SureSelect Focused Exome

When time to answers matters

Benefits

Focused coverage of only the disease-associated regions, suited for benchtop sequencing

- Highly optimized design for deep coverage of targets in HGMD, OMIM, and ClinVar, 98% at 20x, for highly sensitive variant detection.

Sample to Sequencing in a Day

- Complemented by the efficient SureSelect^{QXT} workflow with the novel 90 minute hybridization step for faster time to sequencing

Complete and flexible solutions from sample to data

- Solutions for library prep, enrichment, QC and analysis enables a full workflow solution for analysis of targets using high output or benchtop sequencers

Overview

The introduction of next-generation sequencing has led to a paradigm shift in the way genetic analysis is carried out. By providing single molecule sensitivity, easy detection of rare alleles and those of low frequency even within highly heterogenous samples is enabled. This single molecule detection is done in a massively parallel manner allowing the inherent limitations of Sanger sequencing such as throughput, scalability, speed and resolution to be overcome. Within the constitutional disease research space, what used to entail a combined approach of linkage analysis to establish the disease gene locus, gene cloning and Sanger sequencing to interrogate specific gene candidates for the presence of the disease-associated variant, can now be condensed into a single day whole exome sequencing effort to analyze thousands of genes simultaneously with single molecule resolution. This strategy has led to the huge increase in the number of disease genes identified to cause rare Mendelian disorders[1].

The SureSelect Focused Exome is a highly targeted design that enables analysis of only the disease-associated targets providing deep coverage even on a benchtop sequencer. This design provides 20 or more reads for 95% of targets at 1.5Gb (100x) of sequencing and 98% of targets with 3Gb (200x) of sequencing enabling superior coverage of disease-associated regions even when sequenced on benchtop sequencers, facilitating highly sensitive and accurate variant calling greatly reducing time to answers (Fig 1).

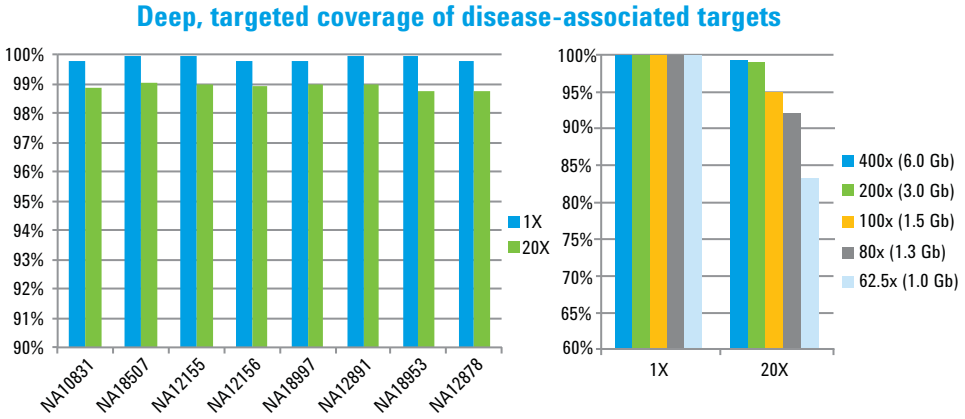

Figure 1. The SureSelect Focused Exome enables deep coverage of disease-associated targets, 98% more with 200x (3 Gb) of sequencing. With a highly uniform design, increased sequencing provides proportional increase in target coverage.

1. Boycott M. et al. Nat. Rev. Genet. 2013

SureSelect Focused Exome

Focused design for deeper coverage

Current methods to elucidate disease-associated variants require labor-intensive methods that are not scalable. Exome-based assays that offer the single-molecule resolution of NGS has the economy of scale that makes it attractive to clinical researchers by providing comprehensive coverage of thousands of targets, increasing the likelihood of identifying that one variant out of a haystack of variants that can be associated with disease. Despite this advantage offered by exome sequencing, it is not compatible with current benchtop sequencers due to their limited output.

The SureSelect Focused Exome enables targeted analysis of only those regions that have previously been associated with disease such as those annotated within HGMD, OMIM and ClinVar. When compared with other similar targeted solutions, the highly optimized design of this panel coupled with the proven SureSelect technology, provides a single assay that allows for interrogation of more disease-associated regions for more comprehensive analysis of samples, making it well suited for the study of rare idiopathic disorders (Fig 2). Addition of custom content using SureDesign lets you tailor this panel to meet your specific design requirements.

Faster answers with confidence

Reduced turn-around time to answers is important in clinical research sequencing. SureSelect Focused Exome is compatible with SureSelect^{QXT}, the fastest target enrichment workflow that enables 3.5x faster workflow compared to other competing solutions. This combined solution of SureSelect Focused Exome and SureSelect^{QXT} enables an efficient workflow from sample-to-sequencing reducing turn-around time while providing confident variant calling.

Targeted analysis using SureCall which provides guided workflows for single or trio analysis of samples enables facilitated identification of single nucleotide variants (SNPs), indels as well as copy number variants (CNVs). Furthermore, hotspot analysis is enabled through the known variants workflow which enables locus-specific detection of variants, greatly reducing time to answers.

Better Coverage of Disease-Associated Targets

A.

	SureSelect Focused Exome	Competitor I
Target Size	12 Mb	12 Mb
DATABASES COVERED		
HGMD_cds	76.80%	73.63%
OMIM_cds	45.45%	41.01%
ClinVar	83.45%	81.54%
Customization	Yes	No
1 Day Workflow	Yes	No

B.

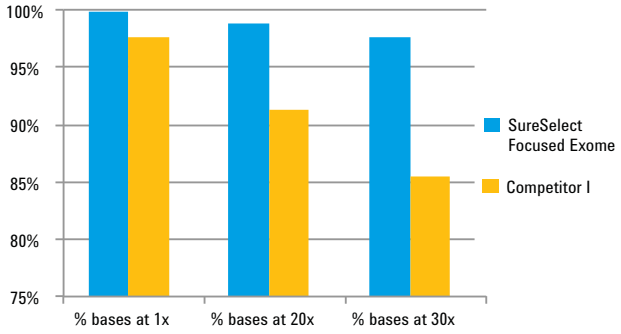

Figure 2. The highly targeted design of the SureSelect Focused Exome captures more disease-associated regions enabling more comprehensive analysis of samples (A). Optimized for performance, it enables deeper coverage of targets from HGMD, OMIM and ClinVar compared to other disease-associated capture solutions in the market when sequenced with the same average coverage enabling more sensitive and accurate variant profiling (B).

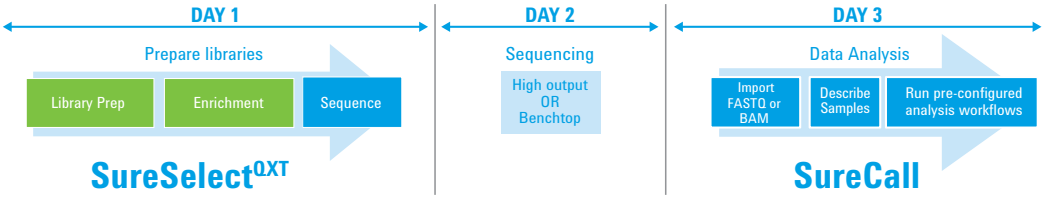

Figure 3. Faster workflows from sample to data are enabled by using SureSelect^{QXT}, the only single-day capture solution, and SureCall, a guided raw data to variants software for single sample or trio analysis.

Ordering Info

	16 rxn	96 rxn	96 auto
SureSelect^{XT} Focused Exome	5190-7787	5190-7788	5190-7789
SureSelect^{XT} Focused Exome Plus 1	5190-7790	5190-7791	5190-7792
SureSelect^{XT2} Focused Exome	5190-7797	5190-7798	5190-7799
SureSelect^{XT2} Focused Exome Plus 1	5190-7800	5190-7806	5190-7807

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