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

Ind-01-20-V001

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 Screening can also be performed on the parents to help prevent any further genetic disorders.

Who benefits from WES Diagnostic?

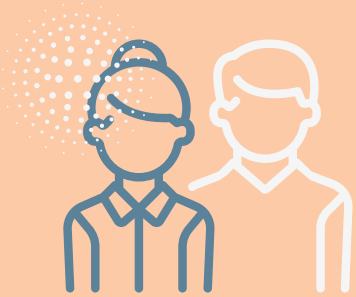
Indicated for individuals or families (new-borns, children, or adults) with undiagnosed genetic disorders.

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**
- **Determine the most effective treatment plan for the patient based on their genetic variant**

WES Screening

WES Screening is 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 WES Screening?

This test is strongly recommended for:

- **Couples with family history of genetic disease**
- **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 Screening?

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

Genetic disorders can be prevented.

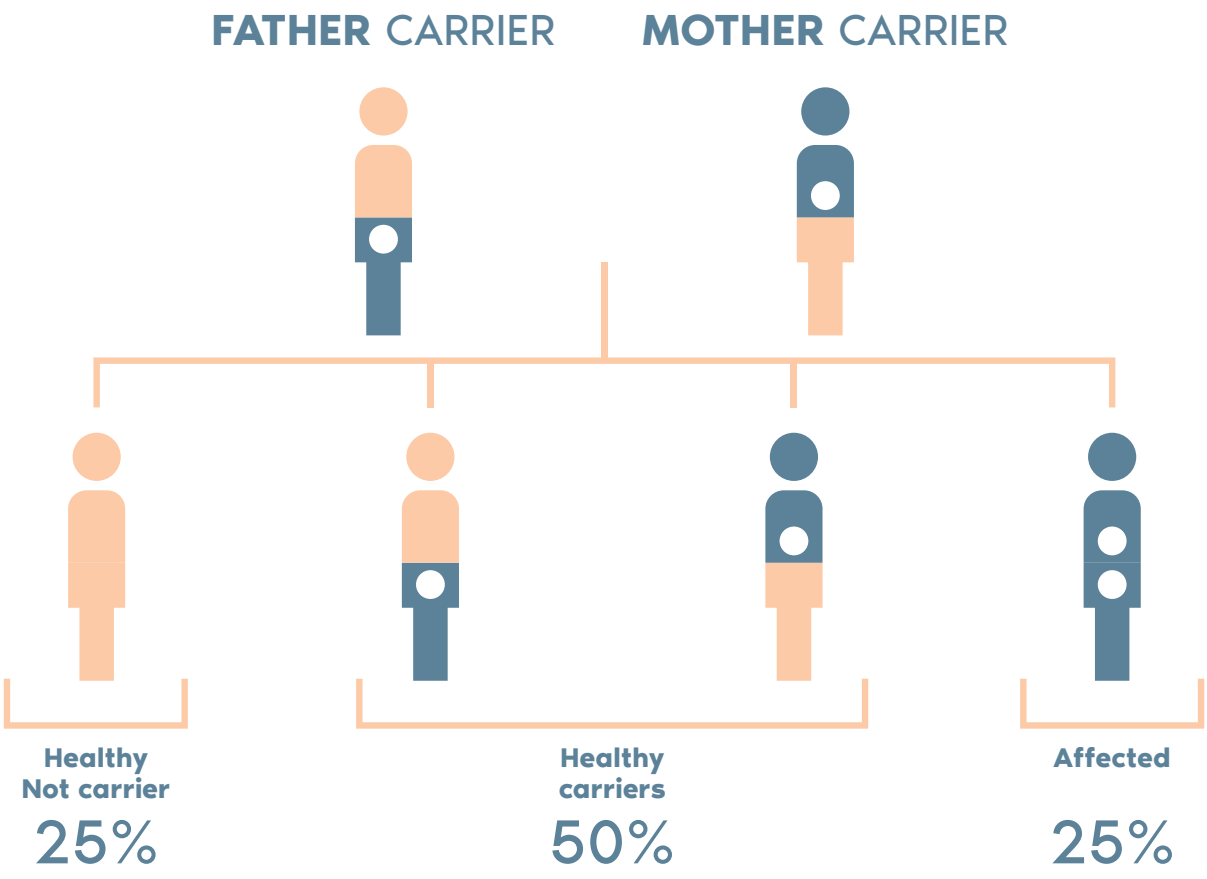
What happens if I'm a carrier?

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%*.

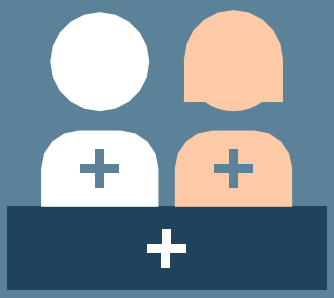
*autosomal recessive



WES **testing options**

	WES Screening
Indications	Couples with family history of genetic diseases, consanguineous couples, or healthy couples
Objective	Determine if a couple is at risk of having a child with a genetic disorder
Genes	2,500
WES **Couples**	Screening Couple Report
TAT	**30 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
Diagnostic

Indications

Affected individuals with an undiagnosed genetic disorders

Objective

Identify the genetic change that leads to the patient's condition/disease

Genes

24,000

Screening Couple Report

Diagnostic Affected Report

WES **Families**

TAT

6 to 7 weeks

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.

For WES Screening, a comprehensive gene list containing approximately 2500 genes linked to autosomal recessive disorders and around 250 genes that are associated with X-Linked conditions.

In addition, all couples are screened for 7 of the most common genetic conditions through alternative sequencing methods which includes: Congenital Adrenal Hyperplasia, Spinal Muscular Atrophy, Duchenne Muscular Dystrophy, Hemophilia A, alpha-thalassemia, and Fragile-X.

Speak with our Genetic Counsellor to review your case and understand how WES can help you
Phone: +91 011 66517833/34

SOME OF THE MOST COMMON MONOGENIC DISORDERS DETECTED:
Beta-Thalassemia
Duchane's Muscular Dystrophy
Sickle cell Anaemia
Propionic Acidemia
Spinal Muscular Atrophy
Cystic Gibrosis
Haemophilia
Fragile - X
Congenital Adrenal Hyperplasia
Polycystic Kidney Disease

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