

Research Human Whole Genome Sequencing

Product Overview

Broad Clinical Labs (BCL) offers a variety of research Whole Genome Sequencing (WGS) services. Leveraging over thirty years of experience in human whole genome sequencing and a deep connection to Broad scientists, BCL has [powered](#) some of the world's largest WGS projects, from the Human Genome Project, to the 1000 Genomes Project, NHLBI TOPMed, gnomAD, All of Us, and more. Our operation is built for scale, with access to the most cutting-edge sequencing technologies, providing advantages in cost and turn-around-time with best-in-class sample qualification, automation, and bioinformatics. Data is generated using the same trusted laboratory and analytical processes that have produced more than half a million genomes to-date¹. Our standard offerings, listed below, include PCR-free sample preparation (though PCR-plus sample preparation is available for low input samples) and sequencing on Illumina NovaSeq X Plus instruments in our CLIA licensed, CAP-accredited lab:

- **Research PCR-free WGS** with 30X, 60X, or 80X short read² coverage.
- **Research PCR Plus WGS** with 30X coverage

What's Included

- Sample receipt and sample fidelity QC (96 SNP fingerprinting)
- De-multiplexing, aggregation, and alignment using the Illumina DRAGEN™
- Single nucleotide variants (SNVs), small insertions and deletions (InDels), and copy number variants (CNVs).

¹ <https://broadinstitute.github.io/gp-dashboard/>

² We also offer long read WGS to 12X or 24X coverage on the Pacific Biosciences Revio™ sequencing technology.

**Research WGS as low as
\$350**

Pricing dependent on number of samples and project details

Input Requirements

- Whole blood in lavender-top (EDTA) tube or;
- Saliva, buccal swabs, and buffy coats collected in approved devices or;
- Extracted DNA; $\geq 1\mu\text{g}$ at $\geq 10\text{ ng/ul}$ (350 ng of material used for processing, with remaining material available for rework and QC).
- Research WGS from FFPE, fresh frozen tissue, and cell pellets is also available.
- Minimal participant metadata including relevant identifiers and biological sex.
- Data delivery location.

Data Deliverable

Data will be delivered through a secure portal via a Google bucket, or alternatively can be delivered via AWS or Azure.

- Research WGS: CRAM and VCF files.

Broad Clinical Labs (BCL) also offers multiple clinical Whole Genome Sequencing (WGS) services, all of which start from a $\geq 30\text{X}$ coverage PCR-free whole genome backbone. To learn more about our clinical offerings visit broadclinicallabs.org