BROAD CLINICAL LABS

PRODUCT DATASHEET

Clinical Human Whole Genome Sequencing with Technical Report

Clinical WGS without interpretation
$499
Volume-based discounts available for larger projects

Product Overview

Broad Clinical Labs (BCL) offers multiple Clinical Human Whole Genome Sequencing (WGS) services[1], all of which start from a ≥30X coverage PCR-free whole genome backbone.

The Clinical Human WGS - Technical Report delivers high-quality sequencing data - aligned reads and genomic variant calls - without clinical interpretation, enabling CLIA-certified entities to interpret and report results within their own distributed testing models. Sample preparation is PCR-free with sequencing on Illumina NovaSeqX Plus Platform in our CLIA licensed, CAP-accredited lab. Alignment and variant calling are performed using the Illumina DRAGEN™ platform. Every clinical whole genome sample automatically includes a comprehensive data package as a technical deliverable.

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 900,000 genomes to-date[2].

What's Included

- Sample receipt and QC
- De-multiplexing, aggregation, and alignment using Illumina DRAGEN™
- Automated pharmacogenomics (PGx) pipeline: Dragen-based star-allele and targeted-caller data for 24 key pharmacogenes, including complex genes CYP2D6 and CYP2B6
- ≤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.

- CRAM, VCFs, and technical report (pdf)
- PGx Data package: six standardized JSON, TSV, and gVCF files covering star-allele assignments and specialized targeted-caller results for CYP2D6 and CYP2B6

Product Specifications

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

[1]Clinical WGS with interpretation services also available [2]https://broadinstitute.github.io/gp-dashboard/