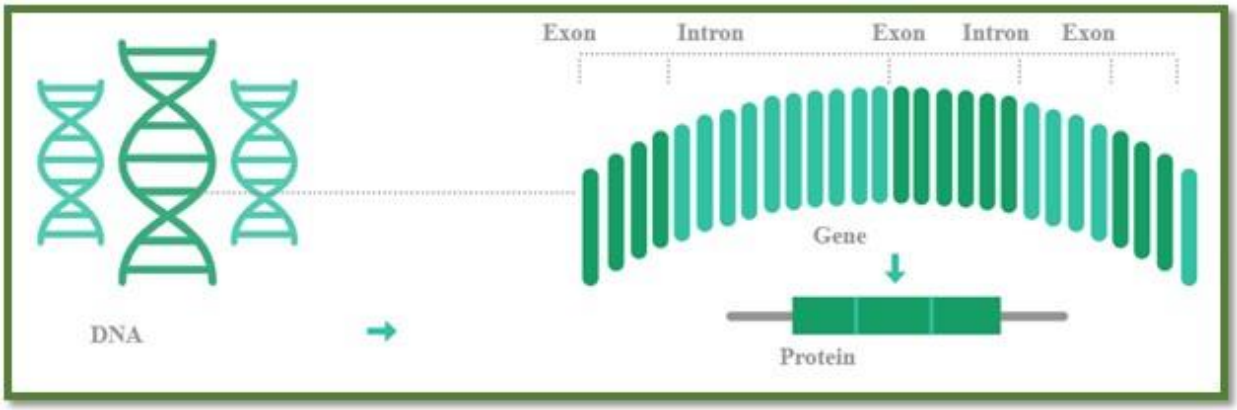
Clinical Exome Sequencing

What is Exome?

- Exome is defined as 1-2% of the genome that encodes proteins (Exons).
- It is collection of exons, that is protein-coding segments plus their affiliated 5'- and 3'-untranslated regions.



What is Clinical Exome?

- Clinical Exome Sequencing is Study of only clinically relevant exonic region (Expressed Region) of the whole Genome.
- As most of the disease-causing mutations (about 85%) detected to date are in the exonic regions of gene.
- Covers over 6000 genes reported to be clinically relevant in OMIM* and/or HGMD* database.
- Focuses on the part of the genome we understand best, the clinically relevant exonic region of the genes.

Why Clinical Exome?

- To diagnose genetically heterogenous diseases such as: Intellectual disability/Development delay, Muscular dystrophy, Ataxia etc.
- To diagnose unclear or atypical presentation of a genetic disorder.
- Analysis of trios (parental genes) significantly improves the diagnostic yield for genetically heterogeneous disorders

How our Clinical Exome is better than Others:

Clinical Exome Sequencing	Oncquest	Eurofins	Dr Lal path Labs
No of Genes Covered	8342	5200	6500
CNV Analysis	Yes	Yes	Yes
CAP Accredited (Proficiency Testing)	Yes	No	No

Test Details:

S. No.	Test Name	Test Code	MRP	Technique	Run days at Section	TAT / Reported on
1	Clinical exome + Mitochondrial genome sequencing (GEFS + Genetic Panel)	SMO10426	40000	Next Generation Sequencing (NGS)	Every day if received before 1:00 p.m.	56th working days by 7:00 p.m.
2	Clinical Exome Sequencing	SMO10421	30000	NGS with Sanger validation		40 Working Days
3	Clinical Exome sequencing plus CNV	SMO10427	88000	(NGS + CNV)		