

GENOMIC SERVICES

Product Data Sheet

— Highlights

- Improved coverage across genome
- Reduction in allele distribution for indel calling
- Fewer false-positive indel calls

PCR-Free Human Whole Genome Sequencing

Preparation of sequence-ready libraries without the need for PCR amplification presents advantages in data quality for human whole genome sequencing. These advantages include a marked reduction in base specific biases that are attributed with DNA polymerases. Additional benefits to PCR-Free Human Whole Genome Sequencing significant improvements in the sensitivity to detect and reduction in false positive observations when calling indel and copy number variants.

Product Details:

This product combines PCR-Free sample preparation with Illumina HiSeq X[™] Ten v2 chemistry run to 30X or 60X Mean Coverage. This combination of highly optimized sample preparation with the most prolific sequencing capacity to date represents the highest fidelity, highest throughput, and fastest turnaround human whole genome sequencing observed to date.



Figure 1. GC Bias Plot showing improved performance of PCR Free sequencing using v2 HiSeq XTen chemistry (Blue) when compared to PCR Free using v1 chemistry (Red) and PCR + v2 chemistry (Green).



Figure 2. Improved library complexity and coverage resulting from PCR-Free WGS sample preparation.

What's Included:

- Size selected library preparation with a median insert size of 400bp +/- 25%
- Illumina HiSeq X[™] Ten Sequencing (150bp paired reads) to specified mean target coverage
- Sample identification QC and contamination fingerprinting via genotyping
- Please note that this product is solely applicable to human samples

Data Deliverable:

- De-multiplexed, aggregated Picard BAM file
- VCF file available upon request
- Data accessed via secure online digital transfer

Input Requirements:

- 350ng of purified genomic DNA
- Fresh/frozen tissue, blood, or cell pellets that preferably yield >500ng of DNA (separate charge will be applied for extractions)
- Minimum Sample data including collaborator participant ID, collaborator sample ID, gender
- Tumor/normal or case/control pairs must be received together if indel co-cleaning is required.
- * Samples failing to meet product input requirements can be attempted "on risk" and will be subject to billing regardless of data quality. FFPE samples are automatically deemed "on risk."

For more information please visit web: genomics.broadinstitute.org email: genomics@broadinstitute.org