suma genomics
simplifying rare diseases

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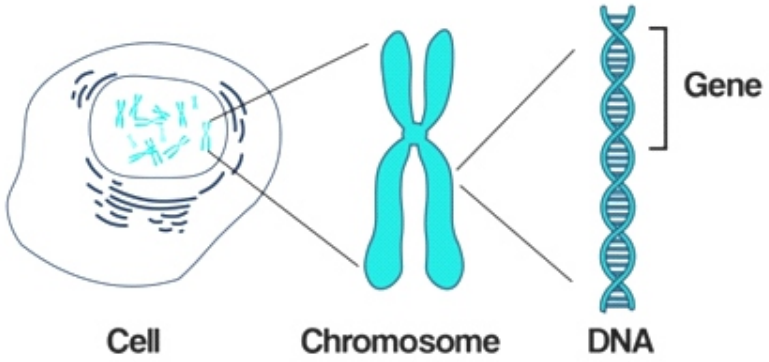
Whole Exome Sequencing - A Guide for Patients and Families

This guide has been prepared to briefly inform you and to clarify the queries that you may have on Exome Sequencing (ES) or Whole Exome Sequencing (WES).

If you are a clinician/geneticist, please check the WES guide for clinicians.

Our body is made up of trillions of cells and each of our cells has information stored and packed in the form of chromosomes. Each of these cells would have 46 chromosomes, 23 from each parent.



To simply put Chromosomes that are made of DNA are somewhat similar to books; the DNA consists of genes; genes are made up of 3 letter codons that are basically made by various combinations of 4 chemical components- A,T,G,C that could be compared to the pages from the respective book. People generally have two copies of most genes, one copy obtained from each parent.

The Exome, which is the protein-coding region of all Genes makes proteins that are essential for the structure and function of our body, and some of these genes also makes us the way we are- unique from others! However, Exome accounts for only 1 to 2% of our DNA. Now imagine some of the letters or pages from the book i.e genes go missing, or are changed; these are called mutations (genetic changes or gene variants). While some mutations do not affect us, some of them do cause minor to major changes in our health, our body, and bodily functions. Past research has provided sufficient evidence to indicate that most genetic conditions arise due to mutations in the exonic (protein coding) regions of the gene.

What is Whole Exome Sequencing (WES)?

Exome Sequencing or Whole Exome Sequencing is a test that looks for changes in approximately ~19,300 genes genes at once. Your clinician/ geneticist suggests you a WES because other tests have not helped diagnose the reason behind signs and symptoms that you/your child or family member is experiencing. The test may be useful for patients whose medical and family histories suggest a genetic cause for their signs and symptoms. WES helps clinicians/geneticists identify the clinical symptoms that may have resulted due to mutations in the exon region of the genes. Oftentimes your doctor may also request testing for your parents/other family members to check for carrier status.

Some laboratories offer to test only 5000-8000 genes which are known to be associated with human diseases (also called 'Mendeliome' or 'targeted exome' or 'focused exome' or incorrectly 'clinical exome'). Suma Genomics does not offer these tests.

What can you expect from the Whole Exome Sequencing (WES) test?

The expected results of this test include positive, negative, inconclusive/variant of uncertain significance and incidental findings.

- **Positive result:** It means the WES test identified a potential genetic change in the genetic material that could explain the clinical symptoms of you or your child or other members.
- **Negative result:** It means the WES test did not identify any clinically relevant changes in the genetic material that could explain the clinical symptoms of you or your child or other members. Your doctor may take decisions on further testing and course of action.
- **Inconclusive/variant of uncertain significance:** This means that the WES test identified a change in the genetic material and we have insufficient evidence to understand the effect of the genetic change and your clinical symptoms. Your clinician might review you or take further actions in this regard.
- **Incidental findings:** Sometimes we may identify the genetic change that could be medically actionable (example: a genetic change that could lead to heart disease or show a risk for a particular type of cancer) but unrelated to your current clinical symptoms. Your doctor will take your permission to reveal incidental findings before you take this test and inform us about your choice. You may opt-in /opt-out from incidental findings. Unless requested specifically, we do not offer these results to you. There might be additional charges for this request.

Why is Genetic counseling essential?

Pre-test counseling: A session of genetic counseling with your doctor/geneticist is highly recommended before you opt for the WES test. Discuss the following with your doctor before undergoing the WES test.

- The likely diagnostic yield of your clinical condition. Sometimes we may not be able to identify any genetic cause.
- Will the WES test result help in the clinical management of you or your child?
- The implications of the WES test results to you and your family members
- The accuracy and the error rate of Exome Sequencing (this depends on the clinical condition, and the gene involved)
- Will your insurance cover the test charges?

Post-test counseling: We do not send the reports to patients or their relatives directly as we believe post-test counseling is essential for genomic tests. The test results will be sent to your referral doctor via a secure email or hard copy (if required). Your referring doctor is solely responsible for all the decisions and possible management plans derived based on the test results.

When can I expect the results of the WES test?

We make our best efforts to provide results at the earliest and assure you a maximum turnaround time of 8 weeks, except in unforeseen or challenging situations.

What happens to the genomic data?

Suma Genomics would store the data in an anonymized form for its internal research purposes; your data is safe, and it would not be sold to a third party. Anonymized data relevant to public health may however be revealed to regulated/authorized bodies for the public good. With your consent, your doctor may ask for your data by reimbursing the expenses towards storage and transfer within a year of ordering the test.

Informed Consent

Informed consent is mandatory for all individuals undergoing Whole Exome Sequencing at Suma Genomics. We accept test request forms from doctors who declare that they have performed genetic counseling for their patients/family members and have obtained their written consent. We do not accept samples and direct requests from patients. For sample consent forms, please see our website.

For further queries and more information, please contact us at office@sumagenomics.in or visit our website-www.sumagenomics.in