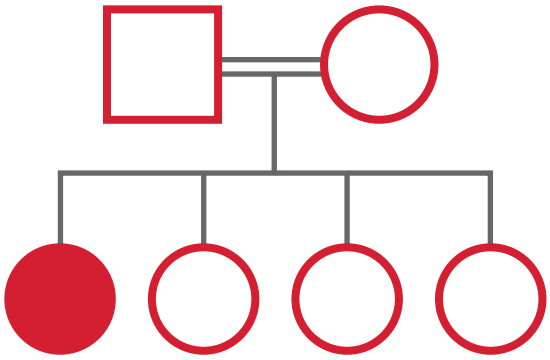
Georgraphic region

Saudi Arabia



Clinical information

3 years old female patient with seizures and hypopigmented spots, compensated septic shock, encephalopathy, and brain edema with descending transtentorial herniation. Her parents are first degree cousins and asymptomatic. They have 3 asymptomatic daughters. A paternal cousin of the index patient is affected by developmental delay and a second paternal cousin is affected by seizures and brain herniation.

Diagnostic journey

✗ **Whole exome sequencing**	Sequencing of all exons (~22,000 genes)
✗ **array-CGH**	Detects copy number variations (CNVs), chromosomal imbalances, regions exhibiting loss/absence of heterozygosity (LOH), uniparental isodisomy (UDP) and even low-level mosaicism.
✗ **Mitochondrial genome analysis**	Covers the entire mitochondrial genome along with 372 nuclear genes related to the mitochondrial diseases
✓ **Whole genome sequencing**	Sequencing of the whole genome: Detected a a heterozygous variant in the TSC2 gene, c.848+281C>T. **DIAGNOSIS: TSC2 - autosomal dominant Tuberous Sclerosis**

> 18 months; > 12,045 €

< 20 days

Powered by whole genome sequencing, **NGS Panel Genomic** avoids additional spending on step wise analysis like deletion/duplication, follow up exome or genome sequencing, mitochondrial analysis, or analysis of new genes associated with the phenotype when new information surfaces.

With NGS Panel Genomic you benefit from:
Highest diagnostic accuracy & utility • Fastest time to results • Significant price saving

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