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

- 4,468 genes from OMIM (Sep-2023 update)
- 2,937 genes from ClinVar (Aug-2023 update)
- 3,931 genes from Orphanet (Nov-2022 update)
- 4,217 genes from HPO (Sep-2022 update)
- ACMG (73 genes; Version 3; incidental findings

Coverage of known pathogenic/likely pathogenic mutations

- High confident pathogenic variants from ClinVar (70,703)
- 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**
- **Latest analysis pipeline using GRCh38.p13 assembly embedded on ADVAT pipeline**
- **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)**