



# Biointellitech: Advanced Genomic Analysis Services

Biointellitech combines biotechnology and artificial intelligence to deliver comprehensive genomic analysis services. Our platform leverages Novogene's industry-leading whole genome sequencing capabilities and our proprietary bioinformatics pipeline to provide researchers with high-quality, publication-ready data for human genome studies.

# Library Quality Control



## Novogene's Cutting-Edge Sequencing Advantages



### State-of-the-Art NGS Technologies

World-leading sequencing capacity using the latest generation of Illumina Novaseq 6000 system, capable of sequencing up to 28,000 human genomes per year.



### Highest Data Quality

Guaranteed Q30 score of 80%, exceeding Illumina's official guarantee of 75%, ensuring exceptional reliability of sequencing results.



### Extraordinary Informatics Expertise

Cutting-edge bioinformatics pipeline and internationally recognized best-in-class software to provide customers with highly reliable "publication-ready data".

# Project Workflow

## Sample Quality Control

Rigorous quality assessment of submitted samples to ensure suitability for sequencing.

## Library Construction

Genomic DNA is sheared into fragments, end-repaired, A-tailed, ligated with adapters, PCR amplified, size selected, and purified.

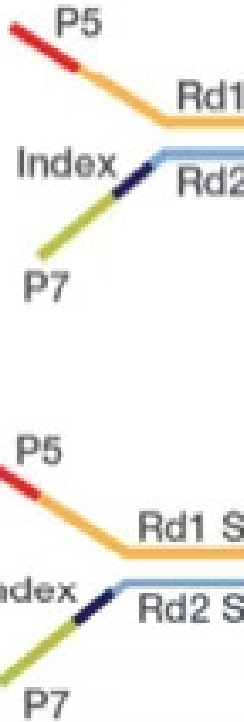
## Sequencing

Libraries are quantified and sequenced on Illumina platforms according to required data amount.

## Bioinformatics Analysis

Comprehensive data processing including quality control, alignment, variant calling, and annotation.

Double-stranded cDNA  
(from RNA protocol)



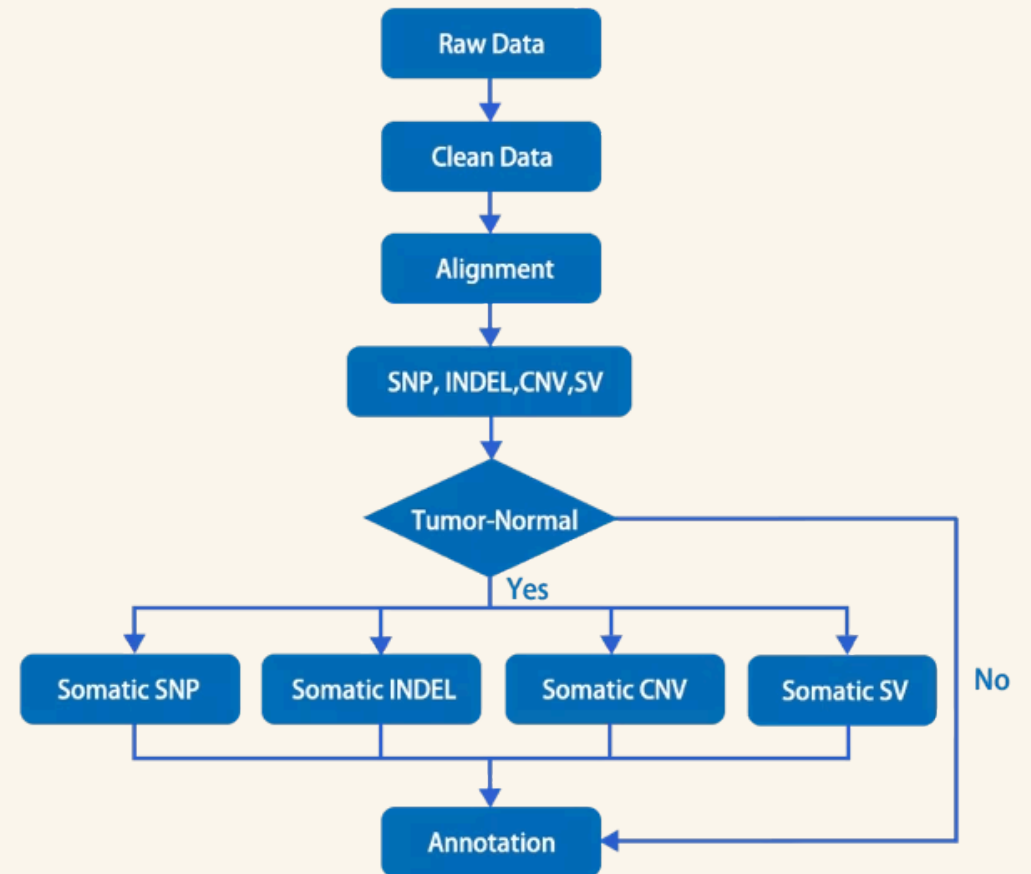
D. Ligate index adapter



# Bioinformatics Analysis Pipeline

Our comprehensive bioinformatics analysis includes:

1. Data quality control: filtering out reads containing adapters or with low quality
2. Alignment with reference genome, statistics of sequencing depth and coverage
3. SNP/InDel/SV/CNV calling, annotation and statistics
4. Somatic variant analysis for tumor-normal paired samples



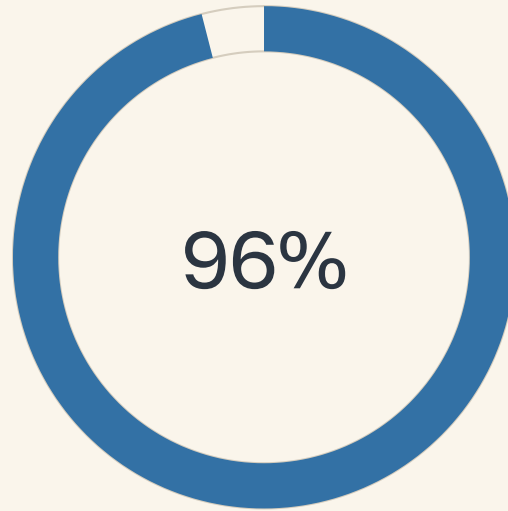
Our pipeline utilizes industry-standard tools including BWA for alignment, GATK for variant calling, ANNOVAR for annotation, and specialized tools for somatic mutation detection.

# Sequencing Quality Metrics



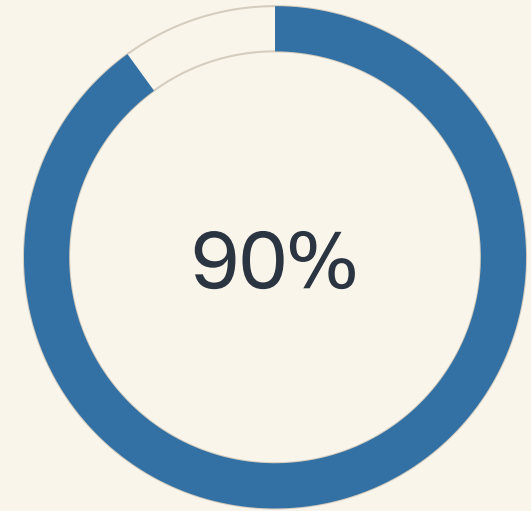
Effective Rate

Percentage of clean reads that pass quality control, demonstrating exceptional data quality.



Q20 Score

Percentage of bases with phred-scaled quality scores greater than 20, indicating high base call accuracy.



Q30 Score

Percentage of bases with phred-scaled quality scores greater than 30, exceeding industry standards.

Our sequencing platform consistently delivers high-quality data with error rates below 0.04%, ensuring reliable downstream analysis results.

# Variant Detection and Analysis



## SNP Detection

Identification of single nucleotide polymorphisms across the genome, with comprehensive annotation of coding consequences, population frequencies, and functional predictions.



## InDel Analysis

Detection of small insertions and deletions, with classification of frameshift and non-frameshift variants and their potential impact on protein function.



## Structural Variants

Identification of larger genomic alterations including deletions, duplications, inversions, and translocations that may impact gene function.

# Comprehensive Variant Annotation

## Functional Annotation

- Protein coding changes
- Genomic regions affected
- Deleteriousness prediction (SIFT, PolyPhen, CADD)
- Conservation scores (GERP++, phyloP)


## Population Databases

- dbSNP identification
- 1000 Genomes Project frequencies
- ExAC population data
- ESP6500 allele frequencies

## Clinical Relevance

- COSMIC cancer mutations
- ClinVar clinical significance
- OMIM disease associations
- GWAS Catalog findings
- HGMD disease mutations





# Somatic Mutation Analysis for Cancer Research

9,936

Somatic SNPs

Detected in tumor sample, including 99 in coding regions that may drive cancer development.

487

Somatic InDels

Small insertions and deletions unique to tumor tissue, potentially affecting gene function.

964

Somatic SVs

Larger structural variants including 679 translocations that may create fusion genes.

280

Somatic CNVs

Copy number variations covering 1.2Gb of the genome, including amplifications of potential oncogenes.

Our tumor-normal paired analysis identifies somatic mutations that may drive cancer progression, providing insights for precision oncology applications.