

PCR-Free Whole Genome Sequencing

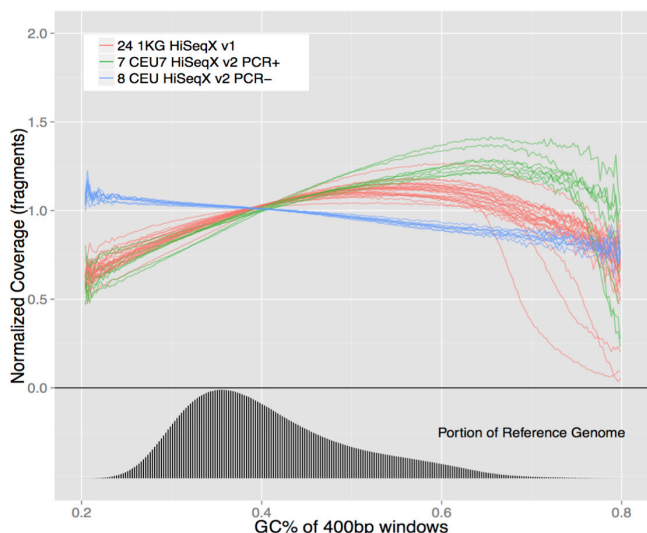
PRODUCT OVERVIEW

Preparation of sequence-ready libraries without the need for PCR amplification presents significant 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 include reduced duplication rates, reduced false positives, and greater sensitivity when calling indel and copy number variants. Our unique approach allows for lower amounts of starting material than most other groups need, even for with PCR WGS.

WHAT'S INCLUDED

- Sample Receipt and Initial QC
- Sample Fidelity QC (96 SNP fingerprinting)
- Library Construction and QC
- 2x150bp Paired Sequencing: 20x, 30x, 60x
- Data Delivery

REDUCTION OF BASE BIASES



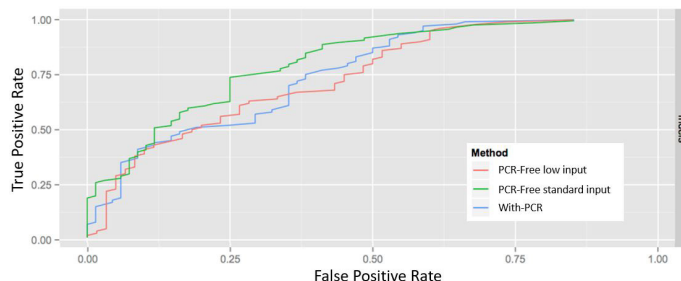
GC Bias Plot showing improved performance of PCR-Free sequencing using v2 HiSeq XTen chemistry (Blue) when compared to PCR Free using v1 chemistry (Red) and PCR + v2 chemistry (Green).

Better evenness of coverage across the whole genome than with-PCR approaches

Improved sensitivity to detect false-positive indel calls

One of the lowest input requirements on the market

HIGHER INDEL SENSITIVITY



Receiver Operating Characteristic (ROC) Curve analysis shows the improved sensitivity of PCR-Free sample prep at standard input (Green) to detect Indels. Low input PCR-Free sample prep (Red) performs similarly to With-PCR (Blue).

INPUT REQUIREMENTS

- 350ng of purified genomic DNA¹
- Fresh/frozen tissue, blood, saliva², or cell pellets that preferably yield >750ng 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

¹ 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."

² Refer to our application notes for special considerations when working with these material types, <http://genomics.broadinstitute.org/products/nucleic-acid-extractions>

DATA DELIVERABLE

- Data accessed via secure online digital transfer
- De-Multiplexed, aggregated Picard CRAM file with contamination-checked, and summary metrics
- Germline Variant Call Format (VCF) available on request