GENEWIZ®

CLINICAL WHOLE EXOME SEQUENCING

GENEWIZ clinical-grade whole exome sequencing (WES) is a cost-effective yet robust alternative to whole genome sequencing that enables deep sequencing of key disease-related loci for use in clinical trials and personalized medical diagnostics. This service has been optimized to produce high-quality sequence information and uniform coverage across the exome. Our validated workflow strictly conforms to standards set by the Clinical Laboratory Improvement Amendments (CLIA) in addition to the College of American Pathologists (CAP).

CLIA
CAP

THE GENEWIZ DIFFERENCE

- **Superior data quality** that exceeds Illumina® benchmarks
- **Population-scale sequencing capacity** for large clinical trials and reference lab overflow
- **Dedicated Ph.D. project managers** provide consultations and ongoing support
- **Rapid turnaround** delivers results in a few weeks
- **High-throughput variant detection** facilitates patient stratification for clinical trials
- **Sample-to-variant calling workflows** with optional in-house Sanger confirmation

DNA Extraction | Library Preparation with Exome Capture | Sequencing on Illumina NovaSeq™ | Variant Calling | Lab Director Signoff (Validated Service Only)

Clinical-grade WES Workflow. Each stage of our modular workflow is completed in a CLIA-certified and CAP-accredited laboratory by certified staff scientists on qualified equipment. Rigorous quality control is performed throughout the process to ensure the highest quality data.

Solid science.
Superior service.

CONTACT GENEWIZ

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Service Levels

GENEWIZ clinical-grade WES is available at two service levels:

- **CLIA-Validated:** This level is recommended if data is used for diagnostic purposes or is reported to patients. You will receive raw data files and a variant report signed by our accredited laboratory director.
- **CLIA Environment:** This more cost-effective option offers greater flexibility for clinical infrastructure work that does not require signoff from a laboratory director.

		CLIA-Validated	CLIA Environment
Report with Lab Director Signature		✓	—
Applications	Germline variant detection	✓	✓
	Somatic variant detection	*Coming Soon*	✓
Laboratory Setting	CLIA-certified & CAP-accredited lab	✓	✓
	CLIA/CAP-certified equipment	✓	✓
	CLIA/CAP-trained personnel	✓	✓
	Secure data server with restricted access	✓	✓
Accepted Sample Types	Genomic DNA	✓	✓
	Whole blood	✓	✓
	Cell pellet	✓	✓
	Fresh frozen tissue	*Coming Soon*	✓
	Saliva	*Coming Soon*	✓
	FFPE	*Coming Soon*	✓
	Other	—	✓
Data Output (Coverage)		≥4.5 Gb (≥50X)	3-30 Gb (40 – 400X)
Variant Calling	Single nucleotide variants (SNVs)	✓	✓
	Insertions or deletions (INDELs)	✓	✓
	Structural variants (SVs)	—	✓
	Copy number variations (CNVs)	—	✓

Performance Specifications

We assessed our WES assay using a combination of Genome in a Bottle (GIAB) reference samples as well as DNA extracted from healthy donor blood and validated by whole genome sequencing. At mean coverage of 50X, SNVs were detected with >99.6% sensitivity, and INDELs were detected with >95.9% sensitivity.

Validated Mean Coverage		≥50x
% bases ≥10X Coverage		97.8%
Single nucleotide variants (SNVs)	Specificity	99.9%
	Sensitivity	99.6%
	Precision	98.7%
Insertions or deletions (INDELs)	Specificity	99.9%
	Sensitivity	95.9%
	Precision	92.7%

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