PRODUCT DATA SHEET

Whole Exome Sequencing

Industry leader in Whole Exome Sequencing
~300,000 Exomes to date

Optimized products drive variant discovery in focused disease areas

PRODUCT OVERVIEW

Leveraging vast experience in the production and analysis of human whole exome sequence data, our research offerings represent the cumulative output of the Broad Institute's knowledge, maximizing specific utility for variant discovery in specific disease areas. Careful analysis of sample inputs, library construction methods, and coverage deliverables results in a set of offerings that provide optimal data to drive scientific discovery.

The Broad Institute Genomic Service's research Whole Exome Sequencing products leverage our experience in producing ~300,000 human exomes to date. Using a content bait set containing 38Mb target territory, this content includes all of our previous exome content plus additional coding content that brings the total coverage of the RefSeq and GENCODE v12 databases to >98%. Our coverage deliverables look closer than mean target coverage, providing valuable insight into the actual percentage of targets achieving a minimum guaranteed coverage.

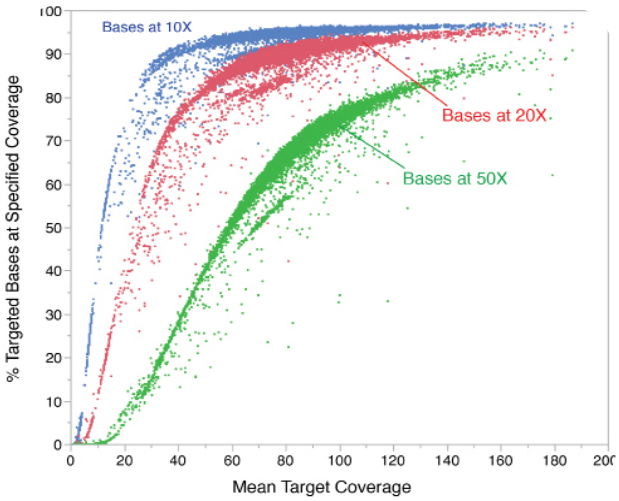
WHAT'S INCLUDED

- Sample Receipt and QC
- Sample Fidelity QC (96 SNP fingerprinting)
- Library Construction, Hybridization and QC
- Sequencing and Data Delivery

DATA DELIVERABLE

- Data accessed via secure online digital transfer
- De-Multiplexed, aggregated Picard BAM file and summary metrics
- Germline Variant Call Format (VCF) and Somatic Mutation Variant Format (MAF) files are also available

COVERAGE ACROSS THE EXOME



Distribution of coverage levels for targeted bases for representative samples sequenced to ~10X, ~20X, and ~50X mean target coverage

EXOME OFFERINGS

Product	Utility	DNA input	Compatible Extractions	Library Construction	Read Length	Coverage Deliverable	Equivalent MTC
Germline Exome	Non-cancer, including population genomics, rare and common disease cohort studies	100ng	Blood, saliva*, cell pellets or buffy coat	Transposase Base	2 x 150bp	85% targeted bases at >20X	~60-80X
Somatic Exome Standard Coverage -Available with Express 28 day TAT	Control for case-control tumor/normal & trio/somatic analysis	100ng	Fresh frozen tissue, blood, FFPE*, saliva*, cell pellets, or buffy coat	Ligation Based	2 x 76bp	80% targeted bases at >20X	~50-80X
Somatic Exome Deep Coverage -Available with Express 28 day TAT	Case/proband for case-control tumor/normal & trio/somatic analysis	100ng	Fresh frozen tissue, blood, FFPE*, saliva*, cell pellets, or buffy coat	Ligation Based	2 x 76bp	85% targeted bases >50X	~120-150X
Liquid Biopsy Germline and Somatic Exome	Compatible with liquid biopsy sample prep	2-20ng cfDNA	Blood, Plasma	Ligation Based	2 x 76bp	Germline 60X Somatic 150X	

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

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GENOMIC SERVICES

FOR MORE INFORMATION

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