Novogene

Clinical Exome Solutions for Rare Diseases

Whole Exome Sequencing (WES) is the most effective tool for diagnosing complicated and multi-system rare diseases. WES searches through all coding regions of currently identified genes, yielding a high chance of correctly diagnosing patients with a complex phenotype. Novogene offers comprehensive and cost-effective clinical exome solutions that meet the needs of patients and your business.

www.novogene.com | clinical@novogeneusa.com

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Why Partner With Us?

- High-quality WES services, bioinformatic analysis, and interpretation.
- Fast turnaround time at a highly competitive price.
- Customized data file types and reports.

Who Benefits?

Hospitals

Transform patient care by providing affordable genetic diagnostic services to your patients.

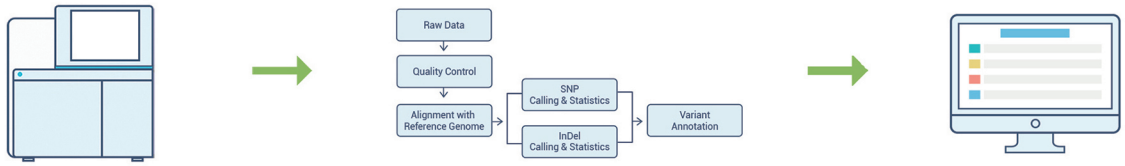
Diagnostic Laboratories

Implement highly complex genetic testing without expensive equipment purchases, validations, or any other associated costs.

Bioinformatics Services

Be a one-stop shop and expand your product offerings with our wet-lab capabilities, providing sequencing data quality you can trust.

Explore Clinical Exome Solutions at Novogene



Whole Exome Sequencing

DNA Extraction
Library Prep
Sequencing

- Performed at Novogene CAP-accredited or CLIA-certified laboratories.
- Data quality with high on-target reads percentage and coverage uniformity.
- Fast Turnaround Time.

Bioinformatic Platform

Data Quality Control
Alignment
Variant Calling & Annotation

- Knowledge-driven NGS analysis, interpretation, and reporting platform based on the GeneCards Suite Knowledge base from >120 data sources and Novogene's in-house database.
- Rapid diagnosis of causal mutations, and prioritization of variants based on their association with patients' phenotypes.
- Evaluation of variants with respect to pathogenicity and causality, which are categorized according to ACMG guidelines.

Interpretation

Variant Evaluation
& Assertion
Test Report & Sign-off

- Report includes primary findings related to patients' phenotypes, medical actionable secondary findings, as well as carrier status for autosomal recessive disorders.
- Clinical interpretation is provided following international best-practice guidelines.

Novogene Corporation Inc.

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Service Features

Sample Types:
Blood, Saliva, Buccal Swab, or DNA

Exome Capture:
Agilent SureSelect Human All Exon V6

Sequencing Platform:
Illumina NovaSeq 6000

Turnaround Time:
10 Days (from sample receival to FASTQ)

CLIA-validated WES Performance Metrics

Mean Sequencing Coverage	103X
Average Raw Data	13.75G
Base Pairs Covered at ≥20x	≥94%
Coverage Uniformity	≥90%
Repeatability	≥ 98% for SNVs ≥ 88% for Indels
Sensitivity	≥ 98% for SNVs ≥ 90% for InDels
Specificity	≥ 99% for SNVs ≥ 98% for InDels
Mapping Rate	≥97%

Custom Deliverables

File / Report Types	Sequencing Only	Sequencing + Analysis	Sequencing + Analysis + Interpretation
FASTQ Files	✓	✓	✓
Data QC Report	✓	✓	✓
BAM Files	✗	✓	✓
VCF Files	✗	✓	✓
Medical Report	✗	✗	✓

Data Releasing Through AWS

Free Data Uploading Service

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COLLEGE of AMERICAN PATHOLOGISTS

Clinical Laboratory Improvement Amendments