Human Whole Genome Sequencing

1. Sample Requirements

1.1 Illumina platform (350 bp insert DNA Library)

Sample Type	Amount (Qubit®)	Volume	Concentration	Purity (NanoDrop™)
Genomic DNA	⩾ 200 ng	⩾ 20 μL	⩾ 10 ng/μL	OD260/280=1.8~2.0 no degradation, no contamination
Genomic DNA (PCR free)	⩾ 1 μg	⩾ 20 μL	⩾ 20 ng/μL	
Genomic DNA from *FFPE	⩾ 800ng	-	-	Fragments should be longer than 1500 bp

* FFPE: Formalin-fixed, paraffin-embedded

1.2 PacBio platform (SMRTbell® DNA Library)

Library Type	Sample Type	Amount	Volume	Concentration	Purity (NanoDropTM/Agarose Gel)
PacBio sequel II DNA CLR library	** HMW Genomic DNA	⩾ 8 μg	⩾ 50 μl	⩾ 80 ng/μl	Fragment size: most of DNA fragment is above 40k; A260/280=1.8~2.0; A260/230=1.5~2.6; ***Nc/Qc=0.95~3.00
PacBio sequel II DNA HiFi library	HMW Genomic DNA	⩾ 15 μg	⩾ 50 μl	⩾ 80 ng/μl	Fragment size: most of DNA fragment is above 30k; A260/280=1.8~2.0; A260/230=1.5~2.6; Nc/Qc=0.95~3.00

** HMW: High Molecular Weight
***Nc/Qc:NanoDropconcentration/Qubitconcentration

1.3 Nanopore platform (Ligation 1D DNA Library)

Sample Type	Amount (Qubit®)	Volume	Concentration	Purity (NanoDrop™)
*HMW Genomic DNA	⩾ 8 μg	⩾ 50 μL	⩾ 100 ng/μL	OD260/280=1.8-2.0; OD260/230=1.5-2.6; fragments should be ⩾ 30 kb;

* HMW: High Molecular Weight

2. Sequencing Parameters

Platform	**Illumina NovaSeq 6000**
Read length	Paired-end 150 bp
Recommended sequencing depth	For tumor tissues: 50×, adjacent normal tissues and blood 30× For rare diseases: 30-50×
Data quality	Guaranteed ⩾ 80% bases with Q30 or higher
***Turnaround time**	4~5 weeks from verification of sample quality to data releasing without bioinformatic analysis

Platform	PacBio Sequel II
Read length	average > 15 kb for Sequel II
Recommended sequencing depth	For genetic diseases: 10-20× For tumor tissues: ⩾ 20×
***Turnaround time	7~8 weeks from verification of sample quality to data releasing without bioinformatic analysis

Platform	Nanopore PromethION
Read length	average > 17 Kb
Recommended sequencing depth	For genetic diseases: 10-20× For tumor tissues: ⩾ 20×
***Turnaround time	6~7 weeks 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 to reference genome; statistics of sequencing depth and coverage
Variant (SNP, InDel, CNV, and SV) 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 SV calling, annotation and statistics Display of Genomic Variants with Circos

Advanced analysis	Methods
Personalized analysis (Cancer & Disease)	HLA typing
	CRISPR/Cas9 Off-target Analysis
	Xenograft Tumor Analysis
	Integration Site Detection

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
		Identification of Driver Mutations in Noncoding Regions
		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)
		Fusion Gene Detection
		Tumor Neoantigen Identification

Advanced analysis	Methods
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

Advanced analysis	Methods
Personalized analysis (Cancer & Disease)	HLA typing
	CRISPR/Cas9 Off-target Analysis
	Xenograft Tumor Analysis
	Integration Site Detection