illumina®

AmpliSeq™ for Illumina Exome Panel

Targeted panel for investigating the protein-coding regions of the genome.

Highlights

- **Fast Exome Library Preparation**
  Prepare 8 exome libraries in < 7.5 hours with < 1.5 hours hands-on time
- **Comprehensive Coverage**
  Access > 97% of exonic content
- **Accurate Data**
  Achieve exceptional target coverage and high uniformity results for accurate variant calls

Introduction

The AmpliSeq for Illumina Exome Panel brings the speed and simplicity of PCR to exome sequencing, enabling researchers to sequence eight exomes in a single run and identify germline variants in less time. Starting with as little as 50 ng DNA, the panel provides uniform and specific coverage of ~59 Mb of exonic content (Table 1). As part of the AmpliSeq for Illumina targeted resequencing solution, the Exome Panel enables quick and accurate assessment of the exome for a deeper understanding of coding mutations.

Table 1: AmpliSeq for Illumina Exome Panel At A Glance

Parameter	Specification
Target Content	> 97% of exonic content
Cumulative Target Size	59 Mb
Variant Types	SNVs, indels[a]
Amplicon Size	206 bp on average
No. of Amplicons	293,303
Input DNA Requirement	50 ng minimum
No. of Pools per Panel	12
Percent Targets Covered at Minimum 500× at Recommended Throughput	> 95%
Coverage Uniformity (percent of targets with > 0.2× mean coverage)	> 90%
Percent On-Target Aligned Reads	> 80%
Total Assay Time	< 7.5 hours[b]
Hands-On Time	< 1.5 hours
DNA-to-Data Time	2.5 days

a. SNVs: single nucleotide variants; indels: insertion/deletions
b. Time represents library preparation only and does not include library quantification, normalization, or pooling

Data on file at Illumina, Inc. 2017

Simple, Streamlined Workflow

The AmpliSeq for Illumina Exome Panel is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.

The Exome Panel is supplied with lyophilized amplicons in a ready-to-use PCR plate.Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 7.5 hours, with <1.5 hours hands-on time. Resulting libraries are normalized, pooled, and then loaded on to a NextSeq™ flow cell for sequencing. Prepared libraries are sequenced using proven sequencing by synthesis (SBS) chemistry on the NextSeq System (Table 2). Up to eight exome libraries can be sequenced in a single run on the NextSeq System.

Resulting data can be analyzed locally using Local Run Manager or easily streamed into BaseSpace™ Sequence Hub. BaseSpace Sequence Hub and Local Run Manager can access the DNA Amplicon analysis workflow for alignment and variant calling. BaseSpace Sequence Hub can access BaseSpace Variant Interpreter, which assists with turning variant call data into annotated results.

Learn more about Illumina sequencing systems at www.illumina.com/systems

Learn more about Illumina informatics at www.illumina.com/products/by-brand/ampliseq/informatics.html

Table 2: Illumina Sequencing System Recommended for Use with the AmpliSeq for Illumina Exome Panel

Sequencing System	Exomes per Run	Run Time
NextSeq System (high output)	8	29 hours

Accurate Data

The AmpliSeq for Illumina Exome Panel delivers exceptional target coverage with high uniformity, resulting in highly accurate variant calls. To demonstrate assay capabilities, five different libraries were prepared from Coriell DNA standards, each with 4–16 replicates, and evaluated using the AmpliSeq for Illumina Exome Panel and NextSeq 550 System. Results showed > 90% uniformity and > 90% on-target alignment (Figure 1). In addition, single nucleotide variant (SNV) calling performance was evaluated. Results showed an average SNV Precision of 98.2% and average SNV Recall of 95.3% (data not shown).

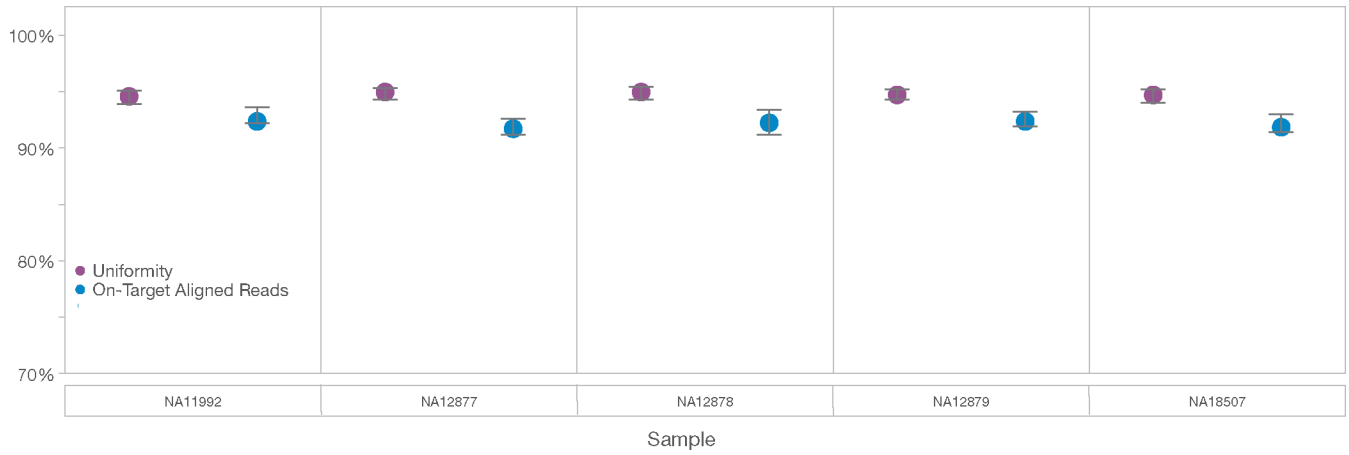

Figure 1: High Coverage Uniformity and On-Target Alignment—AmpliSeq for Illumina Exome libraries were made from Coriell DNA standards (4–16 replicates per DNA) and sequenced on the NextSeq 550 System (high output). Data represents the average of four NextSeq 550 System high output runs, eight samples per run. Error bars indicate variability of technical replicates.

Learn More

To learn more about the AmpliSeq for Illumina Exome Panel, visit www.illumina.com/products/by-type/sequencing-kits/library-prep-kits/ampliseq-exome-panel.html

To learn more about the AmpliSeq for Illumina targeted resequencing solution, read the overview at www.illumina.com/content/dam/illumina-marketing/documents/products/datasheets/ampliseq-for-illumina-targeted-resequencing-solution-data-sheet-770-2017-022.pdf

Ordering Information

Order AmpliSeq for Illumina products online at www.illumina.com

Product	Catalog No.
AmpliSeq for Illumina Exome Panel (8 reactions)	20019166
AmpliSeq for Illumina Library PLUS (24 reactions)	20019101
AmpliSeq for Illumina Library PLUS (96 reactions)	20019102
AmpliSeq for Illumina Library PLUS (384 reactions)	20019103
AmpliSeq for Illumina CD Indexes Set A (96 indexes, 96 samples)	20019105

Illumina, Inc. • 1.800.809.4566 toll-free (US) • +1.858.202.4566 tel • techsupport@illumina.com • www.illumina.com

illumina®