# Clinical Whole Exome Sequencing with Somatic 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.

The clinical research lab provides 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 Somatic Whole Exome Sequencing Test, which provides the neccessary depth of coverage for somatic variant calling across Tumor/Normal pairs including SNV, indel and CNV calls.

**Table 1. Technical Specifications of Broad CLIA Somatic Exome**

| | | |
|---|---|---|
| Targeted Territory | 38 Mb | |
| Coverage Deliverable | ≥150X MTC | |
| Sample Format | Tumor/Normal Pairs | |
| Input Materials* | 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% | 97.1% |
| Indel | 80% | 89.3 |
| **Analytical Specificity (FPR/Mb)** | | |
| SNV | <1 | 0.304 |
| Indel | <1 | 0.088 |

+Performance metrics represent mean sensitivity across targeted regions with 50bp padding based on 20% Allele Fraction.
*Please contact for additional detail on input material requirement

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

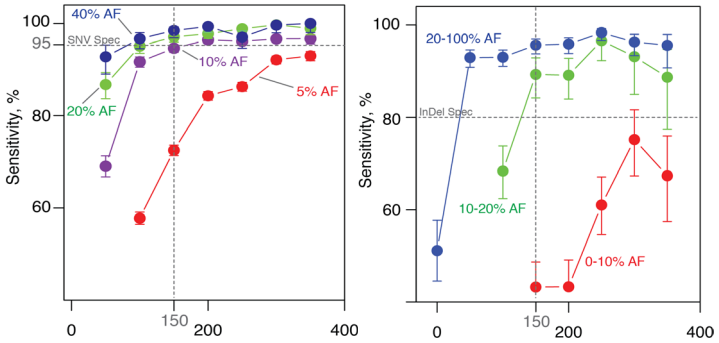


**Figure 1. Somatic SNV (left) and Indel (right) sensitivity at varying allele fractions.**

## Deliverables, Reporting and Data Delivery

The deliverables for the Somatic test is a technical report, Picard-generated aggregated, aligned BAM files, somatic SNV (VCF) and Indel mutation annotation format (MAF) files through the Broad's cancer analysis Firehose pipeline, as well as CNV calls (.called & .called.seg.annotated) delivered through the ReCapSeg algorithm. The technical report outlines process specifications and performance of the sample relative to these specifications. The variant files list all variants identified by the analytics 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.

## Turnaround Time

Processing times is 21 calendar days from sample receipt to delivery of the technical report, VCF/MAF, aggregated Picard BAM files, and ReCapSeg data.

***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).
5. Allen *et al.* **Whole-exome sequencing and clinical interpretation of FFPE tumor samples to guide precision cancer medicine** *Nature Medicine* **20**. 682-8. (2014).