

1.High-Throughput Technologies

Next-Generation Sequencing (NGS)

Whole Genome Sequencing (WGS)

Whole Exome Sequencing (WES)

RNA Sequencing (RNA-Seq)

Evolution of Sequencing

Sanger → Pyrosequencing → SMRT → Nanopore

↔ Enables: Comprehensive mutation profiling, SNP detection, structural variation analysis

2 Bioinformatics Tools

Preprocessing & QC

FastQC, Trimmomatic, Cutadapt

Alignment

STAR, HISAT2, Bowtie

Quantification

featureCounts, HTSeq, Salmon, StringTie2

Splicing Analysis

rMATS, SUPPA

Visualization

ggplot2, Matplotlib, Giotto, Seurat, BEAVR

Integration

Multi-omics, spatial transcriptomics

↔ Supports: Data cleaning, mapping, expression analysis, splicing detection, visual interpretation

3. Biomarker Discovery

Types of Biomarkers

PD-L1, EGFR, BRCA1/2

Applications

Diagnosis, prognosis, therapy selection

Challenges

Data volume, quality control, validation

↔ Requires: Statistical learning, machine learning, predictive algorithms

4. □ Open-Source Repositories

Genomic Data

TCGA, GEO, SRA, ENCODE, Expression Atlas

Research Data

Figshare, Dryad, Harvard Dataverse, re3data

Pathway & Spatial Tools

Reactome, SpatialDB