Human Whole Exome Sequencing

1. Sample Requirements

Sample Type	Amount (Qubit®)	Volume	Concentration	Purity (NanoDrop™)
Genomic DNA	⩾ 300 ng	⩾ 15 μL	⩾ 20 ng/μL	OD260/280=1.8-2.0; no degradation, no contamination
cfDNA/ctDNA	⩾ 30 ng	-	-	Fragments should be in multiples of 170 bp, no genomic contamination
Genomic DNA from *FFPE	⩾ 500 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

	Methods	
Advanced analysis	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