BROAD INSTITUTE

Clinical Whole Exome Sequencing with Germline Analysis

GENOMIC SERVICES

Product Data Sheet

The Broad clinical research sequencing lab is a clinical laboratory accredited by the College of American Pathogists (CAP), licensed by the State of Massachusetts and registered with the Centers for Medicare and Medicaid Services to provide testing under the CLIA regulations[1]. The clinical research sequencing lab was created to further the Broad Institute mission of creating, and making available tools for genomic medicine, and applying them to human disease.

Germline Exome

The clinical research sequencing lab offers Whole Exome Sequencing that leverages the Broad-developed solution-phase hybridization assay[2] and a co-developed with Illumina® whole exome content bait set (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%. Table 1 outlines the technical specifications of our Germline Whole Exome Sequencing Test.

Table 1.
Technical Specifications of Germline Exome Product

Targeted Territory	38 Mb	
Coverage Deliverable	≥100X MTC	
Input Material*	500ng Genomic DNA, Whole Blood, Cells, FFPE, Fresh Frozen Tissue, Buffy Coat	
Sample Prep Method	Broad With-Bead[3]	
Analytical Sensitivity+	**Specification**	**Observed**
SNV	95%	98.8%
Indel	80%	88.4%
Analytical Specificity (FPR/Mb)		
SNV	<1	0.09
Indel	<1	0.33

+Performance metrics represent mean sensitivity across targeted regions with 50bp padding.
* Please contact for additional detail on input material requirements

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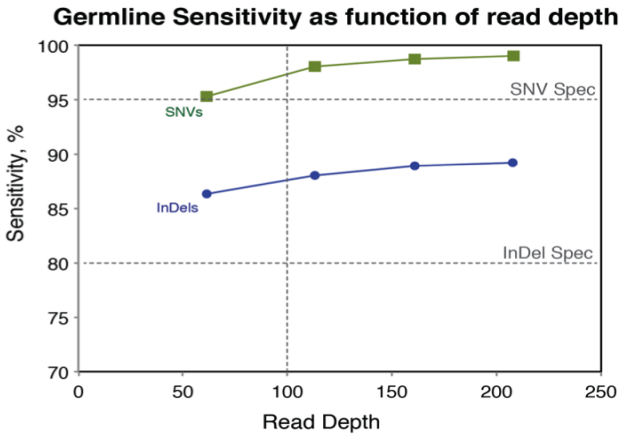

Figure 1.
Germline SNV and Indel sensitivity as a function of read depth

Deliverables, Reporting and Data Delivery

The deliverables for the Germline test include a technical report, as well as a Picard-generated BAM file for the germline exome test. The technical report outlines process specifications and performance of the sample with respect to these specifications. The VCF file lists all single nucleotide and indel variants identified by the analytic pipeline.

Data Access and Retention

All files are accessible via a dedicated, secure data portal. Data will be stored in accordance with relevant federal and state regulations for CLIA or clinical trials testing as appropriate.

Turn-Around-Time

Processing times is 21 calender days from sample receipt to delivery of the technical report, VCF and aggregated Picard BAM files.

References

1. CLIA # 22D2055652; MA License # 5347; CAP # 8707596.
2. Gnirke *et al.* **Solution hybrid selection with ultra-long oligonucleotides for massively parallel targeted sequencing.** *Nature Biotechnology* **27**, 182 - 189 (2009).
3. Fisher *et al.* **A scalable, fully automated process for construction of sequence-ready human exome targeted capture libraries.** *Genome Biology* **12**:R1 (2011).
4. Cibulskis *et al.* **Sensitive detection of somatic point mutations in impure and heterogeneous cancer samples.** *Nature Biotechnology* **31**, 213–219 (2013).