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.

Starting with a 33mb Human Core Exome based on the Consensus Coding Sequence (CCDS) Project we've added 2mb of additional custom content curated from both our germline and somatic research community. Our exome covers the complete mitochondrial genome, ACMG59 genes, and now also targets additional RefSeq and Online Mendelian Inheritance in Man (OMIM) putative gene sequences, Catalogue of Somatic Mutations in Cancer (COSMIC) variants, key promoters and other motifs that have been identified as potential cancer hot spots.

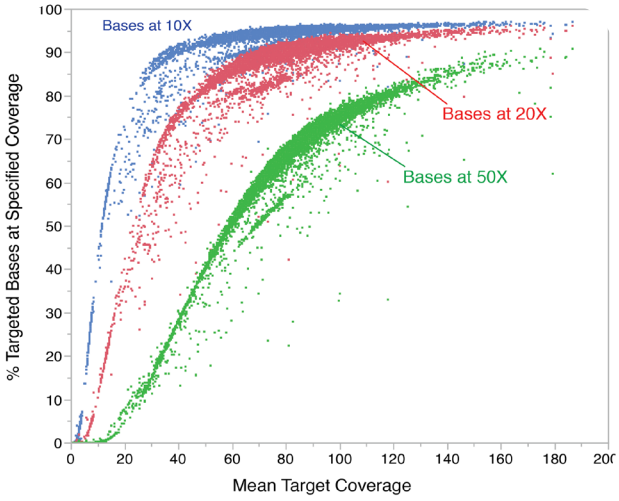
WHAT'S INCLUDED

- 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/CRAM 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	Read Length	Coverage Deliverable	Equivalent MTC
Germline Exome	Non-cancer, including population genomics, rare and common disease cohort studies	100ng	Blood, saliva*, cell pellets buffy coat, or FFPE (for normals)	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, saliva*, cell pellets, or buffy coat	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	2 x 76bp	85% targeted bases >50X	~120-150X
Liquid Biopsy Germline and Somatic Exome	Compatible with liquid biopsy sample prep	5-50ng cfDNA	Blood, Plasma	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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