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Whole Exome Sequencing Test Results

Pathogenic

Mutation known to cause the disease.

Likely Pathogenic

Alterations with strong evidence in favor of pathogenicity

Variant of Unknown significance

Analysis finds a genetic variant that may or may not be associated with a disease.

No mutation detected

No gene mutation was found (normal report)

Why Greenarray- Whole Exome Sequencing Test ?

- ✓ Offers High quality exome sequencing, highly sensitive and specific detection of mutations.
- ✓ Delivers the most accurate and comprehensive exome coverage.
- ✓ Advanced bioinformatics tools are used to improve overall accuracy and sensitivity for calling single nucleotide variants.
- ✓ Clear statement of any limitations that WES may have before and after testing.
- ✓ Reporting of the results in a comprehensive and easy-to-read way.

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Limitations of Whole Exome Sequencing

The following analysis is not performed.

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

References

- Advancing Personalized Medicine Through the Application of Whole Exome Sequencing and Big Data Analytics Pawel Suwinski1Front. Genet., 12 February 2019
- Exome sequencing: what clinicians need to know Sastre L 31 March 2014 Volume 2014:4 Pages 15—27 Dove Press medical journal
- Estimated rare diseases population in South Asian countries

About Greenarray

Greenarray is a molecular diagnostic laboratory. We offer diagnosis of infectious diseases, genetic testing and healthcare information to improve health and wellness. Our goal is to provide high quality affordable and accessible services.

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Whole Exome Sequencing

A comprehensive DNA test for effective prevention & diagnosis of genetic disorders

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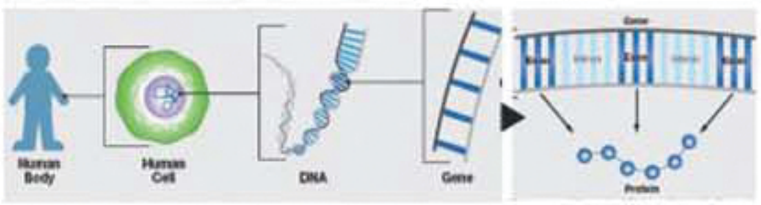
What is an exome ?

The body is made of millions of cells which all contain the same DNA (20 thousand genes) and they make proteins.



In the human genome, most genes are made up of portions called exons, separated by spacer regions called introns. Exons give rise to proteins, while introns have a mysterious spacer function. The set of exons of a genome is called exome.

What is Whole Exome Sequencing ?

Whole Exome sequencing is a comprehensive genetic test where protein – coding regions of DNA (exons) are sequenced that help to identify the variants responsible for causing disease in an individual affected with a disease/condition.

Who should go for Whole Exome Sequencing ?

Whole Exome Sequencing is the most advanced & accurate diagnostic genetic test. It can be performed during at any age and is generally recommended for the following :

Person suspected to have any genetic condition or having family history of gentic condition.

Having a complex medical history that may affect many organs and/or body systems.

Inconclusive previous genetic testing.

For prenatal & at birth analysis of baby's health.

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Test Sample Requirement

Blood
(3-5 ml in EDTA tube)

OR

Extracted DNA sample
(1µg high quality DNA)

How is Whole Exome Sequence testing done ?

Blood sample will be collected and sent to Greenarray labs.

DNA extraction will be performed from collected sample.

Extracted DNA will be subjected to Whole Exome Sequencing using NGS.

Analysis of variants and assessment of pathogenicity will be performed by clinical scientist.

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What are the benefits of Whole Exome Sequencing ?

Genetic testing can help :

- ✓ To make or confirm a diagnosis of a disease or a genetic condition.
- ✓ To make informed medical decisions about you or your family's current and future health.
- ✓ To decide on a personalized treatment and care plan in complex medical conditions.
- ✓ To make decisions about planning family for yourself or for other family members.
- ✓ To offer other family members the opportunity to learn their genetic status and if any genetic risk is involved.

Is Genetic Counseling necessary ?

It is recommended to have genetic counseling before and after genetic testing to learn the benefits and limitations of the test.