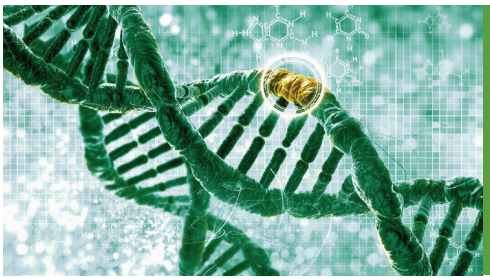
Novogene

Human Whole Exome Sequencing



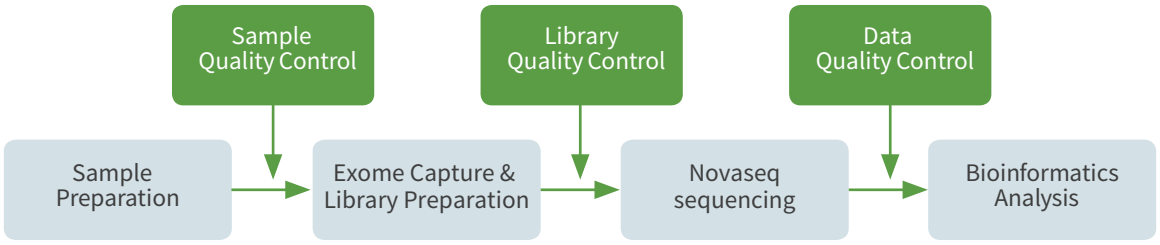
Exome sequencing provides a cost-effective alternative to whole genome sequencing, as it targets only the protein coding region of the human genome responsible for a majority of known disease-related variants. Whether you are conducting studies in rare mendelian disorders, complex disease, cancer research, or human population studies. Novogene' s comprehensive human whole exome sequencing (hWES) service provides a high-quality, affordable, and convenient solution.

Extensive experience with >3000 projects

Data quality exceeds Illumina's official guarantee

In house pipeline to meet different analysis requirement

Project workflow



Sequencing parameter

Platform	Illumina NovaSeq 6000
Read length	Pair-end 150
Read length	Pair-end 150
Recommended Sequencing Depth	For Mendelian disorder/rare disease: effective sequencing depth above 100× (12G) For tumor samples: effective sequencing depth above 200× (24G)
Data quality	Guarantee Q30 ⩾80%
Turnaround time	23 working days from verification of sample quality to data delivery （<24 samples）

Samples requirement

Library Type	Sample Type	Amount Required	Volume	Concentration	Purity
Short insert library (180-280 bp)	Genomic DNA	⩾400 ng	⩾ 20 μL	⩾ 20 ng/μL	OD260/280=1.8~2.0 No degradation, no contamination
	FFPE Genomic DNA	⩾800 ng	-	-	Fragments should be longer than 1000 bp

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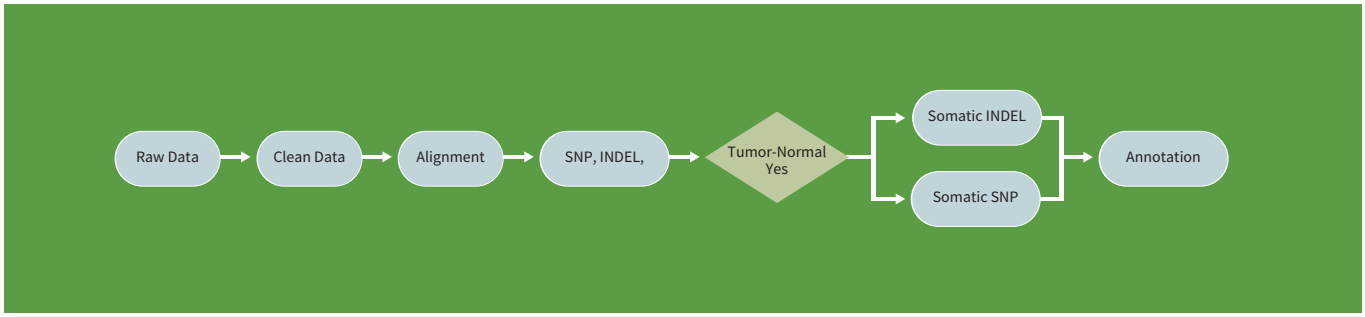
Analysis Pipeline



Standard Analysis

- Data quality control
- Alignment with reference genome, statistics of sequencing depth and coverage
- SNP/InDel/SV/CNV calling, annotation and statistics
- Somatic SNP/InDel/SV/CNV calling, annotation and statistics (paired tumor samples)

Advanced Analysis

- Tumor evolution analysis (Cancer)
- Tumor neoantigen identification (Cancer)
- Candidate variant identification (Disease)
- Linkage analysis (Disease)
- Xenograft tumor analysis (PDX)

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Novogene Data

Below are Novogene data from human genome sequencing projects.

Sample name	Raw Data[1]	Effective (%) [2]	Q30 (%)[3]	Mapped (%) [4]	Average sequencing depth[5]	Genome coverage (%) [6]	Percentage of Genome with ⩾ 4X coverage (%) [7]	Percentage of Genome with ⩾ 10X coverage (%) [8]	Percentage of Genome with ⩾ 20X coverage (%) [9]
Novo 1	6.5	98.17	88.74	99.72	67.3	99.7	99.4	98.1	92.2
Novo 2	9	98.99	90.20	99.86	91.54	99.9	99.7	99.2	97.1
Novo 3	12.3	98.57	93.19	99.88	117.46	99.9	98.5	98.2	94.8
Novo 4	15.2	98.71	93.22	99.81	179.56	99.6	99.3	98.5	96.6
Novo 5	18.7	98.96	93.53	99.85	188.92	99.8	99.5	98.6	96.4
Novo 6	19.8	98.81	92.41	99.84	215.3	99.6	99.4	98.8	97.5

1 Original sequencing data (in gigabases).
2 Percentage of clean reads from all raw reads.
3 Percentage of reads with an average quality greater than Q30.
4 Percentage of total reads that mapped to the reference genome (UCSC hg38).
5 Average sequencing depth.
6 Percentage of genome covered by sequencing.
7 Percentage of bases in genome with a sequencing depth 4x.
8 Percentage of bases in genome with a sequencing depth 10x.
9 Percentage of bases in genome with a sequencing depth 20x.

Publications using Novogene's expertise

Year	Journal	Article
2019	Cancer Res	Multiregion sequencing reveals the genetic heterogeneity and evolutionary history of osteosarcoma and matched pulmonary metastases
2019	Neural Plasticity	New genotypes and phenotypes in patients with 3 subtypes of Waardenburg Syndrome identified by diagnostic next-generation sequencing.
2019	International Journal of Cancer	Preliminary exploration of potential molecular therapeutic targets in recurrent and metastatic parathyroid carcinomas.
2019	Eur Respir J	Germline BMP9 mutation causes idiopathic pulmonary arterial hypertension
2019	Nature Communications	Recurrent GNAQ mutation encoding T96S in natural killer/T cell lymphoma
2018	J Cell Mol Med	Whole-exome sequencing identifies a novel mutation of GPD1L
2018	Cell	Mutational landscape of secondary Glioblastoma guides MET-Targeted trial in brain tumor