BROAD CLINICAL LABS

PRODUCT DATASHEET

Research Human Whole Genome Sequencing

Research WGS starting at $399

Pricing dependent on number of samples and project details

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-date[1]. 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 read[2] coverage.
- **Research PCR-plus WGS** with 30X or 60x short read coverage

What's Included

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

Input Requirements

Research PCR-free WGS:

- Whole blood, saliva, buccal swabs, buffy coats, fresh frozen tissue, cell pellets collected in approved devices or;
- Extracted DNA; ≥1µg total at 10ng/µL - 110ng/µL concentration; 50 - 300µL preferred volume, 30µL minimum acceptable volume
- Minimal participant metadata including relevant identifiers and biological sex.
- Data delivery location.

Research PCR-plus WGS:

- FFPE Scrolls, FFPE Slides; or Genomic DNA (≥300ng total in 50-300µL volume; 6-110 ng/µL preferred concentration, 3 ng/µL minimum acceptable concentration) extracted from blood, saliva, buccal swab, buffy coat, fresh frozen tissue, cell pellet, peripheral blood mononuclear cells (PBMC), or polymorphonuclear leukocytes (PMN) that does not meet the minimum input requirements of the Research PCR-free product
- Minimal participant metadata including relevant identifiers and biological sex.
- Data delivery location.

Data Deliverable

- CRAM file (aligned to hg38)
- Single sample VCF

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

[1] https://broadinstitute.github.io/gp-dashboard/

[2] We also offer long read WGS to 10x, 20x, 40x coverage on the Pacific Biosciences Revio™ sequencing technology.