Whole Exome Sequencing

**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

**EXOME OFFERINGS**

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

**DATA DELIVERABLE**

- Data accessed via secure online digital transfer
- De-Multiplexed, aggregated Picard BAM file with contamination-checked, 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

**FOR MORE INFORMATION**

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