1. Sample Requirements

Sample Type	Amount (Qubit®)	Volume	Concentration	Purity
Genomic DNA	≥400 ng	≥20 μL	≥20 ng/μL	OD260/280=1.8-2.0; No degradation, no contamination
MDA product/Single Cell Amplified DNA	≥1 μg	≥20 μL	≥20 ng/μL	OD260/280=1.8-2.0; No degradation, no contamination
Genomic DNA from FFPE *	≥0.8 μg	≥20 μL	≥20 ng/μL	OD260/280=1.8-2.0; No degradation, no contamination

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× (6G) For tumor sample: effective sequencing depth above 100× (12G)
Data quality	Guaranteed ≥80% bases with Q30 or higher
Turnaround time	22 working days from verification of sample quality to data releasing without bioinformatic analysis (depending on sample size); additional turnaround time needed for bioinformatic analysis

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	**Method**	
	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 Identificaiton
	Monogenic disease	Candidate Variant Filteration
		Analysis under dominant / recessive model
		Linkage Analysis
		Region of Homozygosity Analysis (ROH)
	Polygenic disease	Candidate Variant Filteration
		Analysis under dominant / recessive model
		Linkage Analysis
		Region of Homozygosity Analysis (ROH)
		De novo SNV/INDEL Analysis