

Human Whole Exome Sequencing

1. Sample Requirements

Sample Type	Amount (Qubit®)	Volume	Concentration	Purity (NanoDrop™)
Genomic DNA	≥ 400 ng	≥ 20 µL	≥ 20 ng/µL	OD260/280=1.8-2.0; no degradation, no contamination
cfDNA/ctDNA	≥ 50 ng	-	-	Fragments should be in multiples of 170 bp, no genomic contamination
Genomic DNA from *FFPE	≥ 300 ng	-	-	Fragments should be longer than 1000 bp

*FFPE: Formalin-fixed-paraffin-embedded

2. Sequencing Parameters

Platform	Illumina NovaSeq 6000
Read length	Paired-end 150 bp
Recommended sequencing depth	For Mendelian disorder/rare disease: effective sequencing depth above 50× (6 G); For tumor sample: effective sequencing depth above 100× (12 G)
Data quality	Guaranteed ≥ 85% bases with Q30 or higher
**Turnaround time	22 working days from verification of sample quality to data releasing without bioinformatic analysis

**Turnaround time varies depending on the project volume.

3. Data Analysis Contents

Standard Analysis
Data quality control: filtering reads containing adapter or with low quality
Alignment with reference, statistics of sequencing depth and coverage
SNP and InDel calling, annotation and statistics
Somatic variant detection (only apply for tumor-normal paired samples) SNP calling, annotation and statistics InDel calling, annotation and statistics CNV calling, annotation and statistics

Advanced analysis		Methods
Cancer	Screening for Predisposing Genes (feasible if only normal samples are provided)	
	Mutational Spectrum & Mutational Signature	
Driver gene analysis	Identification of Known Driver Genes	
	Significantly Mutated Gene & Pathway Analysis	
	Mutation Relation Test of Significantly Mutated Genes	
	Identification of Driver Genes Based on Mutation Clustering Bias	
	Identification of Driver Somatic CNVs	
	Mutation Site Displaying	
Tumor heterogeneity analysis	Tumor Purity & Ploidy Estimation	
	Intra-tumor Heterogeneity Analysis	
	Tumor Evolution Analysis (One normal and at least 3 tumor samples from the same patient are needed)	
	Tumor Neoantigen Identification	
Monogenic disease	Candidate Variant Filtration	
	Analysis under dominant / recessive model	
	Linkage Analysis	
	Region of Homozygosity Analysis (ROH)	
Polygenic disease	Candidate Variant Filtration	
	Analysis under dominant / recessive model	
	Linkage Analysis	
	Region of Homozygosity Analysis (ROH)	
	De novo SNV/InDel Analysis	