CENTOGENE
THE RARE DISEASE COMPANY

NGS Panel Genomic: the fastest way to diagnose a patient

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Georgraphic region

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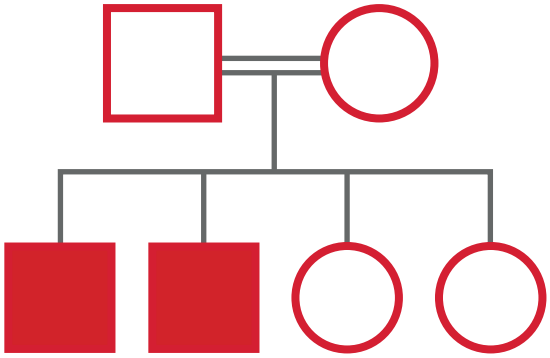
Clinical information

9 years old male patient with osteopenia, recurrent bone fractures, low bone density, abnormality of dental color, and gray sclera. Parents are healthy and consanguineous. They have another affected son with suspicion of osteogenesis imperfecta and additional symptoms, and 2 healthy children.



Diagnostic journey

✗ **Whole exome sequencing**

Sequencing of all exons (~22,000 genes)
A homozygous variant in the CREB3L1 gene, c.1259-13A>G, was identified in patient´s brother.

✓ **Whole genome sequencing**

Sequencing of the whole genome: Detected a homozygous large deletion within the TMEM38B gene, which is consistent with a genetic diagnosis of osteogenesis imperfecta type XIV.

DIAGNOSIS: Osteogenesis imperfecta type XIV – TMEM38B gene

> 7 months; > 5,350 €

< 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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