

IMMUNE



EAR



DENTAL



NEUROLOGY



LIVER



ENDOCRINOLOGY



REPRODUCTIVE



MUSCULAR
SKELETAL



METABOLIC



LUNG



HEMATOLOGY



SKIN

ADVAT EXOME PANEL

ADVAT
EXOME SEQUENCING

ANDERSON CLINICAL EXOME TEST

CLINICAL EXOME SALIENT FEATURES

- Clinical Exome is a custom focused exome curated in-house by experts at Anderson and covers 6161 genes encompassing both nuclear and mitochondrial genes.
- This exome provides better coverage of disease associated genes including coding variants, splice variants. It gives very high diagnostic utility at a low cost compared to whole exome sequencing
- Clinical Exome Panel is enriched for disease associated genes (with strong evidence) and genes with limited but emerging evidence from OMIM, Orphanet and other sources.

ADVAT CLINICAL EXOME PANEL

Enhanced coverage of disease associated genes with strong, moderate or supporting evidence in literature and databases

4,848

Genes from
OMIM
(Oct-2023
update)

34 Million

Records
submitted in
ClinVar (Oct
2023 update)

4,489

Genes from
Orphanet
(Nov-2022
update)

13,000

Phenotypes
from HPO
(Sep 2022
update)

ACMG

(73 genes;
Version 3;
incidental
findings)

Coverage of known pathogenic / likely pathogenic mutations

High confident pathogenic and likely
pathogenic variants from **ClinVar (2,18,000)**

In-house reported novel variants from
1000+ clinical reports

- Coverage of mitochondrial genome (37 genes)
- Phenotype based analysis, using ADVAT, a Anderson developed proprietary tool
- Clinical Exome has baits designed to cover multiple gene annotation sources and multiple transcripts facilitating highly sensitive and accurate variant calling
- The analytical sensitivity is >99% for SNPs and 93% for indels
- Repeatability, reproducibility, sensitivity and specificity were tested and were on par with global standards (99% for SNVs)

INDICATIONS FOR CLINICAL EXOME TEST?

- The clinical presentation or family history indicates the presence of an underlying genetic cause.
- Testing for diseases with genetic heterogeneity.
- Individual with an unidentified genetic disorder.
- To enable medical intervention and/or treatment.
- To validate the suspected genetic diagnosis.

ANDERSON WHOLE EXOME TEST

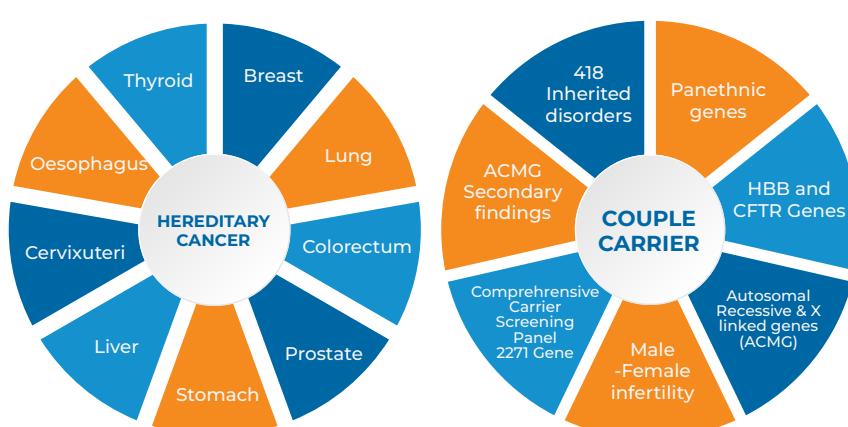
Whole Exome Sequencing (WES), sequences the complete coding region of the genome. It is designed to examine all the coding regions and splice junctions of the genome. This method can be used to identify variations in the protein-coding region of any gene, rather than in only a select few genes. Because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

WHOLE EXOME SALIENT FEATURES

- Uniform coverage across exome region with a mean depth of >80-100X.
More than 98% of targeted basepairs covered at $\geq 10x$
- All protein-coding regions along with the intron-exon boundary regions of ~24383 genes (including autosomal recessive, dominant and X-linked) and nuclear encoded mitochondrial genes
- Comprehensive detection and analysis of SNVs
- Requisite quality control steps throughout the workflow from the laboratory sample processing till the interpretation ensures consistency, validity and accuracy of results
- Report reviewed by Clinical Geneticist
- Pre and Post Test Genetic Counselling



Anderson Latest Technology in Genetic Testing helps to detect rare diseases and rare genetic variants



RARE DISEASES

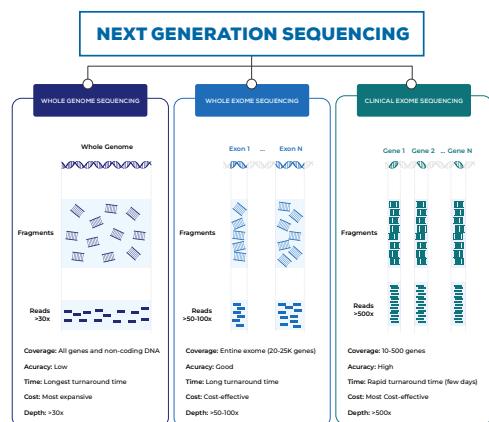
- Skeletal Dysplasia
- Batten Diseases
- Von Hippel - Lindau Syndrome
- Cone Rod Dystrophy
- Parkinson's Disease Seizures
- Developmental Delay
- Wilson Disease
- Epilepsy and other neuro diseases
- Acanthocytosis Chorea
- Alzheimer's Disease
- Batten Diseases
- Chronic Kidney Disease
- Cone Rod Dystrophy
- Cystic Fibrosis
- Hurler Syndrome
- Joubert Syndrome
- Lamellar Ichthyosis
- Mitochondrial Disorder
- Neurofibromatosis
- Prader Willi Syndrome
- Wolfram Syndrome

INDICATIONS FOR WHOLE EXOME TEST?

- May be contemplated for specific patients when the combination of symptoms does not permit a precise diagnosis or characterization of the suspected genetic condition.
- Circumstances in which a delayed differential diagnosis could substantially affect the patient's quality of life.
- In some instances, a staged diagnostic approach can significantly escalate both expenses and time.
- The physician is unable to offer any viable diagnosis based on the symptoms.
- When there is no alternative method available to confirm the diagnosis.

ANDERSON VARIANT ANALYSIS TOOL [ADVAT PIPELINE]

Anderson Latest Technology in Genetic Testing helps to detect rare diseases and rare genetic variants



EXOME SALIENT FEATURES

- Coverage of alternate gene transcripts (MANE,GENCODE, REFSEQ).
- Coverage of Non-Coding Pathogenic variants(HGMD/ClinVar).
- 100% Coverage of Mitochondrial genome.
- Indian Database to identify rare genetic variants.
- Reanalysis after years can be done without any charges.
- Hg 38 assemble exome sequencing.
- Anderson Variant Analysis Tool (ADVAT) pipeline is an automated tool for Single, Duo and Trio analysis.

ADVAT PANEL

Clinical exome sequencing (6161) | Whole exome sequencing (24383) | Couple carrier sequencing (2271) | Hereditary Cancer sequencing (143) | Mitochondrial genome sequencing (37) | Whole Genome sequencing

- ADVAT pipeline (International lab standard workflow)
- Asian Population & Clinical database (Updated every month)
- Specialized single, couple and trio workflow
- Specific panel for each disease
- Reanalysis for additional phenotype
- Specialized clinical genetic & genetic counsellor
- Data storage & accessibility for 5 years
- Automated reporting
- Low cost, Short TAT

Sample Type	Sample Requirement
EDTA Whole Blood	3-5 ml in EDTA tube
Purified Genomic DNA	1µg high quality DNA (50-100 ng/microlitre)
Amniotic fluid (AF)	7-10ml
POC	250ng



FOR MORE DETAILS, CONTACT US AT

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