BROAD CLINICAL LABS

PRODUCT DATASHEET

Clinical Human Whole Genome Sequencing with Interpretation Report

Clinical WGS with panel interpretation starting at $749
Clinical WGS with interpretation $1000
Pricing dependent on panel, number of samples and project details

Product Overview

Broad Clinical Labs (BCL) is a CLIA-licensed/CAP accredited lab offering multiple clinical Whole Genome Sequencing (WGS) services with interpretation[1], all of which start from a ≥30X coverage PCR-free whole genome backbone and include an interpreted report. Genomic variants are annotated, prioritized, and analyzed using a tertiary analysis variant interpretation platform, then reviewed and signed out by our team of board-certified clinical laboratory geneticists. Interpretation options include[2]:

- The Actionable Genetic Screening Panel, which is based on the ACMG-recommended list of actionable secondary findings and guidelines outlined by ACMG[3] and includes pathogenic and likely pathogenic variation in this set of genes.
- The BCL Cancer Genetic Risk Panel analyzes 76 genes linked to cancer predisposition syndromes, covering a broad range of cancer types — including breast, gynecologic, gastrointestinal, skin, genitourinary, CNS, endocrine, and hematological. It supports clinical diagnosis confirmation, personal and family risk assessment, and informed medical decision-making.
- Diagnostic whole genome analysis, which will return pathogenic and likely pathogenic variants, as well as variants of uncertain significance (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).
- Targeted Interpretation: BCL is contracted to perform technical-only cWGS or cBGE. The customer then identifies potentially reportable variants in their patient population using their own tertiary analysis pipeline. Subsequently, BCL performs Targeted Interpretation on the reportable variant(s), and issues a clinical report summarizing these findings.

What's Included

- Sample receipt and QC
- De-multiplexing, aggregation, and alignment using Illumina DRAGEN™
- Clinical interpretation of single nucleotide variants (SNVs), small insertions and deletions (InDels), and copy number variants (CNVs)
- ≤28-day TAT from sample receipt, subject to project size and capacity (discussed at project initiation)

Input Requirements

- Capillary blood; whole blood in lavender-top (EDTA) tube; saliva, buccal swabs, and buffy coats collected in approved devices; or CLIA-compliant extracted DNA (≥1µg at ≥10 ng/uL).
- 350 ng of DNA is used for processing, with the remaining material available for rework and QC.

Data Deliverable

Data will be delivered through a secure portal via a Google bucket or API integration.

- Clinical WGS technical data, consisting of CRAM, VCFs, and metrics files
- Clinical WGS interpretation report[4] (PDF and/or JSON)

Product Specifications

- Mean Coverage: ≥ 30x
- Percent Bases @20x: ≥ 90%
- Percent Mapped Bases: ≥ 75%
- Additional Secondary Metrics

[1] Clinical technical-only WGS (no interpreted report) also available [2] Depending on project sizes and needs, BCL is also able to offer custom screening panels for healthy populations and custom diagnostic panels designed for a specific indication or phenotype. [3] PMID: 35802134 https://pubmed.ncbi.nlm.nih.gov/35802134/

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