GENDIA

WHOLE EXOME SEQUENCING (WES) test

DESCRIPTION : The WES test sequences and analyses 214.405 exons dispersed throughout the genome.

INDICATIONS :

- Patients without molecular diagnosis who have exhausted currently available genetic testing
- Patients with a long differential diagnosis or genetically very heterogeneous disorder (eg deafness, retinitis pigmentosa, cardiomyopathy) that makes sequential testing cost-prohibitive

REQUIREMENTS :

- Informed consent if needed locally
- Clinical information about the patient and other affected family members
- Information on the parents : affected ? consanguineous ?
- A pedigree

SAMPLE : 30 ug of DNA or 10 ml EDTA blood of the proband and both parents sent at room temperature to GENDIA (Emiel Vloorsstraat 9, 2020 Antwerp, Belgium).

TURNAROUND TIME : The WES test takes < 2 months to complete from arrival of the sample in the lab to the result report.

METHODS :

- Approximately 37 Mb (214.405 exons) of the Consensus Coding Sequences (CCS) are enriched from fragmented genomic DNA by > 340.000 probes designed against the human genome (Nextera Rapid Capture Exome, Illumina)
- The generated library is sequenced on an Illumina NextSeq or HiSeq 4000 platform (Illumina).

GENDIA – *Genetic Diagnostic Network*
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- All exons and intron boundaries (+/-20 bp) are analysed.

- Relevant variants identified by NGS are Sanger sequenced to exclude NGS artefacts.
- All relevant variants are in-house analysed for their possible pathogenicity and clinical relevance.

TRIO ANALYSIS : It is advised to analyse the DNA from the affected father together with that of both parents (trio analysis). Comparing the proband's exomic data to that of the parents will significantly increase the sensitivity of the test as it will maximize the likelihood of identifying the disease-causing variant(s), and decrease the chance of obtaining variants of unknown significance (VUS).

LIMITATIONS : The following analysis is <u>not</u> performed :

- Analysis of introns
- Analysis of the mitochondrial genome
- Deletion-duplication testing
- Analysis of repeats (eg FMR1 repeat)

RESULTS : The results will be sent to you personally by email. If you want to have third parties to receive a copy, you can indicate this on the submission form below.

What will be in the report : Pathogenic variants, likely pathogenic variants and variants of unknown significance (VUS) in genes that are :

- Previously implicated in a human disorder similar to the affected individual
- Could be hypothesized to be related to the cause of the disorder due to their relationship to other genes or particular function

What will <u>not</u> be in the report ?

- Carriership of recessive variants
- Late-onset dominant variants not related to the proband's disorder (eg BRCA1)
- Benign and likely benign variants

PRIVACY and CONFIDENTIALITY: Your sample and the DNA extracted from it will only be used for the WES test. Your DNA will be stored at the performing test laboratory in Germany, unless you ask GENDIA to destroy it. The results from the WES test will be stored at GENDIA and the performing test laboratory in Germany, GENDIA will comply with the laws and regulations of Belgium and the EU regarding data privacy and the collection, use, processing and storage of patient information, and the performing test laboratory in Germany will comply with the laws and regulations of Germany regarding data privacy and the collection, use, processing and storage of patient information. The results of the WES test will only be reported to you personally, and never to third parties, including family members, insurance companies and physicians, unless you ask GENDIA to do so.

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GENETIC COUNSELING: A consultation with genetic counseling and discussion of the test results and the follow up steps is indicated certainly when a pathogenic gene variant was identified. Such consultation is available at GENDIA : you can make an appointment by emailing at info@gendia.net

PRICE : 3 possible test prices :

- 1500 Euro for a single patient
- **2500 Euro for trio analysis (patient and 2 parents) : ADVISED TEST**
- 2700 Euro for trio analysis of 3 affected patients in a family with autosomal dominant inheritance

PAYMENT : The payment is due within 2 months after the invoice date.

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