Overview

Whole exome sequencing is a powerful technology used to analyze protein coding regions of the genome. This massively parallel analysis, coupled with the single molecule resolution of next-generation sequencing, has played a major role in accelerating disease gene discovery by providing a cost-effective, single reaction method for analysis of the exons that constitutes the 20,000 or so genes in the genome. The power of this technology extends to not only translational research, but also clinical research. For translational research, exome sequencing allows for the analysis of all protein coding regions to study the effect of DNA variants on normal biology and disease pathogenesis. In the clinical research setting, specifically in the study of rare disorders, the simultaneous analysis of these targets enable the facilitated detection of disease-associated variants out of the 20,000 to 50,000 that can be found in the human exome. Today, with the higher standards set for target coverage to increase confidence in variant detection, the challenge is to provide comprehensive designs that deliver complete target coverage.

Key Benefits

Most comprehensive content, an exome solution for every application
- Up-to-date core content from relevant databases, targets more exons including hard-to-capture regions
- Easily add UTRs for translational research, COSMIC for cancer research, or custom content for specific applications

Deep and complete target coverage for definitive answers
- Highly optimized design for superior coverage and uniformity
- Increase breadth of coverage of targeted regions

Complete and flexible solutions from sample to data
- Compatible with SureSelect workflows for faster time to results
- Solutions from design, library prep, sample QC, automation and data analysis

Figure 1. SureSelect Human All Exon V6’s highly optimized bait design coupled with stringent capture workflows enables a high on-target metric that results in a specific mapping of reads to targets for deep coverage. This exome targets updated content from relevant databases, including hard-to-capture regions, for comprehensive analysis of protein-coding regions.
The SureSelect Human All Exon V6 is a high performing design that targets updated content from databases relevant to both the clinical and translational researchers. This content, which includes targets that in previous designs and other commercial designs have proven to be a challenge to capture, enables comprehensive coverage of protein coding regions (Figure 1).

In addition, with optimized bait design and boosting strategies that improve on coverage uniformity, the SureSelect Human All Exon V6 is able to provide high 20x end-to-end coverage of targeted exonic regions (Figure 2). To demonstrate, the coverage of genes important in constitutional research are shown. This improvement helps address the current observation of the inverse correlation between depth of coverage and breadth of coverage providing balanced coverage, maximizing variant information output for per exome sequenced.

Deep and Complete Target Coverage, Comprehensive and Accurate VariantCalling

The utility of exome sequencing is evaluated as a balance between relevant information gained and cost of sequencing. In this respect, content definition becomes an application-specific question. To address the needs of the researchers in the constitutional space, typical reference databases used to curate and annotate new genes and transcripts were mined for SureSelect Human All Exon V6 + UTR targets to facilitate detection of disease-associated variants. For cancer researchers on the other hand, the SureSelect Human All Exon V6 also provides an option that includes targets from both TCGA and COSMIC. These databases catalog variants that play roles in the initiation and progression of cancer, providing a better understanding of disease pathogenesis. Addition of custom content using SureDesign further enables this exome to be tailored to specific applications.

From Translational to Clinical Research, a Complete Exome Solution Tailored for Every Application

Accelerated workflows are critical to providing reduced turn-around time from sample to data. The SureSelect Human All Exon V6 is compatible with SureSelect workflows using the shortest hybridization times, as little as 90 minutes, providing faster sample to sequencing.

Data analysis using SureCall enables simplified workflows for single sample or trio analysis, suited for constitutional research or analysis of paired tumor and normal samples, typical in cancer research, addressing another critical bottleneck in translational or clinical research (Figure 3).

Figure 3. Faster workflows from sample to data are enabled by using SureSelectQXT, the only single-day capture solution, coupled with SureCall, a guided analysis software for single sample, paired, or trio analysis.

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