ACTIA Inherited Genetics

MEDGENOME

Whole Exome Sequencing

Whole Exome Sequencing (WES), sequences the complete coding region of the genome. It is designed to examine all the coding regions and splice junctions of the genome. This method can be used to identify variations in the protein-coding region of any gene, rather than in only a select few genes. Because most known disease causing mutations occur in exons, whole exome sequencing can be used more effectively than whole genome sequencing.

When should you consider a Whole Exome test?

1. Can be considered for certain patients where the combination of symptoms does not allow an exact diagnosis/phenotype of suspected genetic disease.
2. Situations where delayed differential diagnosis may have a significant impact on the patient's quality of life.
3. In certain cases where a stepwise diagnostic strategy often substantially increases costs and time
4. Physician cannot provide any plausible diagnosis from the symptoms
5. Where there is no other alternate technique to confirm the diagnosis and to end the diagnostic odyssey

Who should undergo Whole Exome Test?

1. Patient with undiagnosed genetic disease (extensive evaluation and multiple genetic tests, without identifying the etiology)
2. Whole Exome Analysis can be used to identify variants inherited from the parents causing recessive disease or dominant disease. Additionally, de novo variants that occur in the offspring but are not present in either of the parents can also be detected

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WHY should you consider MedGenome's Whole Exome test?

Confronting the diagnostic challenge with whole exome sequencing is the best choice when you need a fast and cost-effective one-step solution to complete the diagnostic process of complex and unsolved cases.

1. Uniform coverage across exome region with a mean depth of >80-100X. More than 98% of targeted base pairs covered at ≥10x
2. All protein-coding regions along with the intron-exon boundary regions of ~20800 genes (including autosomal recessive, dominant and X-linked) and nuclear encoded mitochondrial genes
3. Comprehensive detection and analysis of both SNVs and CNVs. Sensitivity of detecting CNVs is 75-99% depending on the length and zygosity(het/homo) of the del/dup
4. Requisite quality control steps throughout the workflow from the laboratory sample processing till the interpretation ensures consistency, validity and accuracy of results
5. Report reviewed by Clinical Geneticist
6. Samples process at our College of American Pathologist (CAP) approved laboratory
7. Free Pre and Post Test Genetic Counselling

What are the test methodology?

Next Generation Sequencing (NGS)

Using genomic DNA extracted from blood, the coding regions of all the genes are captured and sequenced simultaneously by NGS technology on an Illumina platform. The sequence data that is generated is aligned and analyzed for sequence variants.

Test sample requirements

Blood
(3-5ml in EDTA tubes)

OR

Extracted DNA samples
(1µg high quality DNA)

Required forms

- Relevant clinical information including all the clinical presentations and symptoms
- Test request form

Turn around time {TAT)

- 21 Working Days for NGS

FREE GENETIC COUNSELLING

Reach Us

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