

PRODUCT DATA SHEET

Human Whole Genome Sequencing

PRODUCT OVERVIEW

Leveraging over twenty years of experience in human whole genome sequencing, the Broad Institute powers the world's largest WGS research projects, spanning from: The Human Genome Project, to the 1000 Genomes Project, to NHLBI TOPMed and more. Our operation is built to scale, utilizing the most cutting edge sequencing technologies, providing advantages in cost and turn-around-time with best-in-class sample qualification and bioinformatics. The WGS workflows and data deliverables are designed in hand with our academic community, allowing the Broad Institute to provide global researchers the tools to recognize the promise of human whole genome sequencing.

WHAT'S INCLUDED

- Sample Receipt and Initial QC
- Sample Fidelity QC (96 SNP fingerprinting)
- Library Construction and QC
- 2x150bp Paired Sequencing: 30x, 60x
- De-multiplexing, aggregation, and alignment (Bwa-Mem)
- Data Delivery

WHOLE GENOME OFFERINGS

| Product | Utility | DNA input | Compatible Extractions | Library Construction | Read Length | Coverage Deliverable |
|--------------------------------|---|-----------|--|---------------------------|-------------|----------------------|
| Standard Coverage Human WGS | Medical & population genetics, disease research, and evolution studies. Limited input material | 100ng | FFPE*, fresh frozen tissue, blood, saliva*, cell pellets, or buffy coats | PCR library amplification | 2 x 150bp | 30X |
| Deep Coverage Human WGS | Rare disease population studies, and Case/proband for case-control tumor/normal & trio/somatic analysis Limited input material | 100ng | FFPE*, fresh frozen tissue, blood, saliva*, cell pellets, or buffy coats | PCR library amplification | 2 x 150bp | 60X |
| Standard Coverage PCR-Free WGS | Medical & population genetics, disease research, and evolution studies. | 350ng | Fresh frozen tissue, blood, saliva*, cell pellets, or buffy coats | PCR-Free | 2 x 150bp | 30X |
| Deep Coverage PCR-Free WGS | Rare disease population studies, and Case/proband for case-control tumor/normal & trio/somatic analysis | 350ng | Fresh frozen tissue, blood, saliva*, cell pellets, or buffy coats | PCR-Free | 2 x 150bp | 60X |
| Common Disease WGS | Screening and large population studies | 350ng | Fresh frozen tissue, blood, saliva*, cell pellets, or buffy coats | PCR-Free | 2 x 150bp | ~20x |

* refer to our application notes for special considerations when working with these material types, www.genomics.broadinstitute.org/products/nucleic-acid-extractions



Deep experience in producing WGS data
>100,000 Whole Genomes to date

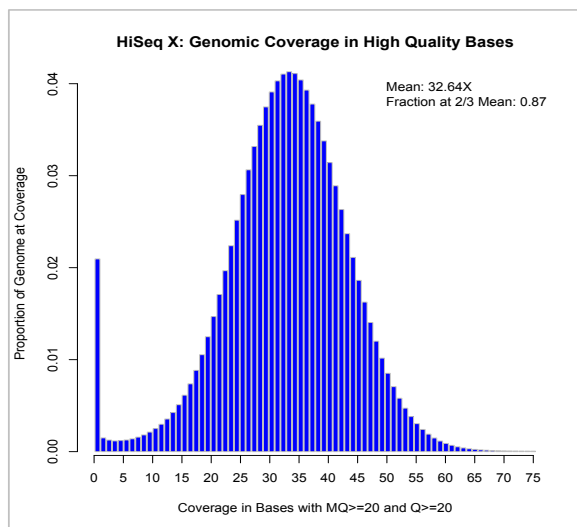
Clinical-Grade Quality Management Systems

Workflows and Deliverables defined by the Broad Institute research community

DATA DELIVERABLE

- Data accessed via secure online digital transfer
- De-Multiplexed, aggregated Picard BAM file with summary metrics

COVERAGE ACROSS THE GENOME



Genome-wide Coverage Distribution of representative sample sequenced to mean coverage of ~30X

FOR MORE INFORMATION

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