Novogene

Whole Exome Sequencing

Novogene Whole Exome Sequencing (WES) is a sequencing method that employs high-throughput sequencing of exon regions of more than 20,000 genes per individual, that are enriched through sequence capture technology. WES targets all protein-coding regions (~1% of the whole genome) responsible for 85% of known disease-causing variants. It provides molecular diagnosis of genetic diseases and enables the exploration of novel mutations and new pathogenic mechanisms.

Test Performance

Mean Sequencing Coverage ≥100×	Average Raw Data ≥12 Gb	Base Pairs Covered at ⩾20× ≥93%
Repeatability ≥ 95% for SNVs ≥ 95% for InDels	Sensitivity ≥92% for SNVs ≥ 92% for InDels	
Specificity ≥99% for SNVs ≥ 99% for InDels	Mapping Rate ≥99%	

Turnaround time

2 weeks from sample receival to report.

Specimen type

Whole blood in an EDTA purple-top, Buccal Swab, Saliva, or gDNA.

Certification

ISO17025.

Contact us

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