BROAD CLINICAL LABS

PRODUCT DATASHEET

Clinical Human Whole Genome Sequencing

Clinical WGS with interpretation $1000
Clinical WGS w/o interpretation $499
Pricing dependent on number of samples and project details

Product Overview

Broad Clinical Labs (BCL) offers multiple clinical Whole Genome Sequencing (WGS) services, all of which start from a ≥30X coverage PCR-free whole genome backbone.

1. Clinical technical-only WGS (no interpreted report), with aligned data and genomic variants as data deliverables **or**
2. Clinical WGS with interpreted report. 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. Interpretation options include[1]:

- The Actionable Genetic Screening Panel, which is based on the ACMG-recommended list of actionable secondary findings and guidelines outlined by ACMG[2] and includes pathogenic and likely pathogenic variation in this set of genes.
- 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).

Leveraging over thirty years of experience in human whole genome sequencing and a deep connection to Broad Institute 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 for clinical WGS services is generated using the same trusted laboratory and analytical processes that have produced more than 600,000 genomes to-date[3]. All offerings include PCR-free sample preparation and sequencing on Illumina sequencing instruments in our CLIA licensed, CAP-accredited lab.

What's Included

- Sample receipt and fidelity QC (96 SNP fingerprinting)
- 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

- Whole blood in lavender-top (EDTA) tube; saliva, buccal swabs, and buffy coats collected in approved devices; or DNA extracted in a CLIA compliant manner (≥1µg at ≥10 ng/uL). 350 ng of DNA is used for processing, with the remaining material available for rework and QC.
- Minimal participant metadata, including biological sex and relevant identifiers
- A test order from an authorized individual
- A signed services agreement with BCL
- Data delivery location

Data Deliverable

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

- Clinical technical-only WGS: CRAM, VCF, and technical report (pdf)
- Clinical WGS with interpreted report: interpretation report[4] (PDF and/or JSON), delivered through a secure online portal or API with sequencing data (CRAM) available upon request.

[1] 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. [2]PMID: 35802134 https://pubmed.ncbi.nlm.nih.gov/35802134/ [3]https://broadinstitute.github.io/gp-dashboard/
[4] Note: This service does not currently include other variant types, including complex structural variants and repeat expansions.