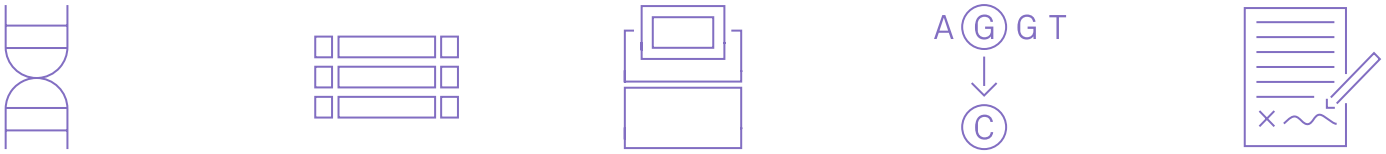
Genomics & Analytical Services | Preclinical & Clinical

Clinical Whole Genome Sequencing

Azenta Life Sciences' clinical-grade whole genome sequencing (WGS) offers unparalleled precision and accuracy for unbiased exploration of the human genome. Our PCR-free WGS workflow empowers clinical researchers to detect disease-related genetic variants in both coding and non-coding regions. In addition, the sequencing data can be used to identify novel variants of unknown significance and reanalyzed once additional disease-related information becomes available. Our validated assay strictly adheres to requirements set forth by the Clinical Laboratory Improvement Amendments (CLIA) and the College of American Pathologists (CAP).

The Azenta Life Sciences Difference

- **Superior data quality** that exceeds Illumina® benchmarks
- **PCR-free workflow** reduces bias and increases precision in challenging genomic regions
- **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
- **Population-scale sequencing capacity** for large clinical trials and reference lab overflow



DNA Extraction

PCR-Free Library Preparation

Sequencing on Illumina NovaSeq™

Variant Calling

Lab Director Signoff (Validated Service Only)

Clinical-Grade WGS Workflow. Each stage of our modular workflow is performed 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.

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Service Levels

Azenta Life Sciences' clinical-grade WGS 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	✓	✓
	Fresh frozen tissue	*Coming Soon*	✓
	Saliva	*Coming Soon*	✓
	FFPE	*Coming Soon*	✓
Data Output (Coverage)		≥90 Gb (≥30X)	45 – 270 Gb (15 – 90X)
PCR-Free Workflow		✓	✓
Variant Calling	Single nucleotide variants (SNVs)	✓	✓
	Insertions or deletions (INDELs)	✓	✓

Performance Specifications

We assessed our PCR-free WGS workflows using the well-characterized Genome in a Bottle (GIAB) reference samples, as well as DNA extracted from healthy donors. At a minimum mean aligned coverage of 22X, SNVs were detected with >99.8% sensitivity, and INDELs were detected with >95.5% sensitivity. Further, variant calling precision was consistently above 98% across several GIAB replicates.

Validated Mean Coverage		≥22×
% bases ≥10X Coverage		95%
Single nucleotide variants (SNVs)	Specificity	99.99%
	Sensitivity	99.80%
	Precision	99.70%
Insertions or deletions (INDELs)	Specificity	99.99%
	Sensitivity	95.5%
	Precision	98.9%

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