**TruSight™ One Sequencing Panel**

The largest sequencing panel available, targeting > 4800 genes and enabling labs to expand and streamline their sequencing portfolio.

### Highlights

- **Unmatched Content and Performance**
  Targets > 4800 genes associated with human diseases

- **Single Panel Replaces Many**
  Expand and streamline your sequencing portfolio with one assay and one workflow

- **Flexible Reporting**
  VariantStudio software enables push-button gene filtering for easy exploration of genomic variant data

### Introduction

The TruSight One Sequencing Panel provides comprehensive coverage of > 4800 clinically relevant genes. Laboratories can analyze all the genes on the panel or choose to focus on a specific subset. In this way, a single panel can effectively replace all other sequencing panels. For those needing bioinformatics capabilities, TruSight One comes with VariantStudio software, a simple, yet powerful tool for analysis, classification, and reporting of genomic variants. Now there's just one workflow to follow and one procedure for managing genomic samples. The result is an integrated sample-to-report process capable of supporting an entire next-generation sequencing (NGS) portfolio (Figure 1).

### Comprehensive Assay

The TruSight One Sequencing Panel covers 12 Mb of genomic content, including 4813 genes associated to a clinical phenotype. This enables labs to focus on genes with proven relevance, rather than wading through excess data that may not be of immediate value. The panel is designed for sequencing a trio on a MiniSeq™ or MiSeq® Systems, 12 samples on a NextSeq® System using a mid-output flow cell, or 36 samples on a HiSeq® 2500 System in rapid run mode, while achieving 20× minimum depth of coverage at more than 95% of the targets.*

### Content Design Strategy

Developed by Illumina, the TruSight One Sequencing Panel focuses on the exonic regions harboring disease-causing mutations. It was designed to cover the most commonly ordered molecular assays. Using TruSight One, labs can now perform all these investigations on-site, with one panel. Genomic targets were identified based on information in the Human Gene Mutation Database (HGMD)¹, the Online Mendelian Inheritance in Man (OMIM) catalog², GeneTests.org³, Illumina TruSight sequencing panels⁴, and other commercially available sequencing panels. Combining data from these sources ensures that the TruSight One panel covers all genes currently reviewed in clinical research settings for a truly comprehensive assay.

### Superior Coverage

The TruSight One Sequencing Panel features a highly optimized probe set that supports analysis of a large number of variants. Starting with only 50 ng of high-quality DNA input, the panel delivers comprehensive coverage of the targeted exonic sequences. The panel includes > 125,395 80-mer probes, each constructed against the human NCBI37/hg19 reference genome. The probe set was designed to enrich for ~62,000 exons, spanning 4813 genes of interest (Table 1).

---

* Percentage is calculated by averaging the mean coverage for each exon, not each base.

---

**Figure 1: One Seamless Sample-to-Report Workflow** — The Illumina TruSight One Sequencing Panel and VariantStudio software allow you to go from DNA sample to report in just 4 days.*

---

For Research Use Only. Not for use in diagnostic procedures.
**Table 1: Coverage Details**

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cumulative Target Region Size</td>
<td>12 Mb</td>
</tr>
<tr>
<td>No. Target Genes</td>
<td>4813</td>
</tr>
<tr>
<td>No. Target Exons</td>
<td>~62,000</td>
</tr>
<tr>
<td>Probe Size</td>
<td>80-mer</td>
</tr>
<tr>
<td>No. Probes</td>
<td>125,395</td>
</tr>
<tr>
<td>Target minimum coverage</td>
<td>20×</td>
</tr>
</tbody>
</table>

**Figure 2: Probe Footprint** — With an approximately 500 bp DNA library (insert size of 300 bp), the probe will enrich 350–650 bp centered around its midpoint.

TruSight One probes were made using an iterative process of design and functional testing to ensure the highest performance and uniformity, optimizing sequencing capacity and enabling unmatched multiplexing. The result is at least 20× coverage of 95% of the regions on the panel.

**DNA Sequence Report**

The Illumina TruSight One Sequencing Panel and VariantStudio software allow you to go from DNA sample to report in just 4 days.† TruSight One targets 12 Mb of the human genome. The 80-mer probes target libraries of approximately 500 bp (insert size of 300 bp), enriching 350–650 bases centered symmetrically around the midpoint of the probe (Figure 2). This means that, in addition to comprehensive coverage of the major exon regions, the panel provides coverage of exon-flanking regions (splice sites). Focusing on the exome subset with known associations to inherited disease (as indicated by HGMD, OMIM, GeneTests.org, etc.) enables labs to detect variants that affect gene function more efficiently than whole-genome or whole-exome sequencing.

**Flexible, Customizable Data Enrichment**

VariantStudio software tool enables researchers to explore variant data easily, identify biologically relevant variants quickly, and enrich data with biological context. When used with the TruSight One Sequencing Panel, labs can quickly and accurately extract and report on only the disease-relevant information of interest. Using the intuitive software interface, users can effectively create subpanels from a single TruSight One analysis.

---

† Average time for a targeted gene panel. Times may vary depending on panel used.

---

For Research Use Only. Not for use in diagnostic procedures.
Perform the Sequencing

Prepared libraries are loaded onto a flow cell for sequencing with the MiniSeq, MiSeq, NextSeq, or HiSeq System. Simply place the flow cell into the instrument and run. Sequence data are exported as a .vcf file and imported easily into the VariantStudio software for analysis.

Filtered Data Analysis

The TruSight One panel can be filtered to isolate a set of genes or regions for analysis and reporting, enabling one assay to represent multiple assays. Simply generate a gene list and select this list when importing .vcf data files from the MiniSeq, MiSeq, NextSeq, or HiSeq system into VariantStudio. For ease of use, VariantStudio software offers commonly applied filters, including variant quality, population frequency, functional impact, and known disease association. In addition to single-sample filtering, VariantStudio software enables multisample comparisons that accelerate identification of causative variants. To support trio and pedigree filtering, the software provides a collection of filters that identifies variants consistent with specified inheritance modes and patterns of disease progression.

Customizable Reporting

VariantStudio software enables users to customize reports to meet requirements specific to different diseases of interest and sequencing panels. Multiple report templates can be created and stored for later use. When a template is applied to a given sample, the user simply enters or imports sample-specific information from LIMS, combines it with the methodology, a summary of results, and the reported variant categories in VariantStudio (Figure 5). Reports, which are linked to the imported sample information, are then exported in PDF or rich-text formats for downstream use.
Table 2: TruSight One Sequencing Panel Performance

<table>
<thead>
<tr>
<th>Sequencing Kit</th>
<th>Reads Passing Filter (M)</th>
<th>Read Length (bp)</th>
<th>Output (Gb)</th>
<th>Samples per Run</th>
<th>% Targets Covered at 12x Minimum</th>
<th>% Targets Covered at 20x Minimum</th>
</tr>
</thead>
<tbody>
<tr>
<td>MiniSeq Reagent Kit</td>
<td>22–25</td>
<td>2 × 150</td>
<td>7.5</td>
<td>3</td>
<td>97</td>
<td>95</td>
</tr>
<tr>
<td>MiSeq Reagent Kit v3</td>
<td>22–25</td>
<td>2 × 150</td>
<td>7.5</td>
<td>3</td>
<td>97</td>
<td>95</td>
</tr>
<tr>
<td>NextSeq Mid Output Kit</td>
<td>132</td>
<td>2 × 150</td>
<td>40</td>
<td>12</td>
<td>98</td>
<td>97</td>
</tr>
<tr>
<td>HiSeq Rapid Run Mode</td>
<td>up to 300</td>
<td>2 × 150</td>
<td>90</td>
<td>36</td>
<td>97</td>
<td>95</td>
</tr>
<tr>
<td>HiSeq Rapid Run Mode (single flow cell)</td>
<td>up to 600</td>
<td>2 × 150</td>
<td>180</td>
<td>72</td>
<td>97</td>
<td>95</td>
</tr>
</tbody>
</table>

a. Performance is reported for samples with > 50% enrichment. For the MiSeq system, this targets 1,200–1,400 K/mm² raw read density.

Accurate Data

Whether sequencing on the MiniSeq, MiSeq, NextSeq, or HiSeq System, the TruSight One Sequencing Panel yields unmatched uniformity and depth of coverage (Table 2). Trio sequencing on a single MiSeq instrument typically achieves 20× minimum depth of coverage at more than 95% of the targets.

Figure 5: Templates Enable Customizable Reporting — VariantStudio provides guided template generation. Addition of samplespecific information enables customized reporting for different clinical research areas.

Summary

The TruSight One Sequencing Panel and included VariantStudio data analysis software enable a seamless workflow. Using this comprehensive panel, labs can quickly sequence over 4800 genes with an associated clinical phenotype. Filtering results with VariantStudio then enables creation of subpanels that are responsive to customer demands and advances in scientific understanding.

Learn More

To learn more about the TruSight One Sequencing Panel, visit www.illumina.com/trusightone.

Ordering Information

<table>
<thead>
<tr>
<th>Product</th>
<th>Catalog No.</th>
<th>TG Catalog No.a</th>
</tr>
</thead>
<tbody>
<tr>
<td>TruSight One Sequencing Panel</td>
<td>FC-141-1006</td>
<td>TG-141-1006</td>
</tr>
<tr>
<td>(9 samples)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>TruSight One Sequencing Panel</td>
<td>FC-141-1007</td>
<td>TG-141-1007</td>
</tr>
<tr>
<td>(36 samples)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

a. TG-labeled consumables include features intended to help customers reduce the frequency of revalidation. They are available only under supply agreement and require customers to provide a binding forecast. Contact your account manager for more information.

References


Note regarding biomarker patents and other patents unique to specific uses of products.

Some genomic variants, including some nucleic acid sequences, and their use in specific applications may be protected by patents. Customers are advised to determine whether they are required to obtain licenses from the party that owns or controls such patents in order to use the product in customer’s specific application.

For Research Use Only. Not for use in diagnostic procedures.

© 2013-2016 Illumina, Inc. All rights reserved. Illumina, HiSeq, MiniSeq, MiSeq, NextSeq, TruSight, and the pumpkin orange color are trademarks of Illumina, Inc. and/or its affiliate(s) in the U.S. and/or other countries. Pub. No. 0670-2013-015 Current as of 04 January 2016