

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

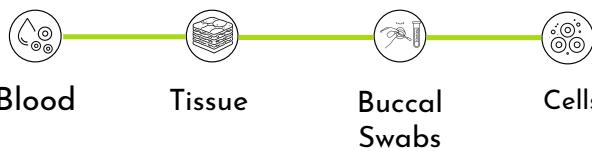
Input



Genomic DNA (Germline)

DNA Quantity: $\geq 100 \text{ ng} - 1 \mu\text{g}$
recommended

Sample Types



Platform Illumina platforms | 150 \times 2 |
100 X Exome, 150 X 2 ~34.9 mb | 8 GB data

Process Map



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