theraCUES
SIGNALS FOR LIFE

Exome Sequencing – Standard (Germline) HG-EXOM-STD

Applications

Method utilizes targeted capture of protein-coding regions (exons) followed by high-throughput next-generation sequencing to identify germline genetic variants within clinically and biologically relevant genes. Sequencing reads are aligned to a reference genome, enabling accurate detection of single nucleotide variants (SNVs) and small insertions/deletions (InDels). Exome sequencing provides a cost-effective alternative to whole genome sequencing for studying inherited diseases, population genetics, and gene–phenotype associations.

Pros

- Protein-coding region
- High coverage of genes encoding proteins
- Well-established clinical interpretation frameworks
- Suitable for inherited disease studies as first line investigation of genetic disorders/population analysis

Notes

- Non-coding regions are not captured
- Structural variant detection is limited to short coding genes
- Coverage may vary across GC-rich regions
- Validation recommended for clinically relevant variants

Input

Genomic DNA (Germline)

DNA Quantity: ≥ 100 ng - 1 µg recommended

Sample Types

Blood | Tissue | Buccal Swabs | Cells

Platform

Illumina platforms | 150 × 2 |
100 X Exome, 150 X 2 ~34.9 mb | 8 GB data

Process Map

Sample DNA Extraction → DNA QC → Capture + Library Prep → NGS Data Acquisition → Data QC → Reference Alignment → Variant annotation → Gene Rank by Phenotype → Reporting

Standard Deliverables

Download Sample Report

- Sample and Library Quality Control (QC)
- Summary of Methods and Raw Data Quality Control
- Reference Genome Alignment Summary
- Variant Identification and Summary Statistics
- Zygosity and Coding, Variant Annotation and Variant Effect Prediction
- Pathogenic variants(if detected)
- Rare variants by specific phenotypes if described