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Whole Exome Sequencing (WES)

Exome analysis is based on next-generation sequencing (NGS), and is used to identify variants in single genes that cause rare genetic diseases. This testing is the method of choice to detect the cause of disease in patients with complex, unspecific symptoms and undefined diagnoses, especially in rare autosomal-recessive disorders (consanguineous parents).

The sum of all coding regions in a genome is called the exome. In humans, this includes 23,000 genes with approximately 50 million bases. With Whole Exome Sequencing (WES), all exons (protein-coding regions) are analyzed. The genetic testing is therefore focused on the analysis of the 1-2% of the human genome where 85% of known pathogenic mutations are found.

Trio Exome Diagnostics is a powerful way to identify causative variants. The parents of the patient are also tested and an exome-wide segregation analysis is performed. This allows variants to be filtered and non-pathogenic variants to be eliminated.

Advantages of Exome Diagnostics:

- The number of variants detected using Exome Diagnostics is very high (> 200,000 per exome).
- Detection rates for Trio Exome Diagnostics are approximately 37%.
- Outstanding price/performance ratio considering the entire exome is screened and a large amount of information is gained. Turnaround times are reasonable.

The detected variants are analyzed and evaluated by an experienced team of scientists and geneticists and summarized in a comprehensive medical report.

Material:	5 ml EDTA-blood
Preanalytics:	Do not freeze
Method:	High-throughput sequencing is carried out by the Illumina HiSeq platform.
Other requirements:	Consent form, detailed description of symptoms
Turnaround time:	4-6 weeks
Literature:	Farwell KD et al. “Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions.“ Genet Med. 2015 Jul;17(7):578-86 Biesecker LG. "Exome sequencing makes medical genomics a reality". NatGen. 2010 Jan;42(1):13–14.

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