

Long Read Human Whole Genome Sequencing

Product Overview

Broad Clinical Labs' Long Read Human Whole Genome Sequencing service, which utilizes Pacific Biosciences Revio[™] technology for sequencing, is ideal for any research applications that benefit from long reads for de novo assembly, methylation profiling, and detection of structural variants and other complex genomic events that may not be fully resolved by short read sequencing.

Broad Clinical Labs process produces sequencing libraries that are tightly sized to ~15 kb, allowing each molecule to be sequenced in multiple passes. This enables error correction using circular consensus, with typical error rates for human samples <1% following error correction.

Additionally, Broad Clinical Labs offers the Nanobind extraction workflow, which isolates high-purity, high-molecular-weight (HMW) DNA from a wide range of sample types, further enhancing support for long-read sequencing.

Long Read Human Whole Genome Sequencing is available at 10X, 20X, and 40X sequencing coverage levels, depending on customer needs.

What's Included

- Sample receipt and Incoming QC (DNA quantification)
- Size selection (~15kb)
- Library construction and QC
- Circular Consensus Sequencing on Pacific Biosciences Revio[™] technology
- Error Correction, Alignment, and resulting Data File Generation
- Data Delivery via Terra Data Repository (TDR)

20X WGS on PacBio RevioTM starting at \$1120 Pricing dependent on number of samples and project details

Input Requirements

- 4 samples & orders in multiples of 4 for the 10X product
- No minimum for 20x or 40x product
- High Molecular Weight Genomic DNA*:
- >3 ug Human HMW DNA, >6 ug HMW DNA preferred at 40-100 ng/uL; Preferred volumes based on biological material types: Blood-110uL, cells-200uL, tissue 110uL
- Minimum Sample metadata, including the following: collaborator participant ID, collaborator sample ID, biological sex of participant
- This service is <u>not</u> recommended for samples with any of the following characteristics:
 - Total DNA quantity < 4 ug, concentration
 <30ng/ul, or volume <100µL
 - DNA is significantly degraded (>50% of DNA is smaller than 40kb), WGA or FFPE derived, A260/230 < 1.80D

Data Deliverable

- BAM File
- FASTQ File



Figure 1. F1 score for six replicates of HG002 run on SPRQ chemistry compared to an HG002 sample run on the previous chemistry. Replicates of HG002 showed comparable performance to previous chemistry and F1 score was consistent across SMRT[™] Cells.

(*) High molecular weight extraction is available as a separate service