

WES

Whole
Exome
Sequencing

Pre-Marital Screening
Pre-Conception Screening
Donor Program Screening
Diagnostic Analysis

❖ Technology

WES uses Next Generation Sequencing (NGS) technology to get the information from millions of DNA fragments. WES analyses 180,000 exons, approx. 24,000 human protein-coding genes

Additional 7 Genes are analyzed using Sanger Sequencing; *FMRI, SMNI, F8, HBA, CYP21A2 (Congenital Adrenal Hyperplasia), CAH (same than CYP21A2 but analyzed by MLPA) and DMD (Duchenne muscular dystrophy)*

❖ How it works



1 Igenomix and Specialists provide Genetic Counseling to patients



2 Doctor prescribes the test for complex genetic disorder cases and family members (if needed)



3 Blood sample is taken from each patient.



4 DNA analysis by NGS and Sanger Sequencing.



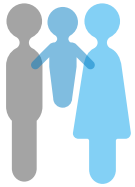
5 Clinically driven interpretation reports are shared with Doctors and Specialists. Igenomix provide Genetic Counseling if it is needed.

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❖ What is WES (Whole Exome Sequencing)?

This is a complete genetic analysis that looks for mutations-variants in the DNA sequence in over 24,000 genes

WES Screening



Used for screening Healthy Couples or Donor Programs

Strongly recommended for couples or donor program when planning to conceive naturally or through an assisted reproduction treatment. Identifies the risk of having a genetically affected baby

WES Diagnostic



Used to diagnose Affected Patients

Specially recommended in complex cases of new-borns or individuals, where clinical symptoms prevail for genetic mutations which need to be identified along with the family members

❖ WES Screening

Pre-Marital, Pre-Conception, Donor Program

Used for screening healthy couples or in donor program, analyses more than 2.600 Genes to report autosomal recessive and X-linked variants (*variants-genes-phenotypes clinically validated by HGMD, OMIM, DECIPHER and ClinVar*)

This test identifies the risk of transmitting recessive disorders to future generations

Although carriers are healthy people, if both parents have same variant in the same gene the probability of having an affected child is 25%. (PGT-M is recommended to avoid this probability)

Results
25
Days

"Individuals" and "Compatibility" results for couples to identify common pathogenic variants with a high risk to be inherited by future generations (as per ACMG and International guidelines)

❖ WES Diagnostic

Unmapped Genetic Disorders
Families with affected members

Specially indicated for families with a history of affected members (new-borns or individuals) with undiagnosed genetic disorder

This test sequences and analyses 180,000 exons, approx. 24,000 human protein-coding genes along with phenotype and family history driven interpretation

Results
6-7
Weeks

Clear results with identified variants following international best-practice guidelines of ACMG/CMSS (American College of Medical Genetics/Council of Medical Specialty Societies)





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