

# Human Whole Exome Sequencing Solutions



85% of disease-causing mutations are found in the coding regions of the human genome. Through exome enrichment, **Whole Exome Sequencing (WES)** can identify the variants from the coding regions and provide comparable and comprehensive genome variations results to Whole Genome Sequencing on research applications in genetic diseases, population genetics and oncology.

Novogene's comprehensive human whole exome sequencing (hWES) service provides a high-quality, affordable and convenient solutions.

## Our Key Features & Advantages



### Wide Selection of Probes & Services

Multiple library choices (Agilent, Twist and IDT probes), custom panels, DNA extractions and a comprehensive data analysis pipelines for your research needs.



### Customized Analyses

Customized data analyses for disease and cancer.



### Competitive Turnaround Time

The strategic locations of our labs across Asia and our extensive NGS system can provide faster turnaround time.



### Clinical Grade Exome Sequencing

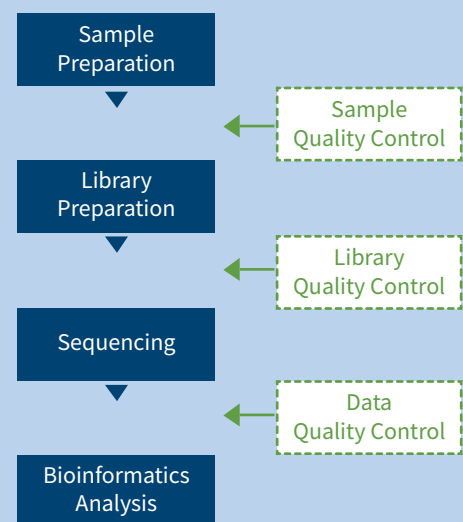
Our CAP and CLIA laboratories ensures that your specimens are processed under the highest quality and in compliance to strict regulations.



### Real-time Project Management

Our Customer Service System (CSS) online platform allows for real-time project tracking 24/7, collaboration with your team and other helpful information.

## Project Workflow



## Sample Requirements

Library Type	Sample Type	Amount	Volume	Concentration	Purity (NanoDrop™)
Human Exome Library	Genomic DNA	≥ 300 ng	≥ 15 µL	≥ 20 ng/µL	OD260/280=1.8-2.0; no degradation, no contamination
	FFPE DNA	≥ 500 ng	-	-	Fragments should be longer than 1000 bp

## Standard Service

Platform	Illumina NovaSeq 6000
Read Length	Paired-end 150
Recommended Sequencing Depth	<ul style="list-style-type: none"> <li>For Mendelian disorder/ rare-disease: sequencing depth above 50x (6G)</li> <li>For tumor sample: sequencing depth above 100x (12G)</li> </ul>
Data Quality	Guarantee ≥ 85% bases with Q30 or higher
**Turnaround Time	Within 2-7 weeks

\*\*For project w/o bioinformatic analysis. TAT is from confirmation of library preparation after sample QC and is subject to change based on sample size, bioinformatic analysis and additional services.

## Standard Analysis Pipeline



Standard Analysis	Advanced Analysis
Data quality control	Tumour evolution analysis (Cancer)
Alignment with reference genome, statistics of sequencing depth and coverage	Tumour neoantigen identification (Cancer)
SNP/ InDel/ SV/ CNV calling, annotation and statistics	Candidate variant identification (Disease)
Somatic SNP/ InDel/ SV/ CNV calling, annotation and statistics (paired tumor samples)	Linkage analysis (Disease)
	Xenograph tumor analysis (PDX)

## Publications

Library Type	IF	Title
Advanced Science	15.84	A Fifteen-Gene Classifier to Predict Neoadjuvant Chemotherapy Responses in Patients with Stage IB to IIB Squamous Cervical Cancer
Nature Communications	12.121	Clonal architecture in mesothelioma is prognostic and shapes the tumour microenvironment

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Please contact your Novogene representative.

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# Human Whole Exome Sequencing Solutions



## Discover which exome probes is best suited for my studies

	Option 1 Clinical grade	Option 2 Cost effectiveness	Option 3 Comprehensive	Option 4 Mouse samples
Probes	IDT: xGEN Exome Hybridization Panel	Agilent SureSelect All Exon V6	Agilent SureSelect All Exon V8 Twist Exome 2.0	Agilent SureSelect TX Mouse
Target size	39M	60M	Agilent V8: 35.1M Twist 2.0: 36.5M	49.6M
Database coverage	RefSeq	CCDS, RefSeq, GENCODE	Agilent V8: CCDS, RefSeq, GENCODE Twist 2.0: CCDS, RefSeq, GENCODE, Clinvar, HCMG, Ensembl	Ensembl, RefSeq
Sample types	gDNA from blood, Saliva Cell lines/ Pellets	gDNA from blood, Saliva Cell lines/ Pellets, FFPE, Other tissues, cfDNA/ ctDNA	gDNA from blood Saliva, Cell lines/ Pellets	gDNA from mouse
TAT*	within 2 weeks	within 4 weeks	6-7 weeks	within 5 weeks
Price	+++	++	+++	+++
Notable features	<ul style="list-style-type: none"> <li>Sequencing performed in CAP laboratories.</li> <li>The xGEN hybridization panel can capture and provide appropriate representation of samples with high GC and AT contents.</li> <li>Fast turnaround time.</li> </ul>	<ul style="list-style-type: none"> <li>Suitable for different types of samples and vast variety of research.</li> <li>Well established exome enrichment probe.</li> </ul>	<ul style="list-style-type: none"> <li>Comprehensive and the most up-to-date coverage of protein coding regions.</li> </ul> <p><b>Agilent V8:</b> Ability to cover the TERT promoter region and other hard-to-capture exons, as well as having a better uniformity of its coverage distribution.</p> <p><b>Twist 2.0:</b> Offers clinically relevant panels such as Clinvar, alongside other genomic databases for translational research.</p>	<ul style="list-style-type: none"> <li>Based on the current UCSC mm9 mouse genome and has a full coverage of the mouse exome by capturing 221,784 exons and 24,306 genes.</li> </ul>

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