WES

Whole Exome Sequencing

A simple DNA test to help prevent and diagnose genetic disorders

The most advanced way to plan for a healthy family

Igenomix®
WITH SCIENCE ON YOUR SIDE

V.2020.01

WES Diagnostic

WES Diagnostic is a comprehensive genetic test that helps identify the disease-causing variant in an individual affected with a disease/condition.

The protein-coding region of DNA (exons) are sequenced. Since most of the disease-causing variants are present in the exon, WES is an efficient technique to determine disease-causing variants that may lead to a particular disease.

WES Diagnostic of the affected individual and their parents can help determine whether the disease-causing variant is inherited.

WES Diagnostic is also performed on the parents to identify and report common variants helping them to prevent any further genetic disorders.

Who benefits from this test?

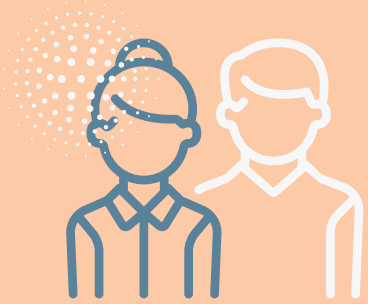
Indicated for individuals or families (new-borns, children, or adults) with undiagnosed genetic disorders.

WES Diagnostic is recommended for the following:

- **Affected individuals with suspected genetic diagnosis**
- **Help Diagnose affected patient with multiple differential diagnoses**
- **If targeted gene testing is negative**
- **Couples with family history of genetic disease**

WES Planning a healthy Family

WES Diagnostic is also an important genetic test that is recommended before planning a family. **This test helps determine whether a couple is at risk of having a child with a genetic disorder.** If parents have one or more variants in common, preventative measures can be taken in order to have a healthy child.



Carriers are usually healthy individuals. However, when both parents carry a variant in the same gene, they are at risk of having an affected child.

Who benefits from this test?

This test is strongly recommended for:

- **Consanguineous couples**
- **Any couple who would like to rule out the risk of having an affected child**

The test can be performed before attempting pregnancy either by natural means or assisted reproductive treatment.

Why to perform WES Diagnostic before planning a family?

Generally, parents realize they are carriers of genetic disorders after an affected child is born.

Genetic disorders can be prevented.

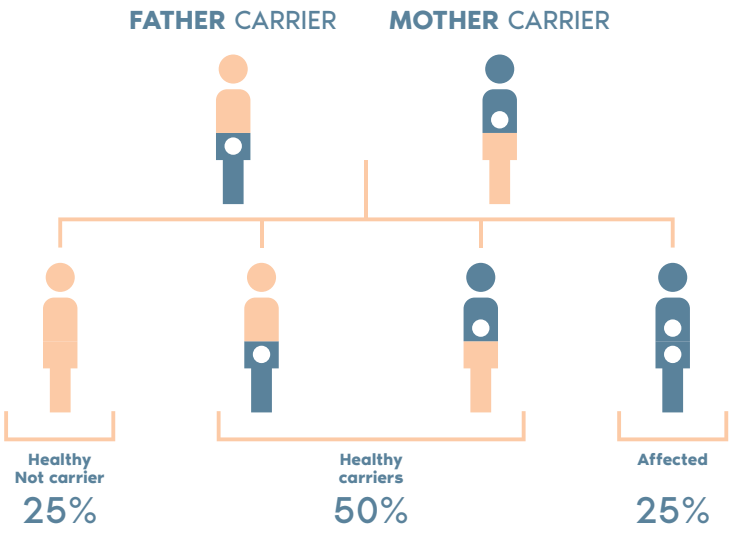
What happens if I'm a carrier?

NOTHING

Carriers of a variant usually do not present symptoms of the genetic condition.

Most individuals are carriers of genetic variants. Although carriers are healthy individuals, if the couple have a disease-causing variant in the same gene, the probability of having an affected child is 25%*.

*autosomal recessive

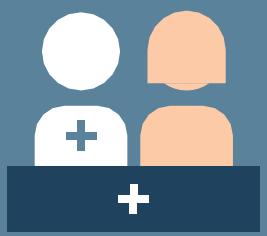


WES **testing options**

WES
Planning a healthy Family

Indications	Consanguineous couples, or healthy couples
Objective	Determine if a couple is at risk of having a child with a genetic disorder
Genes	24.000
WES **Couples**	Couple Report • Recessive, X-linked, Incidental findings • Pathogenic, likely pathogenic
TAT	25 working days
Sample	4-5 ml Blood

What if the couple is at risk?



Genetic counselling is recommended to discuss the possible procedures to conceive a healthy child.

Pre-implantation-Genetic Diagnosis can be performed to reduce the risk of having an affected child through assisted-reproductive treatments

Prenatal Diagnosis following spontaneous conception to determine if disease-causing variant is present in the developing fetus

WES

Family affected with a condition

Indications	Couples with family history of genetic diseases
Objective	Identify the genetic change that leads to the patient's condition/disease
Genes	24.000
WES **Couples**	Couple Report Diagnostic Report • Recessive, X-linked, Incidental finding • Pathogenic, likely pathogenic and VUS
TAT	35 working days
Sample	4-5 ml Blood

What disorders are included?

Whole Exome Sequencing (WES) through Next Generation technology allows the analysis of approximately 24,000 genes.
WES includes in the reports variants classified as pathogenic, likely pathogenic and VUS (in case there is an affected member with a condition). Recessive, X-linked and incidental finding are included in the reports.
In addition, all Couples are screened for 6 of the most common genetic conditions through alternative methods which includes: Congenital Adrenal Hyperplasia, Spinal Muscular Atrophy, Duchenne Muscular Dystrophy, Hemophilia A, alpha-thalassemia, and Fragile-X.
These tests cover the most common pathogenic variants associated with the aforementioned conditions.

If you suspect a clinical indication of one of these 6 conditions, please speak to our genetic counselor to better understand what test is most appropriate.

Speak with our Genetic Counsellor to review your case and understand how WES can help you
Phone: +971 4 5519465

SOME OF THE MOST COMMON MONOGENIC DISORDERS DETECTED IN THE MIDDLE EAST:
Beta-Thalassemia
Sickle Cell Anemia
Epidermolysis Bullosa
Propionic Acidemia
Congenital Adrenal Hyperplasia
G6PD Deficiency
Non-Syndromic Sensorineural Hearing Loss
Mediterranean Fever
Polycystic Kidney Disease
Cystic Fibrosis
Severe Combined Immunodeficiency Disorder
Stuve-Wiedemann Syndrome

(*)The American College of Medical Genetics (ACMG) and The American Congress of Obstetricians and Gynaecologists (ACOG).

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