BROAD CLINICAL LABS

PRODUCT DATASHEET

Long Read Human Whole Genome Sequencing

24X WGS on PacBio Revio™ as low as $1600

Product Overview

Broad Clinical Labs' Long Read Human Whole Genome Sequencing service, which utilizes the 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 Lab's 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.

Long Read Human Whole Genome Sequencing is available at 12X and 24X sequencing coverage levels, depending on customer needs.

What's Included

- Sample receipt and Incoming QC (DNA quantification and assessment of degradation)
- Size selection (~15kb)
- Library construction and QC
- Circular Consensus Sequencing on Pacific Biosciences Revio™ technology
- Error Correction, Aggregation, Alignment, and resulting Data File Generation
- Data Delivery via Google Cloud-based platform

Input Requirements

- Minimum batch size is 2 samples for 12X coverage and 1 sample for 24X coverage

Input Requirements (cont.)

- ≥4ug high molecular weight human DNA (≥30ng/ul minimum concentration), ≥100uL, and at least 50% of genomic DNA should be ≥40kb in size as measured by Broad Clinical Labs upon receipt
- 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 < 1.8OD

Data Deliverable

- Data delivery includes a circular consensus error corrected, aggregated, and aligned output file which can be accessed via a Google cloud based platform.
- Expected output for 12X deliverable is ~37Gb, ≥10kB and for 24X deliverable ~74Gb, ≥10kB

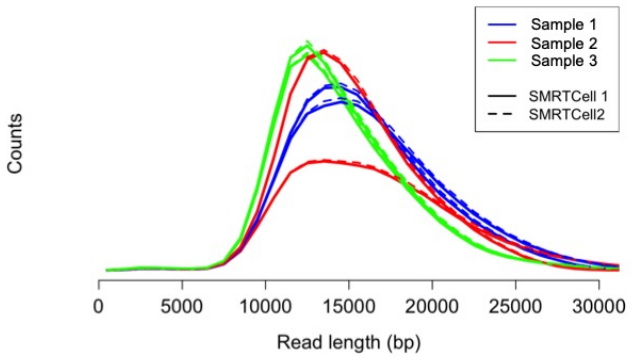

Figure 1. Read length by read count for three WGS samples, each run on two PacBio Revio™ SMRT™ Cells, with the vast majority of reads exceeding 10kB read length and good reproducibility across SMRT™ Cells.