NGS Panel Genomic: the fastest way to diagnose a patient

Prof. Peter Bauer, MD1 • Oliver Brandau, MD1 • Lia Abbasi Moheb, PhD1

1 CENTOGENE AG, Rostock, Germany

Geographic region

Saudi Arabia

Clinical information

Three 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 three 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 heterozygous variant in the TSC2 gene, c.848+281C>T.

DIAGNOSIS: TSC2 - autosomal dominant Tuberous Sclerosis

> 18 months; > 12,045 €

< 20 days with NGS Panel Genomic!

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