Prenatal WES

Veritas
a LetsGetChecked company

PrenatalWES is an innovative prenatal diagnostic test indicated for pregnancies with risk of presenting a genetic alteration

WHAT IS PrenatalWES?

- PrenatalWES is a **diagnostic service for pregnancies with fetal structural anomalies** such as cardiac malformations, short femur, increased nuchal translucency, or intrauterine growth restriction, after ruling out the presence of aneuploidies.

- The test is based on fetal whole exome sequencing enhanced for the **detection of CNVs** (Copy Number Variants), currently studied by microarrays **and pathogenic variants or SNVs** (Single nucleotide Variants) **in any gene that may be related to fetal anomalies**. The aim of the test is the detection of a possible genetic cause in at risk pregnancies **performing a single technique**.

WHY IS IT IMPORTANT?

- The standard procedure in prenatal diagnosis implies the performance of microarrays. In the absence of findings, it is necessary a second assay to sequence specific genes possibly related to the ultrasound findings. This implies a significant delay in diagnosis and eventual therapeutic interventions. **The advanced technology of PrenatalWES allows both assays to be performed in a single test, shortening turnaround time and maximizing the diagnostic yield.**

WHAT WILL YOU RECEIVE

» Genome-wide study of CNVs and SNVs in genes related to fetal malformations or family history.

» Comprehensive report with detailed information about the variants detected and their clinical implication.

» Genetic counseling to the specialist for the interpretation of the patient's results.

HOW TO START

The physician prescribes the test.

An amniotic fluid or chorionic villus biopsy sample is required.

In the laboratory, the presence of common fetal aneuploidies is ruled out within 24 hours, the whole exome is sequenced and analyzed for CNVs and SNVs.

The report is sent to the specialist, who will further discuss the results with the patient. Upon request of the physician, it is possible to refer the patient to Veritas for genetic counseling.

POSSIBLE OUTCOMES

Variants classified as pathogenic or probably pathogenic based on American College of Medical Genetics and Genomics (ACMG) guidelines (PMID: 25741868) are included in the report. Benign variants or Variants of Uncertain Significance (VUSs) are not reported.

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ADVANTAGES

COMPREHENSIVE GENOMIC ANALYSIS

With a single test the analysis of **CNVs and SNVs** is performed, maximizing the diagnostic yield.

REDUCED TURNAROUND TIME

Turnaround time is shortened as both analyses are performed with the fetal exome sequence data.

EXPERT PROFESSIONALS

Team with **extensive experience in prenatal diagnosis and genetic counseling.**

OTHER TESTS

In fetuses with a normal result it is possible to request after birth the extended neonatal screening test myNewborn.

INNOVATIVE TECHNOLOGY

Cutting-edge technology based on whole fetal exome sequencing.

TECHNICAL INFORMATION

- Whole exome sequencing with enhanced regions with 100x average coverage, sequencing more than 99% of the genes of interest at ≥20x
- In-house variant analysis tool
- The Veritas Intercontinental team includes internationally recognized experts with more than 20 years of experience in prenatal diagnosis and genetic counseling. Pioneers in the development and introduction in Europe of innovative prenatal tests into clinical routine

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