BROAD CLINICAL LABS

PRODUCT DATASHEET

Human Whole Genome Sequencing

Research WGS as low as $350
Clinical WGS with interpretation $1000
Clinical WGS w/o interpretation $750

Pricing dependent on number of samples and project details

Product Overview

Broad Clinical Labs (BCL) offers both research and clinical Whole Genome Sequencing (WGS) services, with or without interpretation. 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 instruments in our CLIA licensed, CAP-accredited lab:

- Research WGS with 30X, 60X, or 80X short read[2] coverage.
- Clinical technical-only (no interpretation) WGS; ≥30X short read coverage.
- Clinical WGS with interpretation (panel or whole genome analysis); ≥30x short read coverage.

What's Included

- Sample receipt and sample fidelity QC (96 SNP fingerprinting)
- De-multiplexing, aggregation, and alignment using the Illumina DRAGEN™.

What's Included (cont.)

- Single nucleotide variants (SNVs), small insertions and deletions (InDels), and copy number variants (CNVs).
- For clinical WGS only: ≤28-day TAT from sample receipt subject to project size and capacity (discussed at project initiation).

Clinical Interpretation

All analyses start from our PCR-free whole genome backbone. Genomic variants are annotated, prioritized, and analyzed using the Fabric Genomics platform, then reviewed and signed out by our team of board-certified clinical laboratory geneticists. We offer customizable analyses including the following:

The Actionable Genetic Screening Panel is based on the ACMG-recommended list of actionable secondary finding and guidelines outlined by ACMG[3] and generally includes pathogenic and likely pathogenic variation in this set of genes.

Custom Screening for healthy populations can be requested on a project-by-project basis. Pathogenic and likely pathogenic variation will be reported.

Custom Diagnostic Panels can be designed for a specific indication or phenotype. Pathogenic and likely pathogenic variants can be reported.

Diagnostic whole-genome analysis will return pathogenic and likely pathogenic variants, as well as (VUSs) in genes related to the patient's phenotype. This type of testing can be run on the proband alone, or with one or both parents (trios and duos).

[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.

[3] PMID: 35802134 https://pubmed.ncbi.nlm.nih.gov/35802134/

Input Requirements

- Whole blood in lavender-top (EDTA) tube or;
- Saliva, buccal swabs, and buffy coats collected in approved devices or;
- Extracted DNA (for clinical WGS offerings, must be extracted in a CLIA compliant manner); ≥1µg at ≥10 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.
- For clinical samples, a test order from an authorized individual.
- 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.
- Clinical technical-only WGS: CRAM, VCF, and technical report (pdf).
- Clinical interpretation report[4] (PDF and/or JSON), delivered through a secure online portal or API with sequencing data (CRAM) available upon request.

Clinical WGS with Interpretation - Report Example

Below is an example of a clinical interpretation report, which is delivered in PDF or JSON format.

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Patient
Patient Name: Jane Doe
DOB: 01/01/1990
Patient ID:
MRN: 012345

Sample
Sample ID:
Accession ID: Sample 6
Collection Date: 01/01/2023
Received Date: 01/03/2023
Material Type Received: Bodily Fluid:Saliva
Report Date: Not yet approved

Order
Ordering Clinician: Rosalind Franklin
Ordering Clinical Institution:
Institution Address:

Indication for Testing
Actionable Genetic Screening Panel

Test Name
Genome Sequencing - Actionable Genetic Screening Panel (78 genes), Broad Institute

Test Result
Positive Result A pathogenic/likely pathogenic variant was detected.

GENE	VARIANT	VARIANT CLASSIFICATION	CONDITION	INHERITANCE PATTERN	ZYGOSITY	PARENTAL ORIGIN
BRCA2	c.9076C>T p.Gln3026Ter	Pathogenic	Hereditary breast and ovarian cancer	Autosomal dominant	Heterozygous	Not Tested

A single variant was identified in this test. One pathogenic variant was identified in BRCA2, which is associated with hereditary breast and ovarian cancer, with autosomal dominant inheritance. Additional information about these findings is included in Variant Details.

Recommendations
Genetic counseling is recommended for this individual and their relatives. For assistance in locating nearby genetic counseling services, please contact your provider or find a genetic counselor through the National Society of Genetic Counselors (https://findageneticcounselor.nsgc.org/). For questions about this report and interpretation please contact bcl-support@broadinstitute.org.

These results should be interpreted in the context of the patient's medical and family history. Please note that variant classification and/or case interpretation may change over time if more information becomes available. Periodic reinterpretation of genome sequencing data is recommended and may be requested by a medical provider, especially if new symptoms arise. Due to the limitations of available testing technologies, additional testing may still be indicated for this individual and family. Testing limitations are detailed below.

Broad Clinical Laboratories, LLC.
Commonwealth of Massachusetts Clinical Laboratory License No. 5347
320 Charles St, Cambridge, MA 02141

CLIA No. 22D2055652 CAP No. 8707596
Lab Director Heidi Rehm, PhD FACMG
bcl-support@broadinstitute.org

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[4] Note: This service does not currently include other variant types, including complex structural variants and repeat expansions.