

ADVAT EXOME SEQUENCING



What is Exome?

Exome Sequencing (ES), sequences the complete coding region of the genome. It is designed to examine all the coding regions and splice junctions of the genome. **Anderson Exome test** incorporated with mitochondrial variant analysis and CNVs.

Features of Exome Sequencing

>99% Analytical Sensitivity	All protein-coding regions with the intron-exon boundary regions of ~24383 genes	100% Coverage of Mitochondrial genome	4,947 Genes from OMIM (Dec-2024 update)
Coverage of Known Pathogenic /Likely pathogenic variants (ClinVar 2,18,000)	Uniform coverage with mean depth of >80-100X	98% of targeted basepairs covered at ≥10x	Coverage of alternate gene transcripts (MANE, GENCODE, REFSEQ)

ADVAT Copy Number Variants (CNVs)

Provides precise confirmation of CNVs identified through Whole Genome or Exome Sequencing

Custom-tailored and cost-effective solutions encompassing all genes and CNVs

Identifies both heterozygous and homozygous variations. Delivers high precision for both rare and common CNVs

In-House Validation of 32 Samples

Power of NGS CNV Detection

Detectable events

Supported data types

	Small 150b+	Medium 1-10Kb	Large 10 Kb+	Gene panel	Whole exome	Whole genome
MLPA	✓			✓		
CMA			✓			✓
ADVAT-CNV	✓	✓	✓	✓	✓	✓

- One single testing paradigm
- True simplification of clinical workflow
- Saves time and money - all on site

Autism and Other Neuro developmental disorders (24 CNV regions)

1q21.1	2p16.3 deletion	2q34 duplication	2q37 deletion
2q37.3 deletion	5p deletion	5q35	5q16 deletion
9q34 duplication	15q11.2BP1-BP2 deletion	15q13.3 deletion	15q15 deletion
15q24 deletion	16p11.2 (proximal and distal)	16p12.2 deletion	16p13.11 deletion
16p13.3 deletion	17p13.3 duplication	17q11.2 duplication	17q12
17971.31	Xp11.22 duplication	Xq28 duplication	

HIGHLIGHTS OF ADVAT PIPELINE

- 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



Scan here to visit our website

Central Processing Center
No.26, Rajarathinam Street,
Kilpauk, Chennai - 600010.

Helpdesk:
+91 73059 88862

