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

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 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