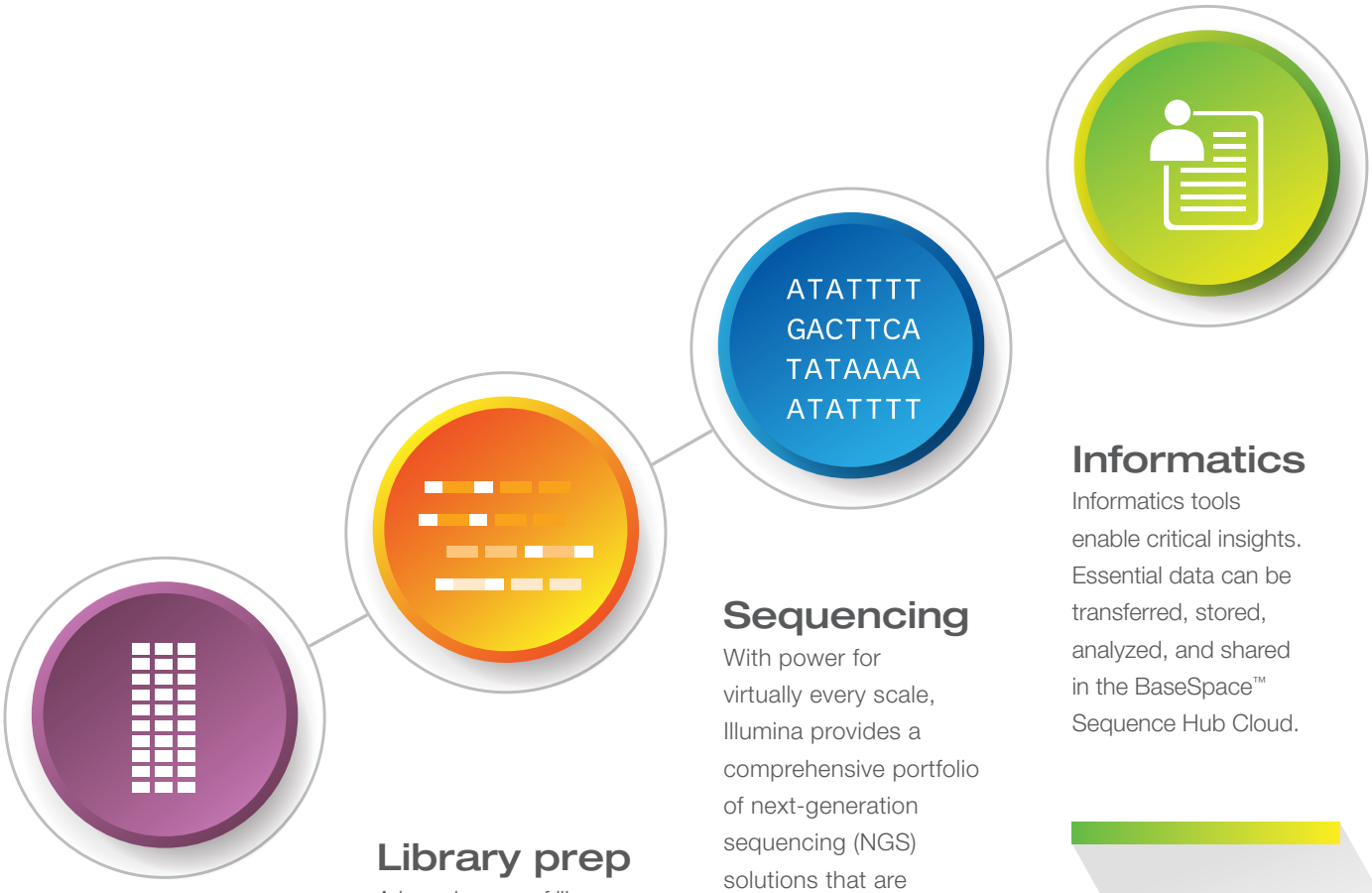
illumina®

Methods Guide

2018 | Version 4



Informatics

Informatics tools enable critical insights. Essential data can be transferred, stored, analyzed, and shared in the BaseSpace™ Sequence Hub Cloud.

Sequencing

With power for virtually every scale, Illumina provides a comprehensive portfolio of next-generation sequencing (NGS) solutions that are accessible for every study and every lab.

Library prep

A broad range of library preparation kits to enable scores of methods and applications.

Infinium™ BeadChips

Powerful, high-throughput genotyping and methylation products with off-the-shelf and custom options.

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METHODS

Genome

Whole-genome sequencing

Targeted resequencing

Custom sequencing

Transcriptome

Epigenome

Infinium genotyping systems and BeadChips

INSTRUMENTS

ANALYSIS TOOLS

Research informatics

Translational informatics

SERVICES

Instrument services, training, and consulting

APPLICATION INDEX

The Highlight 2018

New Technologies

Nextera™ DNA Flex Library Preparation
Now you can use human blood, saliva, and bacterial samples directly in your whole-genome sequencing library preparation. Learn how you can take advantage of rapid sample processing, high data quality with low bias, and low sample input requirements with the Nextera DNA Flex Library Preparation Kit on page 5.

Instruments

iSeq 100 System
Our most affordable sequencing system has arrived. Experience Illumina sequencing quality and reliability in a smaller format, lower throughput instrument. Featuring a touchscreen interface, cartridge-based reagents, and on-board informatics, the iSeq 100 System is ideal for use with small targeted DNA and RNA panels. Learn more on page 118.

Partnerships

At Illumina, we strive to bring the best possible solutions to our customers at a price that has been continuously dropping for the last decade. We've entered into partnerships with companies such as Integrated DNA Technologies (IDT) and Thermo Fisher Scientific, opening up new possibilities for our customers and advancing our central mission: To unlock the power of the genome to improve human health.

IDT DNA and RNA exome panels
In 2017, Illumina and IDT entered into a collaborative agreement to provide the IDT DNA and RNA exome panels to Illumina customers. The IDT DNA and RNA exome panels offer state-of-the-art performance and low cost.

- IDT exome (page 45)
- IDT RNA exome (pages 65 and 71)

AmpliSeq™ for Illumina targeted resequencing solution
In 2018, Illumina and Thermo Fisher Scientific entered into a collaborative agreement to provide Illumina customers access to the AmpliSeq portfolio. Now, customers can take advantage of best-in-class amplicon technology on the best-in-class sequencing technology, saving time, money, and precious sample, while improving confidence in results.

- AmpliSeq for Illumina cancer research panels (page 24)
- AmpliSeq for Illumina genetic disease research panels (page 77)
- AmpliSeq for Illumina Community panels
- AmpliSeq for Illumina Custom panels (page 38)

Nextera DNA Flex

The next era in library preparation

A fast, integrated workflow for a wide range of applications, from human whole-genome sequencing to amplicons, plasmids, and microbial species.

Whole-genome sequencing across various applications

Small
- Human microbiome
- Microbiology
- Public health research
- Metagenomics

Large
- Agrigenomics
- Model organism research
- Plant/animal research

Human
- Cancer genomics
- Variant detection
- Genetic risk studies
- Population genetics

Flexibility across all Illumina sequencers

MiniSeq System | MiSeq System | NextSeq 550 System | NovaSeq 6000 System

Save time and increase efficiency with fewer steps

Mechanical Fragmentation Library Prep
1. DNA extraction
2. DNA quantification
3. DNA shearing
4. Adapter ligation and index tagging
5. Normalization

Enzyme Fragmentation Library Prep
1. DNA extraction
2. DNA quantification
3. Tagmentation and index tagging
4. Normalization

Nextera DNA Flex
1. DNA extraction, tagmentation and index tagging, and normalization

See page 15 for details.

iSeq 100 Sequencing System

Your new lab partner

Makes next-generation sequencing easier and more affordable than ever

Workflow and applications

The iSeq 100 System is ideal for small whole-genome sequencing (eg, bacteria, viruses, plasmids), targeted sequencing of a set of genes or gene regions, gene expression analysis, or 16S metagenomics.

Prep

Prepare libraries for a range of targeted applications, including microbial sequencing and targeted resequencing.

Sequence

Start a run on the iSeq 100 System in less than 5 minutes. Sequencing is complete in 17.5 hours for a 2 × 150 bp run.

Analyze

Monitor and analyze sequencing runs using the on-instrument software and touchscreen interface.

Share

Interface with the iSeq 100 System from any web browser to review real-time data and performance metrics.

Applications and Methods

Gene expression analysis with targeted RNA-Seq

Targeted RNA-Seq focuses on specific transcripts of interest. It is used to analyze gene expression and identify fusion genes. mRNA sequencing (mRNA-Seq) detects known and novel transcripts and measures transcript abundance for accurate, comprehensive expression profiling.

16S rRNA sequencing

16S rRNA sequencing is a culture-free method for identifying and comparing bacteria from complex microbiomes or environments that are difficult to study.

Targeted gene panels

Targeted gene sequencing panels contain defined probe sets focused on specific genes of interest. Both predesigned and custom panels are available.

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See page 118 for details.

AmpliSeq for Illumina

Sequencing amplified

Highly accurate data with a fast, simple workflow

AmpliSeq for Illumina Workflow

Prepare high-quality libraries quickly and simply using a multiplexed PCR-based workflow.

Content selection

Library prep and accessories
5-7 hours total time
1.5 hours hands-on

Sequencing
17-32 hours

Data analysis
Time varies

Save time with ready-to-use AmpliSeq for Illumina panels

BRCA Panel
Targeted research panel investigating somatic and germline variants in *BRCA1* and *BRCA2*

Cancer Hotspot Panel v2
Targeted research panel investigating hotspot regions of 50 genes with known associations to cancer

Comprehensive Cancer Panel
Targeted panel investigating the exonic regions of 409 genes with known associations to cancer

Comprehensive Panel v3
Targeted DNA and RNA research panel investigating variants across 161 genes associated with a range of cancer types

Exome Panel
Targeted research panel for sequencing the protein-coding regions of the human genome

Immune Response Panel
Targeted RNA expression panel investigating 395 genes involved in tumor-immune system interactions

Transcriptome Human Gene Expression Panel
Targeted panel that measures expression levels of >20,000 human RefSeq genes

Choose your targets with custom panels

AmpliSeq for Illumina Custom Panels
Select up to 12,288 amplicons from any species and use with DNA, RNA, or degraded samples

See page 24 for details.

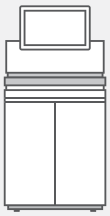
Methods

Whole-genome sequencing (large genomes)

Sequence complex genomes to make discoveries with confidence and fidelity

Key strengths

- Obtain a comprehensive picture of virtually any large genome
- Identify single nucleotide polymorphisms (SNPs) of interest and discover new variants
- Detect copy number and structural variants
- Assemble novel genomes
- Choose from a variety of library preparation kit options for multiple applications

Prepare library	Sequence	Analyze and Report
		
TruSeq DNA PCR-Free Library Prep Kit **TruSeq DNA Nano Library Prep Kit** **Nextera DNA Flex Library Prep Kit**	**NextSeq 550 System** **HiSeq X Series** **NovaSeq 6000 System**	**Available in BaseSpace Sequence Hub** Resequencing Oncology paired analysis Whole-genome phasing
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training TruSeq DNA PCR-Free Library Prep Kit Nextera DNA Flex Library Prep Kit	**Tiered service plans** NextSeq 550 System HiSeq X Series NovaSeq 6000 System	**Proof-of-Concept (POC) Service** Standard application functional testing with your samples **Sequencing Center Implementation Consulting (SCIC)** HiSeq X Series implementation **Validation Services** IQ/OQ/PQ[a]

a. IQ = Installation Qualification, OQ = Operational Qualification, PQ = Performance Qualification

Visit www.illumina.com/largewg for more details.

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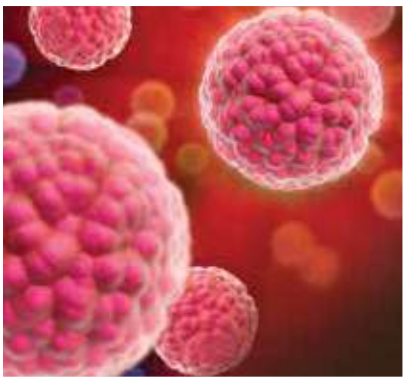
Innovation. Discovery. Application.



Tumor–normal studies (cancer research)
Tumor–normal studies determine the difference between tumor and non-tumor genomes. Researchers can employ whole-genome sequencing (WGS) to monitor genomic changes that occur in a tumor, including the presence of markers for good and poor prognosis.

Population genetics (genome-wide association studies [GWAS])
Population genetics is focused on understanding how allele frequencies differ between ethnicities or geographies for the study of genetic disease prevalence, population origins, and interpenetration of genomic data in general.

Plant research (agrigenomics)
Plant genomes present challenges to researchers because they are highly complex. Long-range sequencing generates long reads that allow scientists to produce more accurate genome assemblies in complex agricultural plant genomes.

Food supply (agrigenomics)
Exponential population growth and a changing climate are creating unique challenges for people working to maintain the food supply. Next-generation sequencing (NGS) is particularly useful in agricultural research, where genomes can be complex and prior knowledge of them can be scarce.

Variant detection (complex disease)
GWAS allows researchers to identify common genetic variants across populations to establish links between those variants and the traits of individuals, including predisposition to disease. Incorporating full 30× WGS into GWAS provides a comprehensive view of genetic variation and enables rare variant discovery.

STEP 1
Choose a library prep method



Product	TruSeq DNA PCR-Free Library Prep Kit	TruSeq DNA Nano Library Prep Kit	Nextera DNA Flex Library Prep Kit
Most important to me	Best[a] genome coverage	High coverage of complex genomes with low input	Simple and versatile sample-to-data workflow
Least important to me	Low DNA input quantity	Assay time	PCR-free workflow
Input quantity	1–2 µg DNA	100–200 ng DNA	1–500 ng DNA
Multiplexing	24–96 samples	24–96 samples	96 samples
Assay time	5 hrs	6 hrs	3–4 hrs
Hands-on time	~4 hrs	~5 hrs	1.5 hrs[b]
Target insert size	350 bp or 550 bp	350 bp or 550 bp	~350 bp
Fragmentation method	Mechanical	Mechanical	Enzymatic

a. Comparisons are within the Illumina WGS (large genomes) portfolio

b. Denotes total workflow time, including DNA extraction, library preparation, and library normalization/pooling steps

STEP 2
Choose a sequencer



Product	NextSeq 550 System		HiSeq X Series	NovaSeq 6000 System		
Most important to me	Convenience, rapid turn around time, and instrument affordability for large genome sequencing applications		$1000 human genome and throughput for population-scale large genome sequencing	Scalable throughput and flexibility for virtually any genome, sequencing method, and scale of project		
Run mode/kit type	Mid-output	High-output	–	S1 Xp	S2 Xp	S4 Xp
Samples/flow cell[a]	1	1	8–16	4	8	24

a. Assuming a 30× human genome

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STEP 3
Obtain results

Cancer research paired analysis ▼

Resequencing ▼

1 Align/Call variants

Tumor Normal B
- Detects somatic variants from a tumor and matched normal sample pair
- Generates a somatic report that includes quality, variants, and a Circos plot
- Works with TruSeq™ DNA PCR-Free, TruSeq DNA Nano, and Nextera DNA Flex Library Prep Kits

Isaac Whole Genome Sequencing B
- Performs alignment and small and large variant calling using algorithms developed by Illumina
- Offers 4× faster alignment speed with the same accuracy as the Burrows-Wheeler Aligner (BWA) Whole Genome Sequencing App

BWA Whole-Genome Sequencing B
- Analyzes whole-genome sequencing data using the Sanger BWA alignment algorithm combined with the Broad's Genome Analysis Tool Kit (GATK) small variant calling algorithm

2 Visualize

Integrative Genomics Viewer (IGV) B
- Enables visualization for the interactive exploration of large, integrated genomic data sets

3 Interpret/Report

BaseSpace Variant Interpreter B
- Enables rapid, rich annotation, filtering, and genomic data interpretation. Customize workflows and summarize findings into structured reports. Determine biological significance of genomic variants within a software framework focused on data security, compliance, and operational efficiency
- Leverage a comprehensive knowledge network of genomic content and leading annotation databases to confidently expedite the interpretation of variants into meaningful data

4 Biological Context

BaseSpace Cohort Analyzer B
- Analyzes complex human subject data for translational research applications on a web-based platform
- Provides access to a large repository of curated public data sets in the world and powerful tools for cohort analysis, and group comparisons of public and proprietary data

B BaseSpace Sequence Hub
Visit www.illumina.com/largewg for more details.

Library prep ordering information

TruSeq DNA PCR-Free Library Prep Kits

TruSeq DNA PCR-Free Library Prep Kits provide uniform coverage for whole-genome library prep for organisms ranging from bacteria to human. The kits offer shortened gel-free workflows, the ability to sequence the most challenging regions, and the power to identify a large number of variants. Libraries prepared with TruSeq DNA PCR-Free Library Prep Kits are compatible with all Illumina sequencing systems.

TruSeq DNA PCR-Free Library Prep Kits are available as single components, allowing users to take advantage of proven Illumina chemistry and use indexes from other sources, such as Integrated DNA Technologies (IDT). This also offers users the opportunity to increase assay plexity and enables accurate assignment of reads and more efficient use of flow cells. The library preparation protocol requires one kit from each component, depending on sample requirements.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
TruSeq DNA PCR-Free Low-Throughput Library Prep Kit		24	20015962
TruSeq DNA PCR-Free High-Throughput Library Prep Kit		96	20015963
Index Adapter Component			
TruSeq DNA Single Indexes Set A	12	24	20015960
TruSeq DNA Single Indexes Set B	12	24	20015961
TruSeq DNA CD Indexes	96	96	20015949
IDT for Illumina - TruSeq DNA UD Indexes	24	96	20020590
IDT for Illumina - TruSeq DNA UD Indexes	96	96	20022370
Training			
TruSeq DNA PCR-Free Library Prep Kit training at customer site			TR-204-0011

TruSeq DNA Nano Library Prep Kit

Preserve precious samples with the TruSeq DNA Nano Library Prep Kit. Prepare sequencing libraries for low- or high-throughput studies from as little as 100 ng of input DNA in less than a day. Bead-based selection reduces the sample loss associated with gel-based selection. This kit is designed to match the ever-increasing read lengths of Illumina sequencing instruments and is compatible with all Illumina sequencing systems.

TruSeq DNA Nano Library Prep Kits are available as single components, allowing users to take advantage of proven Illumina chemistry and use indexes from other sources, such as Integrated DNA Technologies (IDT). This also offers users the opportunity to increase assay plexity and enables accurate assignment of reads and more efficient use of flow cells. The library preparation protocol requires one kit from each component, depending on sample requirements.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
TruSeq DNA Nano Library Prep Kit		24	20015964
TruSeq DNA Nano Library Prep Kit		96	20015965
Index Adapter Component			
TruSeq DNA Single Indexes Set A	12	24	20015960
TruSeq DNA Single Indexes Set B	12	24	20015961
TruSeq DNA CD Indexes	96	96	20015949
IDT for Illumina - TruSeq DNA UD Indexes	24	96	20020590
IDT for Illumina - TruSeq DNA UD Indexes	96	96	20022370

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Nextera DNA Flex Library Prep Kits

Save time and reduce hands-on touch points with On-Bead Tagmentation, producing sequencing-ready libraries in less than three hours. Simplify daily operations with a kit that supports a broad DNA input range (1–500 ng), multiple DNA input types, and flexible for use with small (bacteria, archaea, viruses, and plasmids) to large genomes (human, plant, mouse). Libraries prepared with Nextera DNA Flex Library Prep Kits are compatible with all Illumina sequencing systems.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Flex Lysis Reagent Kit			20018706
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

Proof-of-Concept (POC) Service

POC Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at the Illumina Service Lab, and in-person consultation post-POC to review sequencing run quality control (QC), data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

Contact your local representative to learn more about Illumina products and services available in your region.
800.809.4566 (North America) • 01799 534332 toll-free (Europe, Middle East, Africa) • +61.3.9212.9900 (Australia)
+65.6773.0188 (Singapore) • +81.3.4578.2800 (Japan) • +86.21.6032.1066 (China)

See page 146 of the Services section for Illumina instrument service plan options.

Whole-genome sequencing (small genomes)

Sequence virtually any small genome and make new discoveries with enormous scalability at high throughput

Key strengths

- Sequence thousands of organisms in parallel
- Provide comprehensive analysis of 20 Mb or smaller genomes
- Discover new biomarkers (SNP variants) within a microbial/viral sample

Prepare library	Sequence	Analyze and Report
		
Nextera XT DNA Library Prep Kit **Nextera DNA Flex Library Prep Kit**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available in Local Run Manager Software (MiSeq System/MiniSeq System)** Resequencing **Available in BaseSpace Sequence Hub** Resequencing *De novo* assembly
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Nextera XT Library Prep Kit Nextera DNA Flex Library Prep Kit	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/smallwg for more details.

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Risk monitoring (biopharmaceutical development)
Risk monitoring is a key aspect of biopharmaceutical research and product development. WGS allows researchers to closely track cell line stability and conduct biosafety monitoring.

Effective screening (public health and epidemiology)
Microbiologists often need to screen food and agricultural samples to identify and confirm the presence of specific microbes. Researchers use WGS to identify the bacterial genomes of pathogens, increasing the accuracy of microbial testing.

Bacterial evolution (public health and epidemiology)
Bacterial species change over time through various mechanisms, such as horizontal gene transfer and coevolution with other organisms. High-resolution gene sequencing can inform a wide range of studies, including mutagenesis, directed evolution, the spatial and temporal dynamics of epidemics, and disease transmission.

Disease surveillance (public health and epidemiology)
Ongoing epidemiological and genomic surveillance is key to identifying determinants of transmission and monitoring microbe changes and adaptations over time and geography. With WGS, scientists gain valuable insights into viral or bacterial variants that develop over the course of disease outbreaks.

STEP 1
Choose a library prep method

Product	Nextera XT DNA Library Prep Kit	Nextera DNA Flex Library Prep Kit
Most important to me	Fast workflow with low input for small genomes	Simple and versatile sample-to-data workflow
Least important to me	Amplicons < 300 bp in length	PCR-free workflow
Input quantity	1 ng DNA	1–500 ng DNA
Multiplexing	384 samples	96 samples
Assay time	90 min	3–4 hrs
Hands-on time	15 min	1.5 hrs[a]
Target insert size	300 bp–1.5 kb	~350 bp
Fragmentation method	Enzymatic	Enzymatic
Design[b]	Independently designed customer probes target amplicons that are at least 300 bp in length	–

a. Denotes total workflow time, including DNA extraction, library preparation, and library normalization/pooling steps
b. Nextera XT DNA Library Prep Kit can be used to create libraries based on in-house–developed amplicon solutions

STEP 2
Choose a sequencer

Product	iSeq 100 System	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System (S1 flow cell)
Most important to me	Affordability and efficiency	Simplicity and instrument affordability	Longest read lengths in Illumina portfolio,[a] caters well to microbiology applications	Capacity for whole genome metagenomics and production power for small genome sequencing applications	Production power with lowest cost per sample[a]
Onboard informatics	●	●	●	–	–
***E. coli* genome samples processed/flow cell**	1–4	1–96	1–96	1–384	96–768
Run mode/kit type	Standard	Mid-output/ High-output	Research mode and Dx	Mid-output/ High-output	S1
Flow cells processed/ run	1	1	1	1	1 or 2

a. Comparison among Illumina sequencing portfolio

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STEP 3
Obtain results

Resequencing ▼

1

Align/Call variants

BWA* Whole Genome Sequencing B
- Analyzes WGS data using the Sanger BWA alignment algorithm combined with the Broad's GATK* small variant calling algorithm

*BWA: Burrows-Wheeler Aligner; GATK: Genome Analysis Tool Kit

De novo assembly ▼

Assemble

Assembly workflow L
- Assembles small genomes (< 20 Mb) and best suits the assembly of bacterial genomes, such as *E. coli*
- Uses the EMBL Velvet algorithm and writes assembly results in FASTA format

SPAdes Genome Assembler B
- Assembles genomes from standard bacterial isolates and single-cell multiple displacement amplification (MDA) preparations

Velvet *de novo* Assembly B
- Assembles *de novo* genomes for bacterial samples

2

Analyze

SEAR: Antibiotic Resistance B
- Constructs full-length, horizontally acquired antimicrobial resistance genes (ARGs) from sequencing data sets
- Leverages environmental metagenomics and microbiome experiments

Annotate/Filter

Prokka Genome Annotation B
- Rapidly annotates genes and identifies coding sequences in prokaryotic genomes
- Does not annotate eukaryotic genomes

3

Visualize

The Integrative Genomics Viewer (IGV) B
- Powerfully displays NGS data

B BaseSpace Sequence Hub L Local Run Manager Software
Visit www.illumina.com/smallwg for more details.

Library prep ordering information

Nextera XT DNA Library Prep Kits

Generate sequencing-ready libraries from small genomes (bacteria, archaea, viruses), amplicons, and plasmids in less than 90 minutes with only 15 minutes of hands-on time. Nextera XT DNA Library Prep Kits simultaneously fragment input DNA and tag the fragments with sequencing adapters in a single-tube enzymatic reaction. Nextera XT DNA Library Prep Kits require as little as 1 ng of input, supporting a wide array of sample types. Bead-based normalization eliminates the need for library quantification before pooling and sequencing. Libraries prepared with Nextera XT Kits are compatible with all Illumina sequencing systems.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera XT DNA Library Prep Kit		24	FC-131-1024
Nextera XT DNA Library Prep Kit		96	FC-131-1096
Nextera XT Index Kit	24	96	FC-131-1001
TruSeq Dual Index Sequencing Primer Kit, single read	Single-use kit		FC-121-1003
TruSeq Dual Index Sequencing Primer Kit, paired-end read	Single-use kit		PE-121-1003
Nextera XT Index Kit v2, set A	96	384	FC-131-2001
Nextera XT Index Kit v2, set B	96	384	FC-131-2002
Nextera XT Index Kit v2, set C	96	384	FC-131-2003
Nextera XT Index Kit v2, set D	96	384	FC-131-2004
Training			
Nextera XT DNA Library Prep Kit training at customer site			TR-204-0009

Nextera DNA Flex Library Prep Kits

Save time and reduce hands-on touch points with On-Bead Tagmentation, producing sequencing-ready libraries in less than three hours. Simplify daily operations with a kit that supports a broad DNA input range (1–500 ng), multiple DNA input types, and flexible for use with small (bacteria, archaea, viruses, and plasmids) to large genomes (human, plant, mouse). Libraries prepared with Nextera DNA Flex Library Prep Kits are compatible with all Illumina sequencing systems.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Flex Lysis Reagent Kit			20018706
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

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Proof-of-Concept Service

The POC Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at the Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
iSeq 100 POC Service	20023613
MiniSeq POC Service	20003924
MiSeq POC Service	SP-801-1002
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

Cancer research panels

Reduce cost per sample and analysis burden with highly focused targeted panels that enable deeper coverage of regions of interest

Key strengths

- Analyze key genes or regions of interest to high depth using predesigned, analytically validated panels
- Analyze cancer-relevant genes cost effectively
- Obtain highly accurate results with variants present down to 5% variant allele frequency at 95% sensitivity and specificity

Prepare library	Sequence	Analyze and Report
		
TruSight Sequencing Cancer Panel Tumor 15 Panel Myeloid Panel RNA Pan-Cancer Panel Tumor 170 Panel **AmpliSeq for Illumina** *BRCA* Panel Cancer Hotspot Panel v2 Comprehensive Cancer Panel Comprehensive Panel v3 Focus Panel Immune Response Panel	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available in Local Run Manager Software and BaseSpace Sequence Hub**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training TruSight Tumor 15 Panel TruSight Tumor 170 Panel TruSight Cancer Panel TruSight Myeloid Panel AmpliSeq Library Prep	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof of Concept Service** Standard application functional testing with your samples Cancer Research Bioinformatics Service **Validation Services** IQ/OQ/IPV[a]

a. IQ = Installation Qualification, OQ = Operational Qualification, IPV = Instrument Performance Verification

Visit www.illumina.com/products/by-area/oncology.html for more details.

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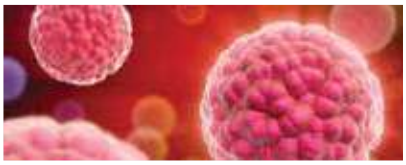
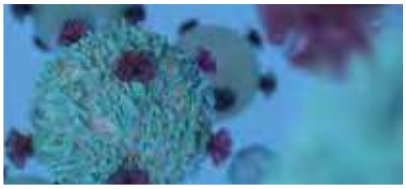


Mutation and somatic variation detection

Cancer researchers often sequence key genes or regions of interest to high depth using predesigned, analytically validated targeted panels. With NGS, scientists can assess multiple genes in a single test with a single streamlined workflow, a short time-to-answer, and accurate and highly sensitive results.



Cancer immunotherapy research

Immuno-oncology is an emerging field that has taken great strides in the fight against cancer, bolstered by a refined understanding of how tumors evade the natural immune response. Leading immuno-oncology researchers are leveraging NGS to discover biomarkers and apply genomics to personalized immunotherapy.

Tumor–normal studies

Cancer whole-genome sequencing (WGS) provides a base-by-base view of the unique mutations present in cancer tissue. It enables discovery of novel cancer-associated variants, including single nucleotide variants (SNVs), copy number changes, and structural variants. By comparing tumor and normal DNA, WGS can also provide a comprehensive view of changes to a specific tumor sample.

Liquid biopsy studies

Cell-free, circulating tumor DNA (ctDNA) can act as a noninvasive cancer biomarker, offering a potential alternative to invasive tissue biopsies. Today, researchers are investigating the use of ctDNA as a biomarker for detecting the presence of tumors in “liquid biopsies” obtained through a simple blood draw.

STEP 1
Choose a library prep method

TRUSIGHT PANELS—AMPLICON-BASED LIBRARY PREP KITS

Product	TruSight Tumor 15 Panel	TruSight Myeloid Panel
Key use	Somatic mutation detection in solid tumors; includes additional QC and analysis support	Somatic mutation detection in myeloid malignancies
DNA input	20 ng	50 ng
FFPE compatibility	Yes	N/A
Genomic content	44 kb (250 amplicons, 15 genes)	~141 kb (568 amplicons, 54 genes)
Read length	2 × 150 bp	2 × 150 bp
Sequencing depth	> 93.5% amplicons at minimum 500×	> 500×
Ideal instrument	MiniSeq System, MiSeq System, NextSeq 550 System	MiniSeq System, MiSeq System, NextSeq 550 System

AMPLISEQ FOR ILLUMINA PANELS—AMPLICON-BASED LIBRARY PREP KITS

Product	AmpliSeq for Illumina *BRCA* Panel	AmpliSeq for Illumina Cancer HotSpot Panel v2	AmpliSeq for Illumina Focus Panel	AmpliSeq for Illumina Comprehensive Panel v3	AmpliSeq for Illumina Immune Response Panel	AmpliSeq for Illumina Comprehensive Cancer Panel
Key use	Analysis of solid breast tumors or germline *BRCA* mutations	Analysis of mutations in common oncogenes	Analysis of genes with known relevance to solid tumors in DNA and RNA	Comprehensive analysis of cancer aberrations in DNA and RNA	RNA panel for analysis of immune cell expression in solid tumor	Comprehensive analysis of cancer aberrations in DNA only
Input	10 ng DNA	10 ng DNA	20 ng DNA/RNA	40 ng DNA/RNA	20 ng RNA/pool	40 ng DNA
FFPE compatibility	Yes	Yes	Yes	Yes	Yes	Yes
Genomic content	2 genes (150 amplicons)	50 gene (207 amplicons)	52 genes (28,783 amplicons)	161 genes (3781 amplicons)	395 genes (395 amplicons)	409 genes (15,992 amplicons)
Read length	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp
Sequencing depth	90× Germline 500× Somatic	950×	1000× DNA 525× RNA	1650× DNA 350× RNA	1M Reads	1660×
Ideal instrument	iSeq 100 System, MiniSeq System, MiSeq System	iSeq 100 System, MiniSeq System, MiSeq System	iSeq 100 System, MiniSeq System, MiSeq System	NextSeq 550 System	NextSeq 550 System	NextSeq 550 System

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TRUSIGHT PANELS—ENRICHMENT-BASED SEQUENCING KITS

Product	TruSight Cancer Panel	TruSight RNA Pan-Cancer Panel	TruSight RNA Fusion Panel	TruSight Tumor 170 RNA/DNA Panel
Key use	Germline mutation detection	Expression, variants, and fusions in RNA	Analysis of RNA Fusions	Analysis of RNA and DNA variants in solid tumors in a single assay
Input	50 ng DNA	10–20 ng RNA	10 ng RNA	40 ng DNA/RNA
FFPE compatibility	No	Yes	Yes	Yes
Genomic content	255 kb (~4000 probes, 94 genes)	1385 genes 57K probes	507 genes 21,283 probes	170 genes 533 kb DNA 358 kb RNA
Assay	Enrichment	cDNA-mediated enrichment	cDNA-mediated enrichment	Enrichment
Read length	2 × 150 bp	2 × 75 bp	2 × 75 bp	2 × 101 bp
Sequencing depth	750× mean coverage	3M reads/sample	3M reads/sample	50M reads/sample
Ideal instrument	MiniSeq System, MiSeq System, NextSeq 550 System	MiniSeq System, MiSeq System, NextSeq 550 System	MiniSeq System, MiSeq System, NextSeq 550 System	NextSeq 550 System, NovaSeq 6000 System,[a] HiSeq 2500 System[b]
UMI Tool Kit compatible	–	–	–	●

a. The TruSight Tumor 170 Panel has not been validated by Illumina on the NovaSeq 6000 System

b. The HiSeq 2500 System is no longer available for purchase; consult with your local service lab regarding system availability

STEP 2
Choose a sequencer





Product	iSeq 100 System	MiniSeq System		MiSeq System		NextSeq 550 System		NovaSeq 6000 System		
Most important to me	Affordability, small footprint, onboard analysis	Onboard analysis, instrument affordability, simplicity		Translational research with multiple run mode options[a]		Ability to run large panels, cytogenetic arrays, and highest[b] bench-top instrument sample throughput		Highest[b] sample throughput, benchtop simplicity, and low cost per sample (base)		
Onboard informatics	●	●		●		–		–		
Production scale for human and nonhuman samples	–	–		–		–		●		
Run mode/kit type	Standard	Mid-output	High-output	Nano	High-output	Mid-output	High-output	S1[c]	S2[c]	S4[c]
Number of panels processed per flow cell										
TruSight Tumor 170	–	–	–	–	–	–	8	32[d]	32[d]	32[d]
TruSight Tumor 15 Panel	4	8	2	–	8	24	–	–	–	–
TruSight Cancer	6	12	24	12	24	96	96	–	–	–
TruSight RNA Pan-Cancer Panel	4	8	24	–	8	24	24	48[d]	–	–
TruSight Myeloid Panel	1	2	8	–	8	40	96	–	–	–
TruSight RNA Fusion	–	–	8	–	8	24	–	48[d]	–	–
AmpliSeq for Illumina *BRCA* Panel	96 germline[d] 12 somatic	96 germline[d] 24 somatic	–	–	96 germline[d] 80 somatic	–	–	–	–	–
AmpliSeq for Illumina Cancer Hotspot Panel v2	16	32	–	96	–	–	–	–	–	–
AmpliSeq for Illumina Comprehensive Cancer Panel	–	–	–	–	–	12	–	52	104	312
AmpliSeq for Illumina Comprehensive Panel v3	–	1	–	3	–	48[d,e]	–	192[d,e]	–	–
AmpliSeq for Illumina Focus Panel	8[e]	16	–	48[e]	–	–	–	–	–	–
AmpliSeq for Illumina Immune Response Panel	–	8	24	–	24	96	–	192	–	–

a. See page 122 in the Instruments chapter to learn more
b. Comparisons among Illumina sequencing portfolio
c. Values supplied represent sequencing performed using the NovaSeq Xp workflow; the standard workflow will have lower throughput values
d. Limited by indexing
e. Combined DNA and RNA libraries

STEP 3
Obtain results

Targeted resequencing ▼

1

Align/Call variants

Local Run Manager (LRM) RNA Fusion Module (RNA Fusion)
- Analyzes RNA Fusions on the MiSeq system with Local Run Manager

AmpliSeq for Illumina Panels B
- Local Run Manager software for on-instrument analysis
- Rapid secondary analysis using the DNA Amplicon or RNA Amplicon BaseSpace Apps

TruSight™ Tumor 15 Analysis App B
- Rapid analysis of TruSight Tumor 15 amplicon sequencing data
- Output is compatible with downstream analysis and reporting applications

TruSight Tumor 170 App
- Rapid analysis of TruSight Tumor 170 sequencing data
- Output is compatible with downstream analysis and reporting applications

Enrichment App
- Rapid analysis the TruSight Cancer Panel data
- Combines the Sanger BWA alignment algorithm with the Broad's GATK small-variant calling algorithm

RNA-Seq Alignment App B
- Read mapping using the TopHat 2 (Bowtie 1), TopHat 2 (Bowtie 2) or STAR aligner
- FPKM estimation of reference genes and transcripts using Cufflinks 2
- Assembly of novel transcripts with Cufflinks 2
- Variant calling (SNVs and small indels) with the Isaac Variant caller
- Optional fusion calling with TopHat-Fusion when TopHat 2 (Bowtie 1) is used or Manta when STAR is used

2

Interpret/Report

BaseSpace Variant Interpreter B
- Perform rapid, rich annotation, filtering, and interpretation of genomic data. Customize workflows and summarize findings into structured reports. Determine biological significance of genomic variants within a software framework focused on data security, compliance, and operational efficiency
- Expedite interpretation of variants into meaningful data with the BaseSpace Knowledge Network containing genomic interpretation content and leading annotation databases to confidently

3

Add biological context

BaseSpace Cohort Analyzer
- Analyzes complex human subject data for translational research applications on a web-based platform
- Provides access to a large repository of curated public data sets in the world and powerful tools for cohort analysis and group comparisons of public and proprietary data

B BaseSpace Sequence Hub L Local Run Manager Software
Visit www.illumina.com/products/by-area/oncology.html for more details.

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Library prep ordering information

TruSight Tumor 170

TruSight Tumor 170, a next-generation sequencing assay designed to cover 170 genes associated with common solid tumors, is an enrichment-based targeted panel that simultaneously analyzes DNA and RNA, covering a wide range of genes and variant types. The comprehensive nature provides laboratories with a deep view into the genetics of cancer.

- Comprehensive coverage of cancer-related variants
  - Assessment of fusions, splice variants, insertions/deletions, single nucleotide variants (SNVs), and amplifications in one assay using DNA and RNA creates efficiencies in sample usage, time, and cost
- Accurate results from low-quality samples
  - Variant detection with as little as 40 ng DNA and RNA input, and as low as 5% mutant allele frequency, maximizes the results from precious formalin-fixed paraffin-embedded (FFPE) samples
- Integrated, streamlined workflow
  - DNA and RNA are prepared in parallel with an integrated workflow following DNA shearing/cDNA synthesis

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight Tumor 170 NextSeq Bundle	16	24	OP-101-1003
TruSight Tumor 170 Library Prep Kit	16	24	OP-101-1004

TruSight RNA Fusion

The targeted TruSight RNA Fusion Panel is a cost-effective solution for detecting gene fusions in multiple cancer types, regardless of origin. Covering 507 fusion-associated genes, a single assay enables researchers to assess most known cancer-related fusions in blood, bone marrow, and FFPE samples and identify novel fusion gene partners. This assay accommodates input amounts as little as 20 ng FFPE RNA or 10 ng fresh-frozen total RNA. The TruSight RNA Fusion Panel provides a sensitive, reproducible, and economical solution for studies of gene fusions in cancer research.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight RNA Fusion Library Prep Kit Set A	12	48	FC-304-1002
TruSight RNA Fusion Library Prep Kit Set B	12	48	RS-304-1003

TruSight Tumor 15

The TruSight Tumor 15 Panel features a comprehensive workflow that simplifies NGS, enabling easy integration into existing lab practices. This panel evaluates 15 significant solid tumor genes in a single assay, offering a more efficient approach to tumor profiling than single-gene testing. Libraries prepared with the TruSight Tumor 15 Panel are optimized for sequencing on the iSeq 100 System, MiniSeq System, and MiSeq System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight Tumor MiSeq 15 Kit	24	24	OP-101-1001
TruSight Tumor 15 Panel with library prep reagents only	24	24	OP-101-1002
TruSight Tumor 15 MiniSeq Kit	12	24	20005610
MiniSeq High Throughput sequencing reagents			MS-102-3003

See Services section on page 146 for Illumina instrument service plan options.
Select Illumina reagents and consumables are available with product attributes that might benefit clinical research laboratories. See page 87 for details.

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TruSight RNA Pan-Cancer

The TruSight RNA Pan-Cancer Panel provides a comprehensive analysis of the cancer transcriptome. Targeting 1385 cancer-related transcripts and genes known to be involved in gene fusions, the TruSight RNA Pan-Cancer Panel enables analysis of cancer samples, including FFPE tissues. It offers the following capabilities:

- Gene expression information, variant calling, and fusion detection with known and novel gene fusion partners
- Optimized, low-input protocol for a wide range of sample types, including FFPE tissues
- Comprehensive view of cancer pathways
- Economical RNA-Seq on a benchtop sequencer

Libraries prepared with the TruSight RNA Pan-Cancer Sequencing Panel are compatible with all Illumina Sequencing Systems.

Ordering information

Product	No. of samples	Catalog no.
TruSight RNA Pan-Cancer Panel, set A	48	RS-303-1002
TruSight RNA Pan-Cancer Panel, set B	48	RS-303-1003

TruSight Myeloid

The TruSight Myeloid Sequencing Panel covers 15 full genes (exons only) and key exonic regions of 39 additional genes, providing a comprehensive assessment of the key genes involved in myeloid malignancies in a single test. The result is an accurate, cost-effective solution for profiling liquid tumors. Libraries prepared with the TruSight Myeloid Sequencing Panel are compatible with the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight Myeloid Sequencing Panel*		96	FC-130-1010
TruSeq Custom Amplicon Index Kit	96	384	FC-130-1003
TruSeq Index Plate Fixture Kit			FC-130-1005
TruSeq Index Plate Fixture and Collar Kit (2 each)			FC-130-1007
Training			
TruSight Myeloid Panel training at customer site			TR-204-0023

*Requires the purchase of a TruSeq Custom Amplicon Index Kit

TruSight Cancer

The TruSight Cancer Sequencing Panel includes genes associated with common and rare cancers, and 284 SNPs found to correlate with cancer through genome-wide association studies (GWAS). The TruSight Cancer Sequencing Panel provides custom oligos targeting identified regions of interest. The panel includes enough product for four enrichment reactions (48 samples) and works with TruSight Rapid Capture Kits. Libraries prepared with the TruSight Cancer Sequencing Panel are compatible with the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	Pooling plexity	No. of indexes	No. of samples	Catalog no.
TruSight Cancer Sequencing Panel (4 enrichments)[a]			48	FC-121-0202
TruSight Rapid Capture Kits	1	1	8	FC-140-1101
	2	2	8	FC-140-1102
	4	4	16	FC-140-1103
	12	24	48	FC-140-1104
	12	24	96	FC-140-1105
	12	96	288	FC-140-1106
Training				
TruSight Rapid Capture Library Prep Panel training at customer site				TR-204-0016

a. Requires the purchase of a TruSight Rapid Capture Kit

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TruSight Oncology UMI Reagents

The TruSight Oncology UMI Reagents reduce background noise in sequencing data, enabling detection of low-frequency variants, such as those found in cell-free DNA (cfDNA). Circulating tumor DNA (ctDNA) may represent a very small fraction of cfDNA, near the limit of detection for NGS. The TruSight Oncology UMI Reagents address this challenge with unique molecular identifiers (UMIs) and error correction software, reducing error rates to < 0.007% and enabling detection of low-frequency variants. Lower error rates increase analytical specificity, resulting in higher confidence in NGS data.

The TruSight Oncology UMI Reagents include UMI adapters and indexes, plus TruSight Oncology DNA library prep and enrichment reagents. Integration of UMIs does not create any extra steps in the library prep workflow and libraries prepared with these reagents are best sequenced on HiSeq or NovaSeq systems due to the high coverage requirements. The UMI Error Correction App aligns reads, then uses UMIs to exclude false positives, reducing variant calling errors. The UMI Error Correction App is available in the cloud-based BaseSpace Sequence Hub or for local installation.

Ordering information

Product	No. of samples	Catalog no.
TruSight Oncology UMI Reagents	48	20024586

AmpliSeq for Illumina *BRCA* Panel

The AmpliSeq for Illumina *BRCA* Panel is a targeted resequencing assay designed for detecting somatic and germline mutations across all exonic regions and the flanking intronic sequences of *BRCA1* and *BRCA2*. It 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. Libraries prepared with the AmpliSeq for Illumina *BRCA* Panel are most compatible with the iSeq 100 System, MiniSeq System, and MiSeq System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina *BRCA* Panel		24	20019168
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

AmpliSeq for Illumina Cancer Hotspot Panel v2

The AmpliSeq for Illumina Cancer Hotspot Panel v2 targets ~2800 mutations in the hotspot regions of 50 genes with known associations to cancer. It 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. Libraries prepared with the AmpliSeq for Illumina Cancer Hotspot Panel v2 are most compatible with the iSeq 100 System, MiniSeq System, and MiSeq System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Cancer Hotspot Panel v2		24	20019161
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

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AmpliSeq for Illumina Comprehensive Cancer Panel

The AmpliSeq for Illumina Comprehensive Cancer Panel provides a targeted resequencing solution for analyzing somatic mutations across 409 genes with known associations to multiple cancer types, including lung, colon, breast, ovarian, melanoma, and prostate. It 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. Libraries prepared with the AmpliSeq for Illumina Comprehensive Cancer Panel are most compatible with the NextSeq 550 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Comprehensive Cancer Panel		24	20019160
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

AmpliSeq for Illumina Comprehensive Panel v3

The AmpliSeq for Illumina Comprehensive Panel v3 offers coverage of 161 cancer-associated genes, including kinases and genes involved in DNA repair. The panel content spans hotspots, full-length genes, copy number variations, intergenic gene fusions, and intragenic rearrangements. It is part of a DNA/RNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Libraries prepared with the AmpliSeq for Illumina Comprehensive Panel v3 are most compatible with the MiniSeq System, MiSeq System, and NextSeq 550 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Comprehensive Panel v3		24	20019109
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

AmpliSeq for Illumina Focus Panel

The AmpliSeq for Illumina Focus Panel targets hundreds of mutations across 52 key genes associated with solid tumors. Using the Focus Panel, researchers can analyze both DNA and RNA concurrently. It is part of a DNA/RNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Libraries prepared with the AmpliSeq for Illumina Focus Panel are most compatible with the iSeq 100 System, MiniSeq System, and MiSeq System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Focus Panel		24	20019164
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

AmpliSeq for Illumina Immune Response Panel

The AmpliSeq for Illumina Immune Response Panel is a targeted resequencing assay for quantitating expression of cancer biomarkers in 395 genes involved in tumor–immune system interactions. It is part of an RNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Libraries prepared with the AmpliSeq for Illumina Immune Response Panel are most compatible with the MiniSeq System, MiSeq System, and NextSeq 550 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Immune Response Panel		24	20019169
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105

MiSeq Reporter Software

Streamlined, automated onboard data analysis workflows

MiSeq Reporter Software is the onboard data analysis tool provided with all MiSeq Systems. The simple and intuitive graphical interface eases primary and secondary data analysis, and helps ensure quality coverage information for each sample.

- Load the sample and walk away
- Automate onboard data analysis and reporting
- Perform data analysis from your desk
- Generate FASTQ files from all workflows
- Stream data to the BaseSpace Platform to use powerful Core Apps

Contact your local representative to learn more about Illumina products and services available in your region.
800.809.4566 (North America) • 01799 534332 toll-free (Europe, Middle East, Africa) • +61.3.9212.9900 (Australia)
+65.6773.0188 (Singapore) • +81.3.4578.2800 (Japan) • +86.21.6032.1066 (China)

See page 146 of the Services section for Illumina instrument service plan options.

Genetic conditions sequencing panels

Target genes and regions relevant to particular diseases or conditions with focused panels

Key strengths

- Efficient detection of rare variants that might be associated with genetic diseases
- Confident identification of causative or inherited rare mutations in a single test
- Cost-effective, streamlined, targeted NGS of specific genetic diseases or conditions

Prepare library

AmpliSeq for Illumina Panels
AmpliSeq for Illumina Custom Panels

AmpliSeq for Illumina On-Demand Panels

TruSight Sequencing Panels
TruSight One Panel

TruSight One Expanded

TruSight Cardio Panel

TruSight Rapid Capture Enrichment Kit

Sequence

iSeq 100 System
MiniSeq System
MiSeq System
MiSeqDx System
NextSeq 550 System
NextSeq 550Dx System
NovaSeq 6000 System

See page 36 to learn how many samples can be run on each sequencing platform.

Analyze and Report

Available on Local Run Manager Software, BaseSpace Sequence Hub
Targeted resequencing

Illumina Informatics Solutions
BaseSpace Sequence Hub
Run management | Analysis | Reporting

Training

Customer site training
TruSight Rapid Capture Kit

Library prep with MiSeq System

Service contracts

Tiered service plans
iSeq 100 System

MiniSeq System

MiSeq System

NextSeq 550 System

NovaSeq 6000 System

Professional services

Proof-of-Concept Service
Standard application functional testing with your samples

Validation Services
IQ/OQ/PQ[a]

a. IQ = Installation Qualification, OQ = Operational Qualification, PQ = Performance Qualification

Visit www.illumina.com/diseasepanels for more details.

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Innovation. Discovery. Application.

Cardiac research (genetic conditions research)
Complex and inherited genetic factors often play a role in cardiac conditions. The TruSight Cardio Panel allows researchers to affordably and accurately identify gene variants that have been linked to disease within 174 genes related to 17 inherited cardiac conditions (ICCs).

Genetic disease research (genetic conditions research)
Inherited diseases can affect children and specific ethnic groups. Targeted sequencing with panels provides insight into the genetic basis of inherited genetic conditions.

Rare genetic disease research (genetic conditions research)
Severe recessive diseases can be the result of rare variants within genes. Targeted sequencing is a simple, comprehensive way to identify these rare variants and increase our understanding of their role in disease onset and progression.

STEP 1
Choose a sequencing panel

Product	Maximum number of samples per flow cell at optimal kit configuration							
	Genomic content covered	Recommended read length	MiniSeq System	MiSeq System v2 chemistry[a]	MiSeq System v3 chemistry	NextSeq 550 System mid-output	NextSeq 550 System high-output	NovaSeq 6000 System
TruSight One Expanded	16.5 Mb	2 × 150 bp	–	–	–	–	24	288
TruSight One v2 MiSeq Combo Kit	12 Mb	2 × 150 bp	–	–	3	–	–	–
TruSight One Panel	12 Mb	2 × 150 bp	3	–	3	12	36	288
TruSight Cardio Panel	244 kb	2 × 150 bp	12	12	–	48	–	–
Contact your local account representative for information on AmpliSeq for Illumina Custom Panels and AmpliSeq for Illumina On-Demand Panels								

a. Limited by indexes

STEP 2
Choose a sequencer





Product	iSeq 100 System	MiniSeq System	MiSeq Series	NextSeq 550 Series	NovaSeq 6000 System
Most important to me	Affordability	Onboard analysis, instrument affordability, simplicity	Translational research with Dx option	Ability to run large panels and highest[1] bench-top instrument sample throughput with Dx option	Highest[a] sample throughput, benchtop simplicity, and low cost per sample (base)
Onboard informatics	●	●	●	–	–
Run mode/kit type	Standard	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	S1, S2, S4
Flow cells processed/ run	1	1	1	1	1 or 2

a. Comparisons among Illumina sequencing portfolio

Visit www.illumina.com/diseasepanels for more details.

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STEP 3
Obtain results

Targeted resequencing ▼

1

Align/Call variants

Enrichment workflow L
- Enables analysis of panels using capture-based methods, such as the Nextera DNA Exome, the TruSight One Panels, and Illumina custom capture panels built in DesignStudio™ Software
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm

Isaac Enrichment B
- Enables rapid analysis of panels using capture-based methods, such as the Nextera DNA Exome, the TruSight One Panels, and Illumina custom capture panels built in DesignStudio Software
- Performs alignment, and small- and large-variant calling using algorithms developed by Illumina
- Offers 4× faster alignment speed with the same accuracy as the BWA Enrichment App
- Provides multilaunch capabilities that allow simultaneous analysis of 96 samples

BWA Enrichment B
- Enables rapid analysis of panels using capture-based methods, such as the Nextera DNA Exome, the TruSight One Panels, and Illumina custom capture panels built in DesignStudio Software
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm
- Provides multilaunch capabilities that allow simultaneous analysis of 96 samples

2

Interpret/Report

BaseSpace Variant Interpreter B
- Enables rapid, rich annotation, filtering, and interpretation of genomic data. Customize workflows and summarize findings into structured reports. Determine biological significance of genomic variants within a software framework focused on data security, compliance, and operational efficiency
- Leverages BaseSpace Knowledge Network containing genomic interpretation content and leading annotation databases to confidently expedite interpretation of variants into meaningful data

3

Add biological context

BaseSpace Cohort Analyzer
- Analyzes complex human subject data for translational research applications on a web-based platform
- Provides access to the largest repository of curated public data sets in the world and powerful tools for cohort analysis and group comparisons of both public and proprietary data

B BaseSpace Sequence Hub L Local Run Manager Software

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Targeted panel ordering information

AmpliSeq for Illumina Custom Panels

AmpliSeq for Illumina Custom Panels enable users to target specific regions of genomic target using content that may not be available in ready-to-use panels. Custom content can be easily designed and ordered online through DesignStudio Software. Targets can be selected based on several preloaded reference genomes. An optimized algorithm automatically designs amplicons based on chosen target candidates. Libraries prepared with the AmpliSeq for Illumina Custom Panel are most compatible with the iSeq 100 System, MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Order online at www.illumina.com/products/by-brand/ampliseq/custom-panels.html

AmpliSeq for Illumina On-Demand Panels

AmpliSeq for Illumina On-Demand Panels provide a choice of > 5000 pretested genes with known content relevant for inherited disease research, including hereditary cancer, primary immunodeficiency, hearing loss, muscular dystrophy, and more. Easily design and order an on-demand panel online using DesignStudio Software. On-demand panels are available for 1 (24 amplicons) to 500 (15,000 amplicons) genes and come in 24- and 96-reaction sizes. Libraries prepared with the AmpliSeq for Illumina Custom Panel are most compatible with the iSeq 100 System, MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Order online at www.illumina.com/products/by-type/sequencing-kits/library-prep-kits/ampliseq-on-demand-panel.html

TruSight One Sequencing Panel

The TruSight One Sequencing Panel provides comprehensive coverage of more than 4800 clinically relevant genes, allowing clinical research labs to analyze all of the genes on the panel or to focus on a specific subset. The panel focuses on the exonic regions harboring disease-causing variants. It was designed to cover the most commonly ordered molecular assays, enabling labs to perform multiple tests with one assay. Libraries prepared with the TruSight One Sequencing Panel are most compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of samples	Catalog no.
TruSight One Sequencing MiSeq Combo Kit[a]	9	FC-141-1006
TruSight One Sequencing Panel	36	FC-141-1007

TruSight One Expanded Sequencing Panel

The TruSight One Expanded Sequencing Panel was developed under the same guiding principles as the legacy panel with an extensive 16.5 Mb of genomic content. The design process for the TruSight One Expanded panel included additional optimization of the existing gene regions, which displayed lower performance, to provide improved coverage. In addition, ~1900 genes recently been associated with clinical phenotypes via reference databases were added to the panel. With the significant increase in gene content for the TruSight One Expanded Panel, it is recommended to be sequenced on the NextSeq 550 System and larger Illumina sequencing systems.

Ordering information

Product	No. of enrichments	Catalog no.
TruSight One Expanded Sequencing Panel	36	115075677

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TruSight Cardio Sequencing Panel

The TruSight Cardio Sequencing Panel targets genes linked to 17 inherited cardiac conditions (ICCs) impacted by genetic predisposition, such as cardiomyopathies, arrhythmias, and aortopathies. The panel includes custom oligos targeting identified regions of interest. Libraries prepared with the TruSight Cardio Sequencing Panel are compatible with the iSeq 100 System, MiniSeq System, MiSeq System, and NextSeq 550 System.

Ordering information

Product	No. of enrichments	No. of samples	Catalog no.
TruSight Cardio Sequencing Kit for MiSeq and MiSeqDx	1	12	FC-141-1010
TruSight Cardio Sequencing Kit for NextSeq and NextSeq Dx	4	48	FC-141-1011
TruSight Cardio Oligos[a]		48	15069654

a. Must be ordered with a TruSight Rapid Capture Kit

Library prep ordering information

TruSight Rapid Capture Kit

TruSight Rapid Capture Kits are all-in-one kits for library preparation and enrichment that allow researchers to prepare TruSight sequencing libraries quickly and easily. TruSight Rapid Capture leverages the speed of Nextera library prep technology. By introducing a unique multiplex pre-enrichment sample pooling, the need for mechanical DNA fragmentation has been eliminated. Use TruSight Rapid Capture with TruSight sequencing panels to maximize the productivity of your lab. Perform library prep and enrichment-based sequence capture for up to 96 samples in only 1.5 days. When sequencing large panels, such as TruSight One, on a MiSeq System, Illumina recommends choosing either a single index or lower number index kit configuration to achieve desired coverage.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight Rapid Capture Kit	1	8	FC-140-1101
TruSight Rapid Capture Kit	2	8	FC-140-1102
TruSight Rapid Capture Kit	4	16	FC-140-1103
TruSight Rapid Capture Kit	24	48	FC-140-1104
TruSight Rapid Capture Kit	24	96	FC-140-1105
TruSight Rapid Capture Kit	96	288	FC-140-1106
Training			
TruSight Rapid Capture Library Prep with MiSeq Training–Customer Site			TR-204-0016

Proof-of-Concept Service

The POC Service will run a subset of your samples with your desired system/application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs/goals, POC sequencing services of your samples at our Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
iSeq 100 POC Service	20023613
MiniSeq POC Service	20003924
MiSeq POC Service	SP-801-1002
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

See page 146 of the Services Section for Instrument Service Plans.

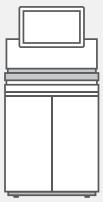
Select Illumina reagents and consumables are available with attributes that may benefit clinical research laboratories. Details on page 87.

Exome sequencing

Investigate the protein-coding regions of the genome with targeted sequencing to uncover genetic influences on disease and population health

Key strengths

- Flexible method for identifying variants across a wide range of applications, including population genetics, genetic disease, and cancer studies
- Comprehensive coverage of expertly selected exonic content
- Cost-effective alternative to WGS, as the human exome contains most known disease-causing variants
- Industry-leading content and increased solution options available due to the development of strategic partnerships

Prepare library	Sequence	Analyze and Report
		
TruSeq DNA Exome **Nextera DNA Exome** **AmpliSeq for Illumina Exome Panel** **TruSeq Library Prep with IDT xGen Exome Research Panel**	**NextSeq 550 System** **NovaSeq 6000 System**	**Available in BaseSpace Sequence Hub** **Isaac Enrichment** **BWA Enrichment**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Nextera DNA Exome	**Tiered service plans** NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples **Validation Services** IQ/OQ/PQ[a]

a. IQ = Installation Qualification, OQ = Operational Qualification, PQ = Performance Qualification

Visit www.illumina.com/exomeseq for more details.

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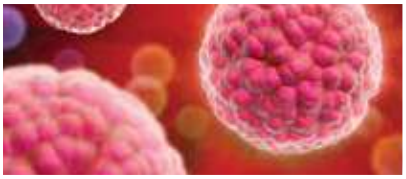
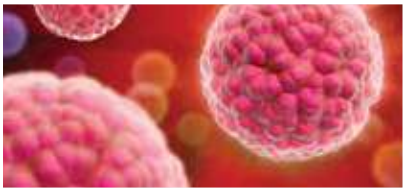


Cancer exome (cancer research)
Sequencing only 1% to 2% of the genome makes it possible to sequence to higher depth more economically, allowing researchers to focus on coding regions only. By performing tumor–normal exome sequencing, researchers can focus on the changes most likely to be tumorigenic.

Mendelian disorders (genetic conditions research)
Genetic conditions can be the result of rare variants that contribute to the inheritance of complex genetic diseases. Exome sequencing allows targeted exploration of human genome protein-coding regions to identify these rare variants that contribute to genetic diseases.

De novo mutation (complex disease)
Neurological and developmental disorders show the high impact of *de novo* mutations on disease risk. Family-based exome sequencing uncovers the mutational processes that occur from one generation to the next. Exome sequencing is a powerful technique to identify disease-causing variants in protein-coding regions offering increased throughput, high accuracy, and the simple workflow from sample generation to data analysis.



Cancer classification and progression (cancer research)
Cancers accumulate numerous genetic changes, but typically, only a few changes actually drive tumor progression. Monitoring cancer gene expression and transcriptome changes with RNA-Seq can help answer research questions on disease classification and progression.

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STEP 1
Choose a library prep method

Product	TruSeq RNA Exome	TruSeq DNA Exome[a]	Nextera DNA Exome	AmpliSeq for Illumina Exome Panel	IDT for Illumina Exome
Most important to me	Ability to assess RNA variants and expression	Highest[b] coverage of coding exons, FFPE samples	Highest[b] coverage of exons; fast assay time	Low input requirement, use with degraded samples, ease and speed of workflow	Industry-leading coverage and data quality
Least important to me	Total Assay time	Assay time	FFPE samples	DNA input amount	Ordering from a single vendor
Target region size	21,415 genes	~45 Mb	~45 Mb	~59 Mb	~51 Mb
Target region description	98.3% RefSeq	99.45% RefSeq 98.83% CCDS 99.68% ENSEMBL 99.68% GENCODE v19	99.45% RefSeq 98.83% CCDS 99.68% ENSEMBL 99.68% GENCODE v19	97% CCDS	Coding sequences of human genes in the RefSeq database
On-target percent	–	> 80%	Average of 75%	> 95%	> 95%
Recommended read length	2 × 75 bp	2 × 75 bp	2 × 75 bp	2 × 150 bp	2 × 75 bp
Kit configurations	48 samples each for Set A and B	24, 48, 72, or 96 samples	8, 24, 48, 72, or 96 samples	12 pools for 8 samples	192 or 1152 samples

a. See page 43 for a list of modular components for TruSeq DNA Exome
b. Comparisons are within the Illumina targeted sequencing portfolio

STEP 2
Choose a sequencer

Product	NextSeq 550 System	NovaSeq 6000 System		
Most important to me	Convenience, rapid turn around time, and instrument affordability for exome sequencing applications	Scalable throughput and flexibility for virtually any exome, and scale of project at lowest[a] price point		
Run mode/kit type	Mid-Output/High-Output	S1	S2	S4
Exomes processed/flow cell	Up to 12	40[b]	80[b]	240[b]

a. Comparisons among Illumina portfolio
b. Based on 8 Gb/sample (~100× coverage of 45 Mb of genomic content)

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STEP 3
Obtain results

Targeted resequencing ▼

1

Align/Call variants

Isaac Enrichment B
- Enables rapid analysis of panels using capture-based methodology, such as Nextera DNA Exome, the TruSight One Panel, and Illumina custom capture panels built in DesignStudio Software
- Performs alignment and small- and large-variant calling using algorithms developed by Illumina
- Offers 4× faster alignment speed with the same accuracy as the BWA Enrichment App
- Provides multilaunch for simultaneous analysis of 96 samples

BWA Enrichment B
- Enables rapid analysis of panels using capture-based methodology, such as Nextera DNA Exome, the TruSight One Panel, and Illumina custom capture panels built in DesignStudio Software
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm
- Provides multilaunch for simultaneous analysis of 96 samples

2

Annotate/Filter

VariantStudio Software B
- Enables extraction of biological knowledge from variant data by providing a rich annotation database, flexible filtering, and a streamlined variant classification and reporting tool
- Leverages leading annotation databases and a powerful filtering interface to rapidly identify disease-associated variants in data sets

3

Interpret/Report

BaseSpace Variant Interpreter
- Enables rapid, rich annotation, filtering, and interpretation of genomic data. Customize workflows and summarize findings into structured reports. Determine biological significance of genomic variants within a software framework focused on data security, compliance, and operational efficiency
- Leverages BaseSpace Knowledge Network containing genomic interpretation content and leading annotation databases to confidently expedite interpretation of variants into meaningful data

4

Add biological context

BaseSpace Cohort Analyzer
- Analyze complex human subject data for translational research applications on a web-based platform
- Provides access to the largest repository of curated public data sets in the world and powerful tools for cohort analysis and group comparisons of both public and proprietary data

B BaseSpace Sequence Hub
Visit www.illumina.com for more details.

Library prep ordering information

TruSeq DNA Exome

TruSeq DNA Exome provides all-in-one library prep and exome enrichment. This kit is optimized for use with both DNA and FFPE samples, delivering uniform and specific coverage of 45 Mb of exonic content with as little as 4 Gb of sequencing. Libraries generated with TruSeq DNA Exome are compatible with the NextSeq 550 System and NovaSeq 6000 System.

Ordering information

Product	Pooling plexity	No. of enrichment reactions	Total no. of samples	Catalog no.
TruSeq DNA Exome	3	8	24	20020614
TruSeq DNA Exome	12	8	96	20020615

Nextera DNA Exome

Nextera DNA Exome provides all-in-one library prep and exome enrichment. This kit is optimized for use with DNA samples, delivering uniform and specific coverage of 45 Mb of exonic content with as little as 4 Gb of sequencing in a rapid single-day. Libraries generated with Nextera DNA Exome are compatible with the NextSeq 550 System and NovaSeq 6000 System.

Ordering information

Product	Pooling plexity	No. of enrichment reactions	Total no. of samples	Catalog no.
Nextera DNA Exome	3	8	24	20020616
Nextera DNA Exome	12	8	96	20020617

TruSeq RNA Exome Library Prep Kits

TruSeq RNA Exome Library Prep Kits use proven TruSeq Stranded RNA Library Prep Kit chemistry combined with efficient sequence-specific captures to generate RNA-Seq libraries that focus on the RNA coding regions from degraded samples. TruSeq RNA Exome kits require as little as 10 ng of input from intact RNA samples or 20 ng of input RNA from degraded samples. Libraries prepared with the TruSeq RNA Exome kit are compatible with the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product[a]	No. of indexes	No. of samples	Catalog no.
TruSeq RNA Library Prep for Enrichment		48	20020189
TruSeq RNA Enrichment (12 enrichments)		48	20020490
Exome Panel			20020183
TruSeq RNA Single Indexes Set A	12	48	20020492
TruSeq RNA Single Indexes Set B	12	48	20020493

a. Use of the TruSeq RNA Exome solution requires a library prep kit, an enrichment kit, a panel, and an index kit

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TruSeq Exome Library Prep Modular Components

Components within the Illumina library preparation kits are available as single components, allowing customers to take advantage of proven Illumina chemistry and use indexes from other sources, such as Integrated DNA Technologies (IDT). This also offers users the opportunity to increase the plexity of their assays and enables accurate assignment of reads and more efficient use of flow cells. The library preparation protocol requires one kit from each component, depending on sample requirements. This assay is most compatible with the NextSeq 550 System and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Components			
TruSeq DNA Library Prep for Enrichment		24	20020181
TruSeq DNA Library Prep for Enrichment		96	20020182
Enrichment Components (order directly from Integrated Digital Technologies)			
IDT - Blocking Oligos		192	1075474 (IDT catalog no.)
IDT - xGen Lock Down Hyb and Wash		192	1072280 (IDT catalog no.)
IDT - Blocking Oligos		1152	1075475 (IDT catalog no.)
DT - xGen Lock Down Hyb and Wash		1152	1072281 (IDT catalog no.)
Probe Panel Component (order directly from Integrated Digital Technologies)			
IDT xGen Exome Research Panel		192	1056114 (IDT catalog no.)
IDT xGen Exome Research Panel		1152	1056115 (IDT catalog no.)
Index Component			
TruSeq DNA UD Indexes	24	96	20020590
TruSeq DNA Single Indexes Set A	12	24	20015960
TruSeq DNA Single Indexes Set B	12	24	20015961

AmpliSeq for Illumina Exome Panel

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. The exome panel is part of an integrated workflow that includes AmpliSeq for Illumina PCR-based library preparation, Illumina sequencing by synthesis (SBS) next-generation sequencing (NGS) technology, and automated analysis. With 59 Mb of genomic content, this assay is most compatible with the NextSeq 550 System and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Exome Panel		8	20019166
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Large Volume	96	96	20019108

Proof-of-Concept Service

The Proof-of-Concept (POC) Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at the Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

Concierge service for your custom panel

A dedicated expert from design to delivery

Custom library panel prep projects might require special attention. Illumina Concierge supports all of your needs, from target region design to functional performance evaluation and optimization to final shipment.

Illumina Concierge

Product content service

- Two levels of Concierge Service: design assistance and product optimization
- Thorough consultation of custom design needs from Illumina experts
- Optimized *in silico* target coverage and minimized gaps
- Functional product performance improvements
- Coordination of ordering and shipping

	Design assistance	Product optimization
Dedicated design expert	•	•
Extended capabilities	•	•
Project management	–	•
Functional testing	–	•
Iterative product enhancements	–	•

Contact your local representative to learn more about Illumina products and services available in your region.
800.809.4566 (North America) • 01799 534332 toll-free (Europe, Middle East, Africa) • +61.3.9212.9900 (Australia)
+65.6773.0188 (Singapore) • +81.3.4578.2800 (Japan) • +86.21.6032.1066 (China)

Metagenomics

Sequence complex microbial samples to identify emerging diseases, analyze microbial communities, and classify microbial species

Key strengths

- Enables bacteria identification and comparison within a mixed population sample
- Accelerates discovery of new SNP markers that translate to virulence or antibiotic resistance
- Promotes accurate evaluation of microbial biodiversity within a given sample
- Contributes to microbial phylogeny and taxonomy

Prepare library	Sequence	Analyze and Report
		
16S Metagenomic Sequencing Protocol **TruSeq DNA PCR-Free Library Prep Kit** **TruSeq Nano DNA Library Prep Kit** **Nextera DNA Flex Library Prep Kit** **Nextera XT DNA Library Prep Kit**	**iSeq 100 System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software, BaseSpace Sequence Hub** Metagenomics **Available on BaseSpace Sequence Hub**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training TruSeq DNA PCR-Free Library Prep Kit Nextera DNA Library Prep Kit Nextera XT Library Prep Kit	**Tiered service Plans** iSeq 100 System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof of Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/metagenomics for more details.

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Bacterial outbreak surveillance (public health and epidemiology)
Outbreak identification and surveillance are critical to understanding virus transmission and variation. Shotgun metagenomics provides additional information about coinfections and novel organisms and supports pathogen–host interaction studies.

Microbiome (agrigenomics)
Bacteria, viruses, and fungi all affect agriculturally important plant and animal species. Metagenomics allows researchers and farmers to manage the health of their herds and plant crops to maximize food production and quality.

Microbiome (environmental studies)
Complex environmental samples often include organisms that cannot be cultured in the laboratory. With shotgun metagenomics sequencing, researchers evaluate bacterial diversity and detect the abundance of microbes in various environments, including unculturable microorganisms.

Microbial identification
Microbial samples are often complex, requiring researchers to identify and compare the populations and activities of the microbes present within a single sample. High-speed, multiplex 16S rRNA sequencing improves the sensitivity and specificity of taxonomic assignments down to the genus level, and sometimes even to the species level.

Human microbiome research
The human gut microbiome varies widely among individuals and populations, and can affect health status. Leveraging shotgun metagenomics, scientists identify the impact of human genetics on variation of the gut microbiome.

STEP 1
Choose a library prep method




Product	TruSeq DNA PCR-Free Library Prep Kit	TruSeq DNA Nano Kit	Nextera DNA Flex Library Prep Kit	Nextera XT DNA Library Prep Kit
Most important to me	Lowest[a] bias to achieve highest[a] coverage	High coverage of complex genomes with low input requirements	Simple and versatile sample-to-data workflow	Fast workflow with low input requirements for small genomes
Least important to me	Low DNA input quantity	Assay time	PCR-free workflow	Amplicons < 300 bp in length
Input quantity	1 µg DNA	100 ng DNA	1–500 ng DNA	1 ng DNA
Assay time	5 hrs	6 hrs	3–4 hrs	90 min
Diversity	≥ 1B unique fragments	≥ 1B unique fragments	≥ 1B unique fragments	< 1B unique fragments
Genome coverage	Complete	Complete	Complete	Slight bias in coverage
Fragmentation process	Mechanical	Mechanical	Enzymatic	Enzymatic

a. Comparisons are within the Illumina targeted sequencing portfolio

STEP 2
Choose a sequencer




Product	iSeq 100 System	MiSeq Series	NextSeq 550 System	NovaSeq 6000 System
Most important to me	Affordability	Long read lengths (2 × 300 bp) 16s metagenomics	Whole-genome metagenomics with convenient, rapid turnaround time and instrument affordability	Production power for whole genome metagenomics with lowest cost per sample[a]
Onboard informatics	●	●	●	–
16s metagenomic samples per flow cell at 20K clusters per sample	180	384 samples v3 600 cycle kit	384	1536
Run mode/kit type	Standard	Mid-output/High-output	Mid-output/High-output	S4 Xp
Flow cells processed/run	1	1	1	1 or 2

a. Comparison among Illumina sequencing portfolio

Visit www.illumina.com/metagenomics for more details.

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STEP 3
Obtain results

Metagenomics ▼

Add biological context

Metagenomics workflow L and 16S Metagenomics B
- Performs taxonomic classification of 16S rRNA-targeted amplicon reads
- Provides interactive visualizations and raw classification output for presample and aggregate analyses

Kraken Metagenomics B
- Assigns taxonomic labels to short DNA sequences with high sensitivity and speed using exact k-mer alignments and a novel classification algorithm
- Optionally performs host read filtering using the Scalable Nucleotide Alignment Program (SNAP) aligner to removed reads that align to the reference genome of the host organism before read classification with the app

MetaPhlAn B
- Profiles the composition of microbial communities from metagenomic shotgun sequencing data
- Relies on unique clade-specific marker genes identified from reference genomes, allowing orders of magnitude speedups and unambiguous taxonomic assignments

B BaseSpace Sequence Hub L Local Run Manager Software

Library prep ordering information

Nextera DNA Flex Library Prep Kits

Save time and reduce hands-on touch points with On-Bead Tagmentation, producing sequencing-ready libraries in less than three hours. Simplify daily operations with a kit that supports a broad DNA input range (1–500 ng), multiple DNA input types, and flexible for use with small (bacteria, archaea, viruses, and plasmids) to large genomes (human, plant, mouse). Libraries prepared with Nextera DNA Flex Library Prep Kits are compatible with all Illumina sequencing systems.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Flex Lysis Reagent Kit			20018706
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

Nextera XT DNA Library Prep Kits

Generate sequencing-ready libraries from small genomes (bacteria, archaea, viruses), amplicons, and plasmids in less than 90 minutes with only 15 minutes of hands-on time and as little as 1 ng of input. Nextera XT DNA Library Prep Kits simultaneously fragment input DNA and tag the fragments with sequencing adapters in a single-tube enzymatic reaction. Bead-based normalization eliminates the need for library quantification before pooling and sequencing. Libraries prepared with Nextera XT DNA Library Prep Kits are most compatible with the MiSeq System and NextSeq 550 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera XT DNA Library Prep Kit		24	FC-131-1024
Nextera XT DNA Library Prep Kit		96	FC-131-1096
TruSeq Dual Index Sequencing Primer Kit, single read	Single-use kit[a]		FC-121-1003
TruSeq Dual Index Sequencing Primer Kit, paired-end read	Single-use kit[a]		PE-121-1003
Nextera XT Index Kit v2, set A	96	384	FC-131-2001
Nextera XT Index Kit v2, set B	96	384	FC-131-2002
Nextera XT Index Kit v2, set C	96	384	FC-131-2003
Nextera XT Index Kit v2, set D	96	384	FC-131-2004
Training			
Nextera XT Library Prep Kit training at customer site			TR-204-0009

a. Sequencing primer kits are only required for use with TruSeq and HiSeq SBS Kits v3

TruSeq DNA PCR-Free Library Prep Modular Kits

TruSeq DNA PCR-Free Library Prep Kits provide uniform coverage for whole-genome library prep for organisms ranging from bacteria to human. The kits offer shortened gel-free workflows, the ability to sequence the most challenging regions, and the power to identify a large number of variants. Libraries prepared with TruSeq DNA PCR-Free Library Prep Kits are compatible with all Illumina sequencing systems.

TruSeq DNA PCR-Free Library Prep Kits are available as single components, allowing users to take advantage of proven Illumina chemistry and use indexes from other sources, such as Integrated DNA Technologies (IDT). This also offers users the opportunity to increase assay plexity and enables accurate assignment of reads and more efficient use of flow cells. The library preparation protocol requires one kit from each component, depending on sample requirements.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
TruSeq DNA PCR-Free Low-Throughput Library Prep Kit		24	20015962
TruSeq DNA PCR-Free High-Throughput Library Prep Kit		96	20015963
Index Adapter Component			
TruSeq DNA Single Indexes Set A	12	24	20015960
TruSeq DNA Single Indexes Set B	12	24	20015961
TruSeq DNA CD Indexes	96	96	20015949
IDT for Illumina - TruSeq DNA UD Indexes	24	96	20020590
IDT for Illumina - TruSeq DNA UD Indexes	96	96	20022370
Training			
TruSeq DNA PCR-Free Library Prep Kit training at customer site			TR-204-0011

TruSeq DNA Nano Library Prep Modular Kit

Preserve precious samples with the TruSeq DNA Nano Library Prep Kit. Prepare sequencing libraries for low- or high-throughput studies from as little as 100 ng of input DNA in less than a day. Bead-based selection reduces the sample loss associated with gel-based selection. This kit is designed to match the ever-increasing read lengths of Illumina sequencing instruments and is compatible with the MiSeq System, NextSeq 550 System, and HiSeq X Series.

TruSeq DNA Nano Library Prep Kits are available as single components, allowing users to take advantage of proven Illumina chemistry and use indexes from other sources, such as Integrated DNA Technologies (IDT). This also offers users the opportunity to increase assay plexity and enables accurate assignment of reads and more efficient use of flow cells. The library preparation protocol requires one kit from each component, depending on sample requirements.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
TruSeq DNA Nano Library Prep Kit		24	20015964
TruSeq DNA Nano Library Prep Kit		96	20015965
Index Adapter Component			
TruSeq DNA Single Indexes Set A	12	24	20015960
TruSeq DNA Single Indexes Set B	12	24	20015961
TruSeq DNA CD Indexes	96	96	20015949
IDT for Illumina - TruSeq DNA UD Indexes	24	96	20020590
IDT for Illumina - TruSeq DNA UD Indexes	96	96	20022370

16S Metagenomic Sequencing Protocol

The 16S Metagenomic Sequencing Protocol describes a method for preparing libraries that allows researchers to sequence the variable V3 and V4 regions of the 16S rRNA gene. This protocol can also be used for sequencing other regions with different region-specific primers. Sequencing libraries prepared with the 16S Metagenomic Sequencing Protocol are compatible with the MiSeq System. The 16S Metagenomic Sequencing Protocol is available at www.illumina.com/metagenomics.

Ordering information

Product	Catalog no.
Nextera XT Index Kit	FC-131-1001
	FC-131-1002
PhiX Control Kit v3	FC-110-3001

Custom sequencing

Capture your genes of interest

Key strengths

- Multiple library preparation options meet a wide variety of needs
  - Choose amplicon sequencing for analyzing degraded samples, genes, and small genomic regions (15–400 kb)
  - Choose enrichment sequencing for fusion discovery and analyzing large genomic regions (400 kb–15 Mb)
- Fully customizable targeted sequencing products are available to meet specific research needs
- Flexible technologies are applicable in a wide range research areas, including cancer studies, population genetics, and complex disease

Prepare library	Sequence	Analyze and Report
		
AmpliSeq for Illumina **Nextera XT DNA Library Prep Kit** **Nextera DNA Flex Library Prep Kit** **Nextera Rapid Capture Custom Enrichment Kit**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software, BaseSpace Sequence Hub** Targeted resequencing
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training AmpliSeq for Illumina Nextera XT Library Prep Kit Nextera DNA Flex Library Prep Kit Nextera Rapid Capture Custom Enrichment Kit	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept service** Standard application functional testing with your samples

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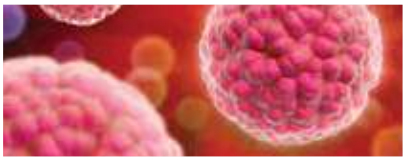
Antibacterial drug resistance (public health and epidemiology)
Drug resistance develops during treatment for certain viral infections as the proportion of naturally occurring drug-resistant variants in the body increases. Public health experts track the development of bacterial drug resistance with targeted sequencing.



Gene targeting (cancer research)
Custom amplicon sequencing is useful for targeting genes known to be involved in diseases, such as cancer. Custom sequencing allows users to select specific targets and achieve research results quickly and efficiently.

Selective breeding (agrigenomics)
Plant and animal breeders strive to emphasize the desirable traits in offspring and make their choices based on physical traits. Custom amplicon sequencing enables breeding decisions to be made based on genetic information.

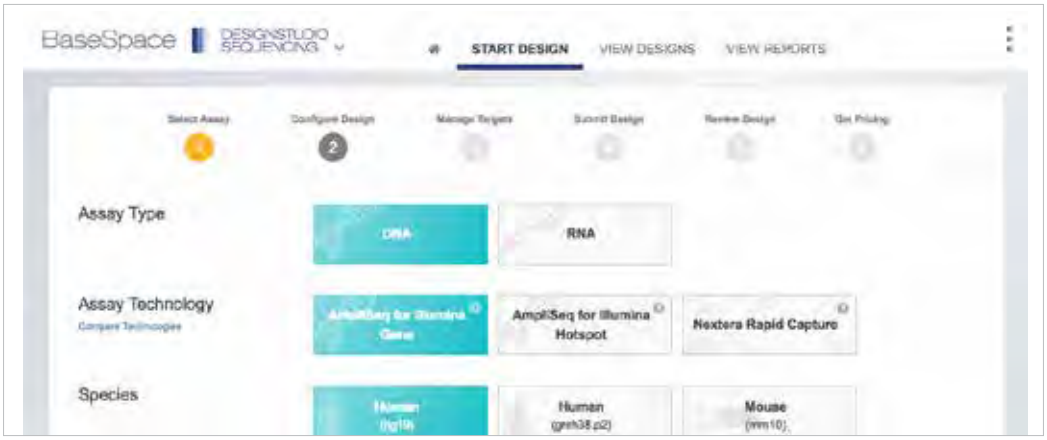
Variant identification (complex disease)
Rare genetic variants sometimes contribute to complex diseases, such as neuropsychiatric disorders. Targeted sequencing enables focused assessment of candidate pathogenic genes and the identification of rare genetic variants that contribute to disease pathogenesis.

Trusted solutions for targeted NGS panel design

DesignStudio Software provides dynamic feedback to help users optimize target region sequencing coverage, reducing the time required to design custom projects. Users can personalize and optimize content to fit the needs of their study.

This web-based software tool is available for the following targeted sequencing products:

- Nextera Rapid Capture Custom Enrichment Kit
- AmpliSeq for Illumina
- TruSeq Targeted RNA Expression Kits



Visit www.illumina.com/customlibraryprep for more details.

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

STEP 1
Choose a library prep method




Product	AmpliSeq for Illumina Custom Panel	Nextera XT DNA Library Prep Kit	Nextera DNA Flex Library Prep Kit	Nextera Rapid Capture Custom Enrichment Kit
Most important to me	FFPE, low input, easy to use	Fastest[a] assay time; lowest[a] DNA input quantity	Simple DNA-to-data workflow	User customizes region of interest
Least important to me	Large genomic regions	Amplicons < 300 bp in length	PCR-free workflow	Preexisting probes
Input quantity	10 ng/pool	1 ng DNA	1–500 ng DNA	50 ng DNA
Assay time	5–7 hrs	90 min	3–4 hrs	1.5 days
Hands-on time	1.5 hrs	15 min	1.5 hrs[b]	5 hrs
FFPE compatibility	Yes	No	No	No
Design	Customers use DesignStudio Software to design probes targeting their regions of interest	Independently designed customer probes target amplicons at least 300 bp in length	Independently designed customer probes target amplicons at least 300 bp in length	Customers use DesignStudio Software to design probes targeting their regions of interest
Fragmentation method	None	Enzymatic	Enzymatic	Enzymatic

a. Comparisons are within the Illumina targeted sequencing portfolio

b. Denotes total workflow time, including DNA extraction, library preparation, and library normalization/pooling steps

STEP 2
Choose a sequencer





Product	iSeq 100 System	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System
Most important to me	Affordability	Onboard informatics, CapEx affordability, speed	Onboard informatics, read-length flexibility, simplicity	Assay flexibility, output scalability, simplicity	Lowest[a] price per sample
Onboard informatics	●	●	●	–	–
Run mode/kit type	Standard	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	S1, S2, S4
Flow cells processed/ run	1	1	1	1	1 or 2

a. Comparisons among Illumina sequencing instrument portfolio based on performance of the S4 flow cell

STEP 3
Obtain results

Targeted resequencing ▼

1

Align/Call variants

Amplicon workflow L and DNA Amplicon B
- Enables rapid analysis of panels using amplicon-based methods, such as the TruSight Cancer Panel, TruSight Myeloid Panel, and AmpliSeq for Illumina Custom Panel built in DesignStudio Software
- Performs alignment using the banded Smith-Waterman algorithm in the targeted regions
- Performs variant calling with the Broad's GATK small-variant calling algorithm, the small-variant caller developed by Illumina, or the somatic variant caller developed by Illumina

PCR amplicon workflow L
- Enables analysis of PCR amplicons prepared with Nextera XT Library Prep Kits
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm

Enrichment workflow L
- Enables analysis of panels using capture-based methods, such as the Illumina custom capture panels built in DesignStudio Software
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm

Isaac Enrichment B
- Enables analysis of panels using capture-based methods, such as the Illumina custom capture panels built in DesignStudio Software
- Performs alignment, and small- and large-variant calling using algorithms developed by Illumina
- Offers 4× faster alignment speed with the same accuracy as the BWA Enrichment App
- Provides multilaunch that allows simultaneous analysis of 96 samples

BWA Enrichment B
- Enables analysis of panels using capture-based methods, such as the Illumina custom capture panels built in DesignStudio Software
- Uses the Sanger BWA alignment algorithm combined with the Broad's GATK small-variant calling algorithm
- Provides multilaunch that allows simultaneous analysis of 96 samples

2

Annotate/Filter

VariantStudio B
- Enables extraction of biological knowledge from variant data by providing a rich annotation database, flexible filtering, and a streamlined variant classification and reporting tool
- Leverages leading annotation databases and a powerful filtering interface for rapid identification of disease-associated variants in data sets

3

Interpret/Report

BaseSpace Variant Interpreter B
- Enables rapid, rich annotation, filtering, and interpretation of genomic data. Customizes workflows and summarizes findings into structured reports. Determines biological significance of genomic variants within a software framework focused on data security, compliance, and operational efficiency
- Leverages BaseSpace Knowledge Network containing genomic interpretation content and leading annotation databases to confidently expedite interpretation of variants into meaningful data

4

Add biological context

Correlation Engine
- Offers a web-based platform for analyzing complex human subject data for translational research applications
- Provides access to the largest repository of curated public data sets in the world and powerful tools for large cohort analysis and group comparisons of both public and proprietary data

B BaseSpace Sequence Hub L Local Run Manager Software

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

Library prep ordering information

AmpliSeq for Illumina Custom Panels

AmpliSeq for Illumina is a streamlined, scalable, amplicon sequencing solution that delivers high-confidence data from low-input DNA and RNA samples. The solution leverages high-performance AmpliSeq chemistry and the Illumina DesignStudio Software to enable researchers to focus their studies on specific genes, regions, or variants of interest with high accuracy. Targets for custom panels can be selected based on several preloaded reference genomes or cultivated from an on-demand gene list. Custom panels can include from dozens to several hundred genes in any species (DNA panel only) to meet a variety of needs.

The AmpliSeq for Illumina workflow requires an AmpliSeq for Illumina panel, AmpliSeq for Illumina Library PLUS Kit, AmpliSeq for Illumina CD Indexes, and the AmpliSeq for Illumina DNA Library Prep Kit (optional). Libraries prepared with the AmpliSeq for Illumina solution are compatible with all Illumina sequencing systems, depending on expected throughput.

Ordering information

Product	No. of samples	Catalog no.
AmpliSeq for Illumina Custom DNA Panel[a]	750	20020495
AmpliSeq for Illumina Custom DNA Panel[a]	3000	20020497
AmpliSeq for Illumina Library PLUS	24	20019101
AmpliSeq for Illumina Library PLUS	96	20019102
AmpliSeq for Illumina Library PLUS	384	20019103
AmpliSeq for Illumina CD Indexes Set A	96 indexes, 96 samples	20019105
AmpliSeq for Illumina DNA Library Preparation Kit		20023392

a. A number of ready-to-use AmpliSeq for Illumina panels are available. Visit www.illumina.com/products/by-brand/ampliseq.com to learn more

Nextera XT DNA Library Prep Kits

Generate sequencing-ready libraries from small genomes (bacteria, archaea, viruses), amplicons, and plasmids in less than 90 minutes with only 15 minutes of hands-on time. Nextera XT DNA Library Prep Kits simultaneously fragments input DNA and tags the fragments with sequencing adapters in a single-tube enzymatic reaction with as little as 1 ng of input. Bead-based normalization eliminates the need for library quantification before pooling and sequencing. Libraries prepared with Nextera XT DNA Library Prep Kits are compatible with the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera XT DNA Library Prep Kit		24	FC-131-1024
Nextera XT DNA Library Prep Kit		96	FC-131-1096
Nextera XT Index Kit	24	96	FC-131-1001
TruSeq Dual Index Sequencing Primer Kit, single read	Single-use kit[a]		FC-121-1003
TruSeq Dual Index Sequencing Primer Kit, paired-end read	Single-use kit[a]		PE-121-1003
Nextera XT Index Kit v2, set A	96	384	FC-131-2001
Nextera XT Index Kit v2, set B	96	384	FC-131-2002
Nextera XT Index Kit v2, set C	96	384	FC-131-2003
Nextera XT Index Kit v2, set D	96	384	FC-131-2004
Training			
Nextera XT Library Prep Kit training at customer site			TR-204-0009

a. Sequencing primer kits are only required for use with TruSeq and HiSeq SBS Kits v3

Nextera DNA Flex Library Prep Kits

Save time and reduce hands-on touch points with On-Bead Tagmentation, producing sequencing-ready libraries in less than three hours. Simplify daily operations with a kit that supports a broad DNA input range (1–500 ng), multiple DNA input types, and flexible for use with small (bacteria, archaea, viruses, and plasmids) to large genomes (human, plant, mouse). Libraries prepared with Nextera DNA Flex Library Prep Kits are compatible with all Illumina sequencing systems.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Flex Lysis Reagent Kit			20018706
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

Nextera Rapid Capture Custom Enrichment Kit

The Nextera Rapid Capture Custom Enrichment Kit provides unparalleled access to genomic regions of interest. This custom assay allows researchers to sequence precious samples faster and more efficiently than ever before, using as little as 50 ng of input DNA. The kit offers add-on functionality to refine content over time, or add regions of unique interest to established panels like Nextera Rapid Capture Exome Kit or TruSight Panel content sets. Libraries prepared with the Nextera Rapid Capture Custom Enrichment Kit are compatible with the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	Pooling plexity	No. of enrichment reactions	Total no. of samples	Catalog no.
Nextera Rapid Capture Custom Enrichment Kit	12	4	48	FC-140-1007
Nextera Rapid Capture Custom Enrichment Kit	12	8	96	FC-140-1008
Nextera Rapid Capture Custom Enrichment Kit	12	8	288	FC-140-1009
Training				
Nextera Rapid Capture Enrichment Kit training at customer site				TR-204-0014

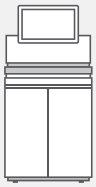
See page 149 for Proof-of-Concept Service information.

Whole-transcriptome and stranded sequencing

Analyze coding and multiple forms of noncoding RNA for a more complete picture of the transcriptome with strand information

Key strengths

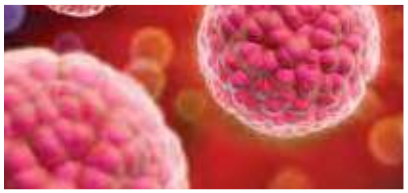
- Captures both known and novel features
- Offers ribosomal reduction options for use with degraded samples
- Enables a more comprehensive understanding of phenotypes of interest

Prepare library	Sequence	Analyze and Report
		
TruSeq Stranded Total RNA with Ribo-Zero Kit Human/Mouse/Rat Gold Plant Globin **TruSeq Stranded mRNA Library Prep paired with Ribo-Zero Kit** Epidemiology Bacteria Yeast	**NextSeq 550 System** **NovaSeq 6000 System**	**Available on BaseSpace Sequence Hub and RNA-Seq** **Gene expression profiling**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training TruSeq Stranded Total RNA Library Prep Kit TruSeq Stranded RNA Library Prep Kit	**Tiered service plans** NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/wholetranscriptome for more details.

Innovation. Discovery. Application.



Cancer gene expression (cancer research)
Cancers accumulate numerous genetic changes, but, typically, only a few changes actually drive tumor progression. Monitoring changes in expression of both coding and noncoding RNA can inform research questions about cancer classification and progression.

Deep transcriptome sequencing (agrigenomics)
Plants have large, repetitive genomes that can make sequencing low-expression genes difficult. Capturing both coding and noncoding RNA provides a more complete view of often complex plant transcriptomes, benefiting a range of applications.

Biomarker identification (complex disease)
Various forms of noncoding RNA have been implicated in a rapidly increasing range of complex diseases. Whole-transcriptome RNA-Seq allows researchers to study coding and multiple forms of noncoding RNA in a single analysis, providing a more complete picture of the biology of complex disease and visibility to a broader range of potential biomarkers.

> Having deep sequencing coverage with RNA-Seq, we've discovered—and have been validating—tens of thousands of novel genes.
>
> **Christopher E. Mason, PhD**, Assistant Professor, Department of Physiology and Biophysics at the Institute for Computational Biomedicine, Weill Cornell Medical College

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STEP 1
Choose a library prep method

Product	TruSeq Stranded Total RNA with Ribo-Zero Kit	TruSeq Stranded mRNA Library Prep paired with Ribo-Zero Kit[a]
Most important to me	Capture coding and noncoding transcriptome	Focused capture of coding transcriptome
FFPE compatibility	Yes	Yes
RNA input	0.1–1 µg	0.1–1 µg
Assay time	1.5 days	2 days
Multiplexing	12–96 samples	12–96 samples
Available organisms	Human, mouse, rat, blood, plant	Disease samples, bacteria, yeast

a. For bacteria, yeast, or disease transcriptome samples, Illumina recommends using the Ribo-Zero Kit designed for your sample type with TruSeq Stranded mRNA Kits

STEP 2
Choose a sequencer

Product	NextSeq 550 System		NovaSeq 6000 System
Most important to me	Flexibility and instrument affordability for gene expression profiling		Lowest[a] cost per sample for production scale RNA-Sequencing
Production scale for human and nonhuman samples	–		●
Run mode/kit type	Mid-output	High-output	S1, S2, S4
Flow cells processed/run	1	1	1 or 2

a. Comparisons among Illumina sequencing portfolio

Visit www.illumina.com/wholetranscriptome for more details.

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STEP 3
Obtain results

RNA-Seq ▼

1 Align/Call variants

TopHat Alignment B
- Maps reads and calls small variants (SNVs and small indels)
- Performs abundance estimations of reference genes and transcripts
- Offers optional fusion calling

2 Assemble/Count

Cufflinks Assembly & DE B
- Assembles novel transcripts
- Performs differential expression (DE) of novel and reference transcripts

Gene expression profiling ▼

Align/Count

RNA Express B
- Aligns RNA-Seq reads with the STAR aligner and assigns aligned reads to genes
- Performs differential gene expression with DESeq2

3 Interpret/Report

BaseSpace Cohort Analyzer
- Provides a web-based platform for analyzing complex human subject data for translational research applications
- Provides access to a comprehensive repository of curated public data sets in the world and powerful tools for large cohort analysis and group comparisons of both public and proprietary data

BaseSpace Correlation Engine
- Provides a web-based platform with an interactive data analysis environment that integrates your data with vast amounts of research data
- Compares Omics molecular profiles from your own experiments with results from a large curated repository of public data assets

B BaseSpace Sequence Hub

Library prep ordering information

TruSeq Stranded Total RNA with Ribo-Zero™ Modular Kits

TruSeq Stranded Total RNA with Ribo-Zero Kits provide a clear and complete view of the transcriptome with a streamlined, cost-efficient, and scalable solution for total RNA analysis. Compatible with a wide range of samples, including low quality and FFPE, these kits combine the benefits of TruSeq RNA Library Prep Kits with ribosomal RNA (rRNA) reduction chemistry. This combination allows analysis of coding and multiple forms of noncoding RNA with precise measurement of strand orientation, uniform coverage, and high-confidence discovery of features such as alternative transcripts, gene fusions, and allele-specific expression. Libraries prepared with TruSeq Stranded Total RNA with Ribo-Zero Kits are most compatible with the NextSeq 550 System and NovaSeq 6000 System. Library preparation requires one library preparation component and one index component.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Preparation Component			
TruSeq Stranded Total RNA Library Prep Kit—Human/Mouse/Rat		48	20020596
TruSeq Stranded Total RNA Library Prep Kit—Human/Mouse/Rat		96	20020597
TruSeq Stranded Total RNA Library Prep Kit Gold		48	20020598
TruSeq Stranded Total RNA Library Prep Kit Gold		96	20020599
TruSeq Stranded Total RNA Library Prep Kit Plant		48	20020610
TruSeq Stranded Total RNA Library Prep Kit Plant		96	20020611
TruSeq Stranded Total RNA Library Prep Kit Globin		48	20020612
TruSeq Stranded Total RNA Library Prep Kit Globin		96	20020613
Index Component			
TruSeq RNA Single Indexes Set A	12		20020492
TruSeq RNA Single Indexes Set B	12		20020493
TruSeq RNA CD Indexes	96		20019792
IDT for Illumina – TruSeq RNA UD Indexes	24	96	20020591
IDT for Illumina – TruSeq RNA UD Indexes	96	96	20022371
Training			
TruSeq Stranded Total RNA Library Prep Kit training at customer site			TR-204-0012

TruSeq Stranded mRNA Library Prep with Ribo-Zero Modular Kit

The TruSeq Stranded mRNA Library Prep paired with Ribo-Zero Kit provides a clear and complete view of the transcriptome with a streamlined, cost-efficient, and scalable solution for mRNA analysis. The kit supports precise measurement of messenger RNA (mRNA) strand orientation for detection of antisense transcription, enhanced transcript annotation, and increased alignment efficiency. High coverage uniformity enhances the discovery of features such as alternative transcripts, gene fusions, and allele-specific expression. Libraries prepared with the TruSeq Stranded mRNA Library Prep paired with Ribo-Zero Kit are most compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System. Library preparation requires one library preparation component and one index component.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Preparation Component			
TruSeq Stranded mRNA Library Prep Kit		48	20020594
TruSeq Stranded mRNA Library Prep Kit		96	20020595
Index Component			
TruSeq RNA Single Indexes Set A	12		20020492
TruSeq RNA Single Indexes Set B	12		20020493
TruSeq RNA CD Indexes	96		20019792
IDT for Illumina – TruSeq RNA UD Indexes	24	96	20020591
IDT for Illumina – TruSeq RNA UD Indexes	96	96	20022371

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Ribo-Zero rRNA Removal Kits

Sequencing is an important tool of discovery for research into human health, disease, development, and more. Add Ribo-Zero Kits to the TruSeq Stranded mRNA Kit library prep workflow to remove unwanted rRNA from the organisms of interest before sequencing to get informative results.

Ribo-Zero Gold rRNA Removal Kit (Epidemiology)

The Ribo-Zero Gold rRNA Removal Kit (Epidemiology) effectively removes cytoplasmic and mitochondrial rRNA from complex samples composed of human, mouse, rat, and a broad spectrum of Gram-positive and Gram-negative bacterial RNA.

Ordering information

Product	Ribosomal removal	No. of samples	Catalog no.
Ribo-Zero Gold rRNA Removal Kit (Epidemiology)	Cytoplasmic and mitochondrial	6	MRZE706
Ribo-Zero Gold rRNA Removal Kit (Epidemiology)	Cytoplasmic and mitochondrial	24	MRZE724

Ribo-Zero rRNA Removal Kit (Bacteria)

The Ribo-Zero Kit (Bacteria) removes rRNA and cytoplasmic (nuclear-encoded) rRNAs from intact and partially degraded mixtures of Gram-positive and Gram-negative bacteria in a single pass. Sequencing data from samples treated with the Ribo-Zero Kit (Bacteria) contain both coding and multiple forms of noncoding RNA.

Ordering information

Product	Ribosomal removal	No. of samples	Catalog no.
Ribo-Zero rRNA Removal Kit (Bacteria)	Cytoplasmic	6	MRZMB126
Ribo-Zero rRNA Removal Kit (Bacteria)	Cytoplasmic	24	MRZB12424

Ribo-Zero Gold rRNA Removal Kit (Yeast)

The Ribo-Zero Gold rRNA Removal Kit (Yeast) is optimized to deplete rRNA, including cytoplasmic and mitochondrial rRNA, from a range of yeast species. Add the Ribo-Zero Gold Kit (Yeast) to the TruSeq Stranded mRNA Kit library prep workflow to remove rRNA from total RNA, enabling users to focus on species of interest and deep analysis of the complex yeast transcriptome. Focused sequencing data enables characterization a novel and low-abundance transcripts.

Ordering information

Product	Ribosomal removal	No. of samples	Catalog no.
Ribo-Zero Gold rRNA Removal Kit (Yeast)	Cytoplasmic and mitochondrial	6	MRZY1306
Ribo-Zero Gold rRNA Removal Kit (Yeast)	Cytoplasmic and mitochondrial	24	MRZB12424

Proof-of-Concept Service

The POC Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at our Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

Contact your local representative to learn more about Illumina products and services available in your region.
800.809.4566 (North America) • 01799 534332 toll-free (Europe, Middle East, Africa) • +61.3.9212.9900 (Australia)
+65.6773.0188 (Singapore) • +81.3.4578.2800 (Japan) • +86.21.6032.1066 (China)

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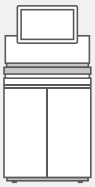
mRNA sequencing

Obtain a clear and complete view of the coding transcriptome

Key strengths

- Enables efficient, comprehensive analysis of the coding transcriptome
- Offers a broad dynamic range that provides high sensitivity and fold change accuracy
- Captures both known and novel features
- Provides an established source of informative biomarkers for a range of phenotypes

Prepare library	Sequence	Analyze and Report
		
TruSeq Stranded mRNA Library Prep Kit **TruSeq RNA Exome Library Prep Kit**	**MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on BaseSpace Sequence Hub** RNA-Seq Gene expression profiling
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training TruSeq Stranded mRNA Library Prep Kit	**Tiered service plans** MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/mRNA for more details.

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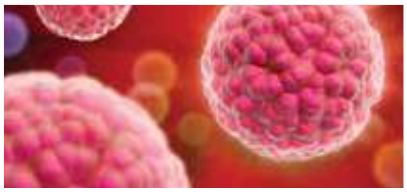
Innovation. Discovery. Application.



Cancer classification and progression (cancer research)

Cancers accumulate numerous genetic changes, but, typically, only a few changes actually drive tumor progression. Monitoring cancer gene expression and transcriptome changes with RNA-Seq can help answer research questions on disease classification and progression.

Gene expression (agrigenomics)

The gene expression profiles in plant and animal cells change through development, in disease, and in response to a broad range of external stimuli. mRNA sequencing enables researchers to capture these changes with high sensitivity and accuracy.

Gene mapping (complex disease)

Differences in gene expression are linked to phenotypic variation between individuals. Differences in gene expression impact regions of the genome involved in the regulation of mRNA expression levels, allowing researchers to better understand the biological mechanisms contributing to complex disease.

Alternative RNA splicing (complex disease)

Alternative RNA splicing has been implicated in a range of complex diseases, including autism and Alzheimer's disease. mRNA sequencing allows scientists to identify both known and previously unidentified splice variants and assess the impact of these events on transcriptome-wide expression patterns.

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STEP 1
Choose a library prep method

Product	TruSeq Stranded mRNA Library Prep Kit	TruSeq RNA Exome Library Prep Kit
Most important to me	Complete coding transcriptome at lowest[a] library prep cost	Complete coding transcriptome in FFPE samples at lowest[a] all-in cost, lowest[a] input requirement
Least important to me	FFPE compatibility	Total assay time
FFPE compatibility	No	Yes
RNA input	0.1–1 µg high-quality total RNA 10–400 ng previously isolated mRNA	10 ng high-quality total RNA 20 ng degraded total RNA
Assay time	9 hrs	2.5 days
Stranded	Yes	Yes

a. Comparisons are within the Illumina RNA-Seq library prep portfolio

STEP 2
Choose a sequencer

Product	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System
Most important to me	Instrument affordability mRNA-Sequencing for small genomes, onboard informatics	mRNA-sequencing for small genomes, read length flexibility, onboard Informatics	RNA-Sequencing for a broad range of genome sizes, Instrument affordability	Lowest[a] cost per sample, scalable for small and large scale research project and operations
Max mRNA samples processed/flow cell (20 M reads per sample)[b]	1	1	2–8	384[c]
Max Gene expression profiling/run (10 M reads per sample)	2–3	2–3	12–36	384[c]
Run mode/kit type	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	S1, S2, S4
Flow cells processed/run	1	1	1	1 or 2

a. Comparison among Illumina portfolio based on individual lane indexing and S4 flow cells

b. Assumes a human-sized transcriptome

c. Limited by available indexes

Visit www.illumina.com/mRNA for more details.

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STEP 3
Obtain results

RNA-Seq ▼

1 Align/Call variants

TopHat Alignment B
- Maps reads and calls small variants (SNVs and small indels)
- Performs abundance estimations of reference genes and transcripts
- Offers optional fusion calling

2 Assemble/Count

Cufflinks Assembly & DE B
- Assembles novel transcripts
- Performs DE of novel and reference transcripts

Gene expression profiling ▼

Align/Count

RNA Express B
- Aligns RNA-Seq reads with the STAR aligner and assigns aligned reads to genes
- Performs differential gene expression with DESeq2

3 Interpret/Report

BaseSpace Cohort Analyzer
- Provides a web-based platform for analyzing human data for translational research applications
- Provides access to a large repository of curated public data sets in the world and powerful tools for large cohort analysis and group comparisons of both public and proprietary data

BaseSpace Correlation Engine
- Provides a web-based platform with an interactive data analysis environment that integrates your data with vast amounts of research data
- Compares Omics molecular profiles from your own experiments with results from a large curated repository of public data assets

B BaseSpace Sequence Hub

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Library prep ordering information

TruSeq Stranded mRNA Library Prep Kit

The TruSeq Stranded mRNA Library Prep Kit provides a clear and complete view of the transcriptome with a streamlined, cost-efficient, and scalable solution for mRNA analysis. The kit supports precise measurement of mRNA strand orientation for detecting antisense transcription, enhanced transcript annotation, and increased alignment efficiency. High coverage uniformity enhances the discovery of features such as alternative transcripts, gene fusions, and allele-specific expression. Libraries prepared with the TruSeq Stranded mRNA Library Prep Kit are compatible with the MiSeq System and NextSeq 550 System. Library preparation requires one library preparation component and one index component.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Preparation Component			
TruSeq Stranded mRNA Library Prep Kit		48	20020594
TruSeq Stranded mRNA Library Prep Kit		96	20020595
Index Component			
TruSeq RNA Single Indexes Set A	12		20020492
TruSeq RNA Single Indexes Set B	12		20020493
TruSeq RNA CD Indexes	96		20019792
IDT for Illumina – TruSeq RNA UD Indexes	24	96	20020591
IDT for Illumina – TruSeq RNA UD Indexes	96	96	20022371

Training	
TruSeq Stranded mRNA Library Prep Kit training at customer site	TR-204-0013

TruSeq RNA Exome Library Prep Kits

TruSeq RNA Exome Library Prep Kits use proven TruSeq Stranded RNA Library Prep Kit chemistry combined with efficient sequence-specific captures to generate RNA-Seq libraries that focus on the RNA coding regions from degraded samples. TruSeq RNA Exome Kits require as little as 10 ng of input from intact RNA samples or 20 ng of input RNA from degraded samples. Libraries prepared with the TruSeq RNA Exome Kit are compatible with the MiSeq System and NextSeq 550 System. Library preparation requires a library preparation component, enrichment component, probe panel, and index component.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Preparation Component			
TruSeq RNA Library Prep for Enrichment			20020189
Enrichment Component			
TruSeq RNA Enrichment			20020490
Probe Panel			
Exome Panel (45 Mb)			20020183
Index Component			
TruSeq RNA Single Indexes Set A	12		20020492
TruSeq RNA Single Indexes Set B	12		20020493

Proof-of-Concept Service

The POC Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at our Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
MiniSeq POC Service	20003924
MiSeq POC Service	SP-801-1002
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	20016091

Targeted RNA sequencing

Design targeted RNA expression panels to focus on RNA sequences of interest or add custom content to fully optimized and experimentally validated panels

Key strengths

- Offers a highly accurate and specific method for measuring transcripts of interest
- Uses qualitative and quantitative information to allow analysis of differentially expressed genes (DE analysis), allele-specific expression measurement, and fusion gene verification
- Measures dozens to thousands of targets simultaneously
- Supports low-quality or FFPE-derived RNA samples

Prepare library	Sequence	Analyze and Report
		
TruSeq Targeted RNA Expression Kits See page 75 for a complete list of targeted RNA expression kits **AmpliSeq for Illumina Transcriptome Human Gene Expression Panel**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software and BaseSpace Sequence Hub Platform** Targeted RNA-Seq App
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

TruSeq Targeted RNA Expression Kits

Custom Kit	Cell Cycle Panel Kit	Neurodegeneration Panel Kit	TP53 Panel Kit
Apoptosis Panel Kit	Cytochrome p450 Panel Kit	NFkB Stem Cell Panel Kit	TWNT Pathway Panel Kit
Cardiotoxicity Panel Kit	Hedgehog Panel Kit		

Training	Service contracts	Professional services
Customer site training TruSeq Targeted RNA Expression Library Prep Kit with the MiSeq System	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/targetedexpression for more details.

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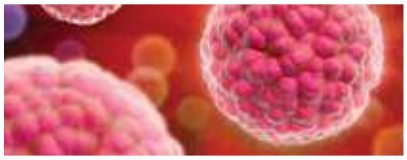

Disease progression (cancer research)
Mapping gene expression and transcriptome changes in cancer is important for understanding tumor classification and progression. Use cancer RNA-Seq to help determine which variants are expressed in cancer samples.

Neural disorders (complex disease)
Neurodegenerative and neurotoxic pathways in the brain are implicated in degenerative disorders such as Alzheimer's disease. With specifically designed targeted RNA expression panels, scientists are able to gain insights into whether these pathways are compromised within study populations.

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STEP 1

Product	TruSeq Targeted RNA Expression Kits		
Input RNA quantity	50 ng	**Multiplexing**	Up to 384
FFPE compatibility	Yes	**Number of assays per panel**	12–1000
Assay time	1.5 d	**Targets per sample**	12–2000
Hands-on time	4 h	**Design**	Add custom content by designing probes targeting regions of interest with DesignStudio Software

STEP 2
Choose a sequencer





Product	iSeq 100 System	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System
Most important to me	Affordability	Onboard informatics, instrument affordability, simplicity	Onboard informatics, read length flexibility, simplicity	Low cost per sample for targeted RNA, flexibility for other methods	High throughput and low cost per sample, using the latest sequencing architecture
Run mode/kit type	Standard	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	Standard

STEP 3
Obtain results

Targeted RNA-Seq ▼

Align/Count

Targeted RNA Kit workflow L
and TruSeq Targeted RNA B

- Supports TruSeq Targeted RNA Kit methods, such as the Hedgehog Panel, Neurodegeneration Panel, and custom RNA panels built in DesignStudio Software
- Detects specific transcript isoforms and gene fusions and reports relative expression values

B BaseSpace Sequence Hub L Local Run Manager

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Library prep ordering information

TruSeq Targeted RNA Expression Custom Components

TruSeq Targeted RNA Expression Kits offer highly customizable mid- to high-plex gene expression profiling and validation, and overcome significant challenges in cost and workflow. These assays deliver a fully integrated solution, including convenient online panel design and ordering, a rapid and simple workflow starting with as little as 50 ng of RNA, and automated on-instrument data analysis. User-defined panels of 12 to 1000 assays can be created to target genes, isoforms, splice junctions, coding region SNPs (cSNPs), and fusion genes with the ability to multiplex up to 384 samples. Libraries prepared with TruSeq Targeted RNA Expression Custom Components are compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of samples	Catalog no.	No. of samples	Catalog no.
TruSeq Targeted RNA Expression Custom Components				
TruSeq Targeted RNA Custom Kit	48	RT-101-1001	96	RT-102-1001
TruSeq Targeted RNA Supplemental Content	48	RT-801-1001	96	RT-802-1001
TruSeq Targeted RNA Expression Fixed Panel Kits				
TruSeq Targeted RNA Apoptosis Panel Kit	48	RT-201-1010	96	RT-202-1010
TruSeq Targeted RNA Cardiotoxicity Panel Kit	48	RT-201-1009	96	RT-202-1009
TruSeq Targeted RNA Cell Cycle Panel Kit	48	RT-201-1003	96	RT-202-1003
TruSeq Targeted RNA Cytochrome p450 Panel Kit	48	RT-201-1006	96	RT-202-1006
TruSeq Targeted RNA Hedgehog Panel Kit	48	RT-201-1002	96	RT-202-1002
TruSeq Targeted RNA Neurodegeneration Panel Kit	48	RT-201-1001	96	RT-202-1001
TruSeq Targeted RNA NFKB Panel Kit	48	RT-201-1008	96	RT-202-1008
TruSeq Targeted RNA Stem Cell Panel Kit	48	RT-201-1005	96	RT-202-1005
TruSeq Targeted RNA TP53 Pathway Panel Kit	48	RT-201-1007	96	RT-202-1007
TruSeq Targeted RNA Wnt Pathway Panel Kit	48	RT-201-1004	96	RT-202-1004
TruSeq Targeted RNA Expression Index Kits			**No. of indexes**	
TruSeq Targeted RNA Index Kit			48	RT-401-1001
TruSeq Targeted RNA Index Kit A			96	RT-402-1001
TruSeq Targeted RNA Index Kit B			96	RT-402-1002
TruSeq Targeted RNA Index Kit C			96	RT-402-1003
TruSeq Targeted RNA Index Kit D			96	RT-402-1004
Training				
TruSeq Targeted RNA Expression Library Prep Kit with the MiSeq System				TR-204-0017

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AmpliSeq for Illumina Transcriptome Human Gene Expression Panel

The AmpliSeq for Illumina Transcriptome Human Gene Expression Panel is a targeted resequencing assay for quantitating gene expression. Starting with as little as 1 ng total RNA (10 ng recommended), the AmpliSeq for Illumina Transcriptome Human Gene Expression Panel yields sensitive, accurate results for gene expression studies, even from low-quality or limited quantity samples, including formalin-fixed, paraffin-embedded (FFPE) tissues. Libraries prepared with the AmpliSeq for Illumina Transcriptome Human Gene Expression Panel are compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Transcriptome Human Gene Expression Panel		24	20019170
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina cDNA Synthesis		96	20022654

Proof-of-Concept Service

The POC Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can empower your laboratory. Receive in-person consultation pre-POC to establish POC needs and goals, POC sequencing services of your samples at the Illumina Service Lab, and in-person consultation post-POC to review sequencing run QC, data, and reports based on your samples. Contact your local Illumina representative for more information.

Ordering information

Product	Catalog no.
iSeq 100 POC Service	20023613
MiniSeq POC Service	20003924
MiSeq POC Service	SP-801-1002
NextSeq POC Service	SP-801-1003
NovaSeq POC Service	2001691

Small RNA sequencing

Generate small RNA libraries directly from total RNA to understand the role of noncoding RNA

Key strengths

- Understand how posttranscriptional regulation contributes to the phenotype of interest
- Identify novel biomarkers
- Capture complete microRNA (miRNA) transcriptome

Prepare library	Sequence	Analyze and Report
		
TruSeq Small RNA Library Prep Kit	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software and BaseSpace Sequence Hub**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Coming soon	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/miRNA for more details.

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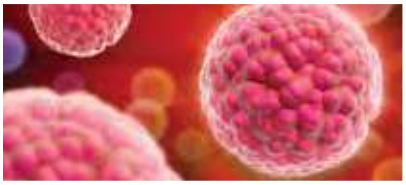

Mutation detection (cancer research)
Researchers often want to analyze precious tumor samples with high sensitivity to detect mutation levels as low as a single copy per cell. miRNA-Seq enables high-throughput profiling or deep-coverage discovery studies of the cancer miRNA transcriptome.

RNA editing and mutation (complex disease)
miRNAs have unique expression profiles in cells of the innate and adaptive immune systems, central nervous system, and a wide range of diseases, including psychiatric disorders. NGS-based profiling enables rapid, high-throughput analysis of the miRNA transcriptome, including insights on RNA editing processes and mutational events, while higher-depth sequencing can be used to discover novel miRNA species.

Analysis

microRNA-Seq ▼

Align/Count

Small RNA B and TruSeq Small RNA Kit workflow L
- Aligns reads against databases for contaminants, mature miRNA, small RNA, and a genomic reference using Bowtie

Library prep ordering information

TruSeq Small RNA Library Prep Kits

TruSeq Small RNA Library Prep Kits provide a simple, cost-effective solution for generating small RNA libraries directly from total RNA.

Ordering information

Product	No. of indexes	Catalog no.
TruSeq Small RNA Library Prep Kit (indexes 1–12)	12	RS-200-0012
TruSeq Small RNA Library Prep Kit (indexes 13–24)	12	RS-200-0024
TruSeq Small RNA Library Prep Kit (indexes 25–36)	12	RS-200-0036
TruSeq Small RNA Library Prep Kit (indexes 37–48)	12	RS-200-0048

See page 149 for Proof-of-Concept Service information.

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B BaseSpace Sequence Hub L Local Run Manager Software

Ribosome profiling

Identify transcripts undergoing translation for visibility to an important layer of gene regulation

Key strengths

- Simplifies translational control research
- Measures gene expression
- Identifies translation start sites
- Predicts protein abundance
- Enables translational and cotranslational process investigation *in vivo*

Prepare library	Sequence	Analyze and Report
		
TruSeq Ribo Profile Kit (Yeast) **TruSeq Ribo Profile Kit (Mammalian)**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**BaseSpace Sequence Hub analysis tools coming soon**
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Coming soon	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Proof-of-Concept Service** Standard application functional testing with your samples

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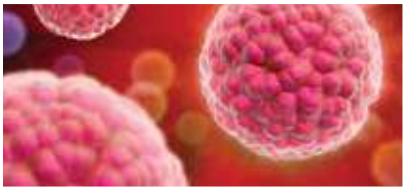

Gene expression dysregulation (cancer research)

Sequencing ribosome-protected mRNA fragments provides critical insight into gene dysregulation in cancer.[1] By measuring the number of ribosomes engaged in translating specific mRNAs, researchers gain insight into functional changes occurring during the development of cancer.

Translational regulation (complex disease)

Regulation of the translation of expressed genes is an important modulator of protein abundance. Ribosome profiling further bridges the gap between genomics/transcriptomics and proteomics, providing visibility to an important layer of posttranscriptional regulation and potential new insights into the mechanisms driving complex disease.

Reference

1. Hsieh AC, Liu Y, Edlind MP, Ingolia NT, Janes MR, Sher A, et al. The translational landscape of mTOR signaling steers cancer initiation and metastasis. *Nature*. 2012;485(7396):55-61.

Library prep ordering information

TruSeq Ribo Profile Kits

TruSeq Ribo Profile Kits capture the portion of the transcriptome associated with ribosomes in preparation for sequencing, providing visibility to an important layer of posttranscriptional regulation. Order the appropriate Ribo-Zero Gold rRNA Removal Kit for your sample. TruSeq Ribo Profile Kits are most compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of samples	Catalog no.
TruSeq Ribo Profile Kit (Mammalian)	12	RPHMR12126
TruSeq Ribo Profile Kit (Yeast)	12	RPYSC12116
Ribo-Zero Gold rRNA Removal Kit (Human/Mouse/Rat)	24	MRZG12324
Ribo-Zero Gold rRNA Removal Kit (Human/Mouse/Rat)	6	MRZG126
Ribo-Zero Gold rRNA Removal Kit (Yeast)	24	MRZY1324
Ribo-Zero Gold rRNA Removal Kit (Yeast)	6	MRZY1306

> Ribosome profiling captures protein abundance information unavailable to mRNA measurements.
>
> **Nicholas Ingolia, PhD**, UC Berkeley

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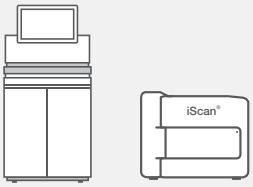
Visit www.illumina.com/ribsomeprofiling for more details.

DNA methylation

Explore the methylome with flexible tools for any project

Key strengths

- Understand the drivers behind gene expression with targeted or genome-wide DNA methylation analysis
- Generate high-quality, reproducible methylomes across tissue types, disease states, and developmental states
- Complement genomic sequencing or RNA-Seq assays for a complete view of genetics and gene regulation
- Identify potential biomarkers for a range of phenotypes

Prepare library	Sequence	Analyze and Report
		
TruSeq Methyl Capture EPIC Library Prep Kit **Infinium MethylationEPIC Kit**	**NextSeq 550 System** **NovaSeq 6000 System** **iScan System**	**Available on BaseSpace Sequence Hub** DNA methylation analysis
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Coming soon	**Tiered service plans** NextSeq 550System NovaSeq 6000 System iScan System	**Proof-of-Concept Service** Standard application functional testing with your samples

Visit www.illumina.com/epigenetics for more details.

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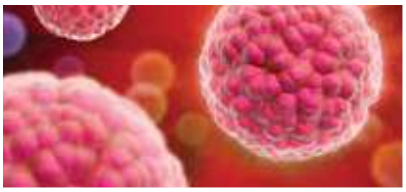

Cancer epigenetics (cancer research)
Studies of cancer epigenetics, such as aberrant methylation and altered transcription factor binding, can provide insight into important tumorigenic pathways. NGS can detect altered methylation patterns and other epigenetic changes in cancer.

Food security (public health and epidemiology)
Environmental changes like deforestation and climate change affect the growth of crops in different areas of the world. Environmental scientists use methylation sequencing to investigate which stress genes in food crops are downregulated in challenging environmental conditions to develop crops adapted to changing conditions.

Plant and animal health (agrigenomics)
DNA methylation regulates development, disease, and environmental responses in plants and animals. Through methylation, researchers can study the health of plant seeds or animal embryos, monitor growth and development over time, and assess disease states and nutrient needs.

Disease risk (complex disease)
DNA methylation has emerged as a consistent and clear contributor to both the risk and development of complex disease. Methylation analysis can identify key methyl markers associated with neurological disorders, such as addiction and schizophrenia, developmental disorders such as autism and metabolic disease, and degenerative diseases such as heart disease and Alzheimer's disease.

STEP 1
Choose a library prep method

Product	Infinium MethylationEPIC BeadChip Kit[a]	TruSeq Methyl Capture EPIC Library Prep Kit
Most important to me	High throughput, large sample number	Cost-effective biodiscovery
Least important to me	Complete methylome coverage	Coverage of regions without known biological importance
No. of CpGs covered	850,000	3.3 M
Input	250 ng FFPE DNA, 500 ng standard DNA	500 ng DNA
FFPE compatibility	Yes	No
Throughput	8 samples/chip	8 samples/run on NextSeq 550 System
Scalability	Automated	Manual
Species	Human	Human

a. The Infinium HumanMethylation450 Array is run on the iScan System. See www.illumina.com/systems/iscan.html for more information

STEP 2
Choose a sequencer

Product	NextSeq 550 System		NovaSeq 6000 System		
Most important to me	Capability for whole-genome and targeted Methyl-Seq, flexibility for other methods		Scalable throughput and flexibility for virtually any genome, sequencing method, and scale of project		
Run mode/kit type	Mid-output	High-output	S1 Xp[a]	S2 Xp[a]	S4 Xp[a]
Human methylation samples sequenced at > 30× coverage per flow cell	1	1	8	8	24
EPIC methyl capture/flow cell	2	8	24[b]	24[b]	48[b]
Flow cells processed/run	1	1	1 or 2		

a. Values supplied represent sequencing performed using the NovaSeq Xp workflow; the standard workflow will have lower throughput values

b. Index limited

STEP 3
Obtain results

DNA methylation analysis (NGS) ▼

DNA methylation analysis (array) ▼

1 Align/Annotate

MethylSeq v2 B
- Conducts sequencing-based DNA methylation analysis that applies the coverage density and flexibility enabled by NGS to enhance epigenetic studies
- Uses Bismark to map bisulfite-treated sequencing reads to the genome of interest and call methylation sites
- Obtains validated data from the TruSeq DNA Methylation Library Prep Kit only

MethylKit App v2
- Analyzes sequencing data for differences in methylation between samples

Align/Annotate/Visualize

Methylation module (GenomeStudio™ Software)
- Generates percent-methylated information for each CpG on the array
- Analyzes integrated controls in a simple graphical module
- Compares methylation across samples on an array
- Creates heat maps, compare samples, and integrate gene expression within one viewer

2 Visualize

The Broad's Integrative Genomics Viewer (IGV) B
- Enables interactive exploration of large, integrated genomic data sets
- The Broad Institute and Harvard developed the app, and Illumina integrated it into the BaseSpace Platform

Library prep ordering information

TruSeq Methyl Capture EPIC Library Prep Kit

The TruSeq Methyl Capture EPIC Library Prep Kit is an enrichment-based targeted sequencing assay that spans the full human methylome. Targeted methylation sequencing (Methyl-Seq) offers a balanced, cost-effective choice between whole-genome bisulfite sequencing and methylation arrays that can support both screening and biomarker discovery study objectives. Libraries prepared with the TruSeq Methyl Capture EPIC Library Prep Kit are compatible with the NovaSeq 6000 System.

Ordering information

Product	Catalog no.
TruSeq Methyl Capture EPIC Library Prep Kit (12 samples)	FC-151-1002
TruSeq Methyl Capture EPIC Library Prep Kit (48 samples)	FC-151-1003

B BaseSpace Sequence Hub

Infinium MethylationEPIC Kit

The unique combination of the comprehensive, expert-selected coverage, high sample throughput capacity, and affordable price of the Infinium Human MethylationEPIC BeadChip Kit make it an ideal solution for large sample-size genome-wide DNA methylation studies. Libraries prepared with the kit are compatible with iScan™ System.

Ordering information

Product	No. of samples	Catalog no.
Infinium MethylationEPIC BeadChip Kit	16	WG-317-1001
Infinium MethylationEPIC BeadChip Kit	32	WG-317-1002
Infinium MethylationEPIC BeadChip Kit	96	WG-317-1003

See page 149 for Proof-of-Concept Service information.

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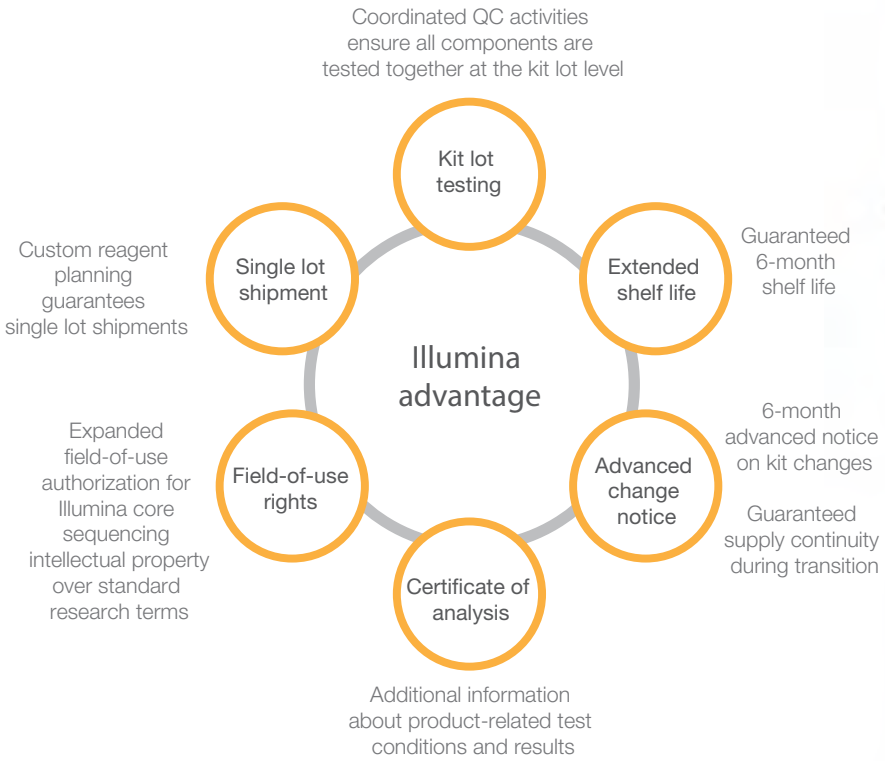
Illumina advantage

Streamline your workflow. Increase your productivity.

Illumina is committed to meeting the unique needs of your laboratory. Several key products now enhance productivity and decrease the costs of consumables used in clinical research. For further information, contact your local account manager.



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Protein–DNA interactions

Quickly and efficiently determine the distribution of DNA-bound protein targets across the genome using ChIP-Seq

Key strengths

- Capture DNA targets for transcription factors or histone modifications across the entire genome of any organism
- Define transcription factor binding sites
- Reveal gene regulatory networks in combination with RNA-Seq and methylation analysis
- Support a wide source of input DNA samples

Prepare library	Sequence	Analyze and Report
		
TruSeq ChIP Library Preparation Kit	**MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on BaseSpace Sequence Hub** DNA/Protein interactions
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training Coming soon	**Tiered service plans** MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**POC Service** Standard application functional testing with your samples

Visit www.illumina.com/epigenetics for more details.

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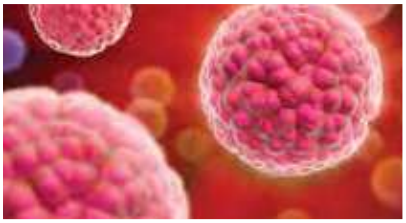

Tumor development and progression (cancer research)

In cancer, the regulation of gene expression is critical to tumor development and progression. Chromatin immunoprecipitation sequencing (ChIP-Seq) provides a sensitive and unbiased method for assessing proteins bound to DNA and their effect on gene regulation using transcription factor regulon mapping, defining binding sites, and correlating changes in histone state to tumor progression.

Protein–DNA interactions (complex disease)

Elucidating the key contributors to complex disease requires a complete view of cell biology. With ChIP-Seq, the intricate interactions of protein and DNA, and changes in the interactions based on cell type, environment, or disease, can be teased out.

Gene regulation (agrigenomics)

The epigenome plays a critical role in plant and animal gene regulation. With ChIP-Seq, researchers can identify the portions of the genome critical to gene regulation, determine the genetic effect of environmental and nutritional changes, and track the physiology of disease and infection.

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STEP 1
Library prep method

Product	TruSeq ChIP Library Prep Kit
Input requirement	5–10 ng DNA
Indexes	24
Input DNA size range	200–800 bp
No. of reads required/sample[a]	5–15M for targeted proteins such as transcription factors > 40M for histone marks and ubiquitous proteins
Assay time	5 hrs
Hands-on time	2.5 hrs

a. Assuming a human-sized genome

STEP 2
Choose a sequencer

Product	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System
Most important to me	Instrument affordability and capacity for small genome ChIP-Seq	Simplicity and capacity for small genome ChIP-Seq with read length options for other applications	Low cost per sample for ChIP-Seq, flexibility for other methods	High throughput and low cost per sample, using the latest sequencing architecture
Run mode/kit type	Mid-output/High-output	Mid-output/High-output	Mid-output/High-output	Standard
Flow cells processed/run	1	1	1	1 or 2

STEP 3
Obtain results

DNA/Protein interactions ▼

1 Align/Call variants

Isaac Whole Genome Sequencing B BSO
- Performs alignment, and small and large variant calling using algorithms developed by Illumina
- Offers 4× faster alignment speed with the same accuracy as the BWA Whole Genome Sequencing App

BWA Whole Genome Sequencing B BSO
- Analyzes ChIP-Seq data using Sanger's BWA alignment algorithm combined with the Broad's GATK small variant calling algorithm

2 Export/Call peaks

Export BAM files from BaseSpace Platform
- Predicts protein–DNA interaction sites from alignments of ChIP-Seq data to a reference
- Zhang Y, Liu T, Meyer CA, Eeckhoute J, Johnson DS, Bernstein BE, et al. Model-based analysis of ChIP-Seq (MACS). *Genome Biol*. 2008;9(9):R137. doi: 10.1186/gb-2008-9-9-r137
- MACS - Model-based Analysis for ChIP-Seq. liulab.dfci.harvard.edu/MACS/index.html. Accessed May 30, 2018

3 Add biological context

The Broad's IGV B BSO
- Enables interactive exploration of large, integrated genomic data sets
- The Broad Institute and Harvard developed the app, and Illumina integrated it into the BaseSpace platform

Library prep ordering information

TruSeq ChIP Library Prep Kits

TruSeq ChIP Library Prep Kits provide a simple, cost-effective solution for generating ChIP-Seq libraries from ChIP-derived DNA. Offering the proven data quality and ease of use of TruSeq DNA Kit sequencing, the kits provide a streamlined ChIP-Seq library prep workflow that leverages reagent master mixes to minimize pipetting and reduce total assay time. The kits have a low DNA input requirement with a range of sample sources. In addition, they offer robust multiplexed sequencing, allowing researchers to optimize the distribution of sequencing output across samples based on read depth requirements. Libraries prepared with TruSeq ChIP Library Prep Kits are compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSeq ChIP Library Prep Kit, set A	12	48	IP-202-1012
TruSeq ChIP Library Prep Kit, set B	12	48	IP-202-1024

See page 149 for Proof-of-Concept Service information.

B BaseSpace Sequence Hub BSO BaseSpace Sequence Hub Onsite

Infinium BeadChip technology

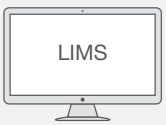
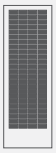
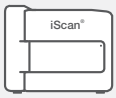
High-throughput SNP genotyping, methylation, and CNV analysis for multiple genomic applications

Infinium BeadChips are ideal for processing thousands of samples quickly and cost effectively to identify variants and some structural variants. Additional advantages include:

- Reproducible, high-quality data at a low per-sample cost
- Fast, high-throughput multiplex processing
- Expert-selected content
  - Coverage of diverse world populations
  - Extensive coverage of common and rare disease conditions
- Custom and semicustom formats
- Fast turn around time from DNA to genotype calls
  - Easy workflow
- Intuitive analysis tools to visualize and analyze data

Key strengths

- Genotype plexity ranging from 500–5M probes
- Ability to detect SNPs, methylation status, CNV, and small insertions/deletions
- High concordance, accuracy, and reproducibility
- Multiple-sample processing with each Infinium BeadChip

Accession and Track	Prepare Sample and Hybridize	Scan BeadChips	Analyze
			
LIMS	**Infinium BeadChips** **Tecan automation** **Automation options** Tecan EVO 150 Base Unit (non-LIMS) Tecan EVO 150 Base Unit (LIMS ready)	**iScan System** **Autoloader 2.x**	**GenomeStudio Software** **Beeline Software** **BlueFuse Multi Software**

Visit www.illumina.com/techniques/microarrays.html for more details.

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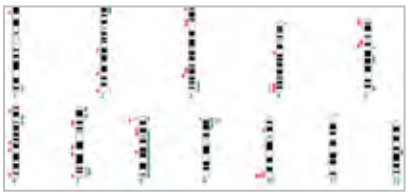
Infinium Human Genotyping Products

Population genetics

Illumina human genotyping arrays contain content supporting whole-genome, targeted genome, and exome analyses. Powered by BeadArray™ and Infinium assay technologies, they deliver exceptional data quality and high-density genomic coverage. Choose from ready-to-use BeadChip arrays with expert-defined content for specific applications or design iSelect™ custom or semicustom microarrays to suit specific research needs. Supporting high-throughput, multiplex processing, Illumina microarrays are ideal for pre-emptive screening research, large-scale population studies, and genome-wide and phenome-wide association studies (GWAS and PheWAS) performed by researchers and biobanks. They also offer the high resolution required for detection of copy number variation (CNV), absence of heterozygosity (AOH), and small indels.

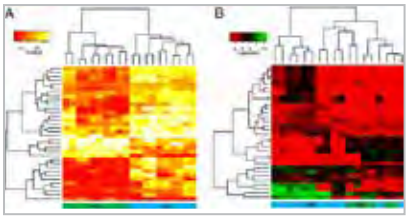
Copy number variation studies

CNVs are genomic alterations that result in an abnormal number of copies of one or more genes. Structural genomic rearrangements such as duplications and deletions can cause CNVs. Like SNPs, certain CNVs have been associated with disease susceptibility. Structural and numerical genetic variations of dosage-sensitive genes are thought to influence the presentation of a phenotype. Accurate profiling of CNV events is crucial for studies associated with cancer and genetic disease.



Fine mapping and candidate gene region genotyping

Genotyping can be used to fine map traits and/or test a genotype–phenotype hypothesis. Quantitative trait loci (QTL) analyses can be performed on the results to characterize how differential gene expression might contribute to phenotypic variation.



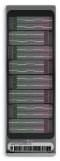
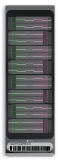
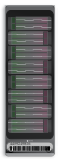
Methylation analysis

Methylation studies enable researchers to understand the functional mechanisms at work in complex disease and metabolism. These are ideal for detecting altered methylation patterns that provide insight into disease pathways and regulation of gene expression.

Human BeadChip Portfolio

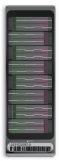
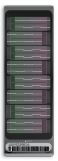
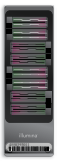
Genome-Wide Human Genotyping BeadChip



Product	Asian Screening Array	Global Screening Array	Multi-Ethnic Global-8 Array	Multi-Ethnic AMR/ AFR-8 Array	Multi-Ethnic EUR/ EAS/SAS-8 Array
Description	East Asian–focused population-scale genetics, variant screening, and precision medicine research	Population-scale genetics, variant screening, and precision medicine research	Complex disease in global populations	Complex disease in Hispanic and African American populations	Complex disease in European, East Asian, and South Asian populations
Key applications	Health screening and precision medicine research, direct to consumer reporting, association studies	Health screening and precision medicine research, association studies	Health screening and precision medicine research, association studies	Health screening and precision medicine research, association studies	Health screening and precision medicine research, association studies
Most important to me	Economical clinical research content and powerful East Asian coverage	Economical clinical research content and global coverage	Clinical research content and high coverage of global populations	Coverage for Hispanic and African populations	Clinical research content and high coverage of European, East Asian, and South Asian populations
Number of loci + add-on real estate	~660K markers	~660K markers	~1.7M markers	~1.4M markers	~1.4M markers
Add-on availability	50K custom add-on	50K custom add-on	245K custom add-on	245K custom add-on	245K custom add-on
Assay type	HTS	HTS	LCG	LCG	LCG
Content source	9000 East Asian whole-genome sequences, 1000 Genomes–phase 3, ClinVar, PharmGKB, NHGRI-EBI GWAS, ExAC	1000 Genomes–phase 3, ClinVar, PharmGKB, NHGRI-EBI GWAS, ExAC	1000 Genomes–phase 3, ClinVar, PharmGKB, NHGRI-EBI GWAS, ExAC	1000 Genomes–phase 3, ClinVar, PharmGKB, NHGRI-EBI GWAS, ExAC	1000 Genomes–phase 3, ClinVar, PharmGKB, NHGRI-EBI GWAS, ExAC

Abbreviations: AMR = Americas, AFR = Africa, EUR = Europe, EAS = East Asia, SAS = South Asia, HTS = High-throughput screening, LCG = Long cap glass, NHGRI-EBI GWAS = National Human Genome Research Institute-European Bioinformatics Institute Genome-Wide Association Study, ExAC = Exome Aggregation Consortium

Product	OmniExpress	OmniZhongHua	Omni2.5	Omni5
Description	Novel trait and disease association discovery with common tag SNPs at an attractive price	Exceptional coverage of common variation found within Chinese (CHB) populations	Comprehensive coverage of common and intermediate SNPs with focus on high LD coverage across diverse populations	High-density array with exceptional coverage of common, intermediate, and rare SNPs
Key applications	GWAS/CNV	Chinese GWAS/CNV	GWAS/CNV	GWAS/CNV
Most important to me	Economical coverage of common variation	Coverage in East Asian populations	Balance between price and rare content	Highest marker density
Number of loci + add-on real estate	~710K markers	~880K markers	2.4M markers	4.3M markers
Add-on availability	30K custom add-on	Not available	250K custom add-on	500K custom add-on
Assay type	HTS	HD Super	LCG	LCG Quad
Content Source	HapMap, 1000 Genomes–3 phases	HapMap, 1000 Genomes	HapMap, 1000 Genomes	HapMap, 1000 Genomes

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Product	Core-24	CoreExome-24
Description	Economical genome-wide backbone built for customization and high throughput	Economical genome-wide backbone and exonic content built for customization and high throughput
Key applications	Association studies, quality control	Association studies, quality control
Most important to me	Cost, ability to detect common variants	Cost, exome content, ability to detect common variants
Number of loci	~300K markers	~550K markers
Add-on availability	+300K custom add-on	+100K custom add-on
Assay type	HTS	HTS
Content source	1000 Genomes – phase 1	1000 Genomes – phase 1

Targeted human genotyping arrays

Product	Exome	OncoArray	PsychArray	ImmunoArray
Description	Access to more than 240K exonic variants to uncover biologically significant associations	Evaluation of genetic variants associated with five common cancers on a single array	Evaluation of genetic variants associated with common psychiatric disorders	Evaluation of genetic variants associated with 14 major autoimmune and inflammatory diseases
Key applications	Analysis of SNPs in coding regions	Germline cancer research	Psychiatric genetics research	Genomic analysis of immunology related conditions
Most important to me	Exonic content, low cost per sample	Markers associated with germline cancer risk	Markers associated with psychiatric genetics	Markers associated with immunology research
Number of loci	~240K markers	~500K markers	~590K markers	~250K markers
Add-on availability	+400K custom add-on	+120K custom add-on	+50K custom add-on	+390K custom add-on
Assay type	HTS	HTS	HTS	HTS
Content source	12K exome sequences	1000 Genomes – phase I, consortium-developed content	1000 Genomes – phase I, 12K exome sequences, consortium-developed content	Consortium-developed content

Infinium nonhuman genotyping arrays

Flexible and powerful predesigned and custom solutions for virtually any species

Agricultural genomics, or agrigenomics (the application of genomics in agriculture), has and will continue to help drive sustainable productivity and offer solutions to the mounting challenges of feeding the world's growing population. Through the constant development of new products and applications, Illumina is continually innovating ways to help agricultural researchers. Our agrigenomics technologies help plant and animal breeders and researchers identify desirable traits, leading to healthier and more productive crops and livestock.

Innovation. Discovery. Application.

Marker-assisted backcrossing

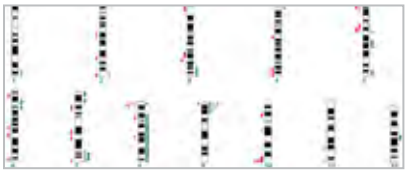
The goal of backcrossing is to move a single trait of interest, such as drought tolerance, high productivity, or disease resistance, from a donor parent to progeny. Marker-assisted backcrossing enables researchers to monitor the transmission of the trait gene via a genetically linked marker that can be easily screened, significantly accelerating backcrossing programs and reducing the time to release of commercially viable plant lines or breeding stock.

Parentage

Genetic markers can be used to identify animals and understand the relationship of offspring to parents. Since a single marker may not yield definitive results, multiple markers are used to increase the probability of identifying the true parent. In linebreeding situations where multiple generations of males or females are present in the herd, the marker results are combined with the breeder's knowledge of possible sires or dams to determine parentage.

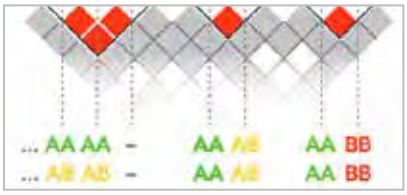
Copy number variation studies

A common form of natural diversity, CNV is heritable. It appears to play an important role in agrigenomics. CNV differences can correlate to health and production traits, and contribute to phenotypic diversity. The distribution of high-density probes can be used to identify CNV breakpoints and ultimately accelerate genetic improvements. Studies are ongoing to determine if the impact of CNVs is the result of differences in copy number for key genetic segments, structural modification of the genome, or a combination of both.



Fine mapping and candidate gene region genotyping

Plant and animal genotyping can be used to create fine map traits and/or test a genotype–phenotype hypothesis. QTL analyses can be performed on the results to characterize how differential gene expression might contribute to phenotypic variation.

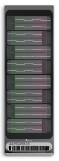


Imputation

Arrays are best designed to select representative SNPs that cover all genetic variations in a population. Using arrays, researchers achieve these results by identifying haplotypes, a collection of alleles that are likely to be inherited all together, that can represent diversity within a set of individuals. Because of their lower costs, simplified analysis, and strong parallel-processing and automation capabilities, DNA arrays and whole-genome SNP imputation are instrumental in analyzing large numbers of samples. Imputation relies on a reference database of fully sequenced genomes to predict genotypes not assayed in a larger sample of individuals. This approach consists of first constructing haplotypes for the samples of interest using the haplotypes from the reference set and then estimating genotypes.

Non-Human Array Portfolio

Standard nonhuman genotyping arrays


Product	GGP Bovine LD	Bovine HD	Bovine LD	Bovine SNP50	Ovine SNP50
Description	Economical array for genomic selection for entire bovine herd	Most comprehensive genome-wide bovine genotyping array for both dairy and beef cattle	Economical array for genomic selection for entire bovine herd	50K array containing highly informative SNP for major cattle breeds	50K array containing highly informative SNP for major porcine breeds
Key applications	Genomic selection, imputation, and parentage	Genomic selection, biodiversity, and genetic mapping	Genomic selection, imputation, and parentage	Genomic selection, biodiversity, genetic mapping, and parentage	Genomic selection, biodiversity, and genetic mapping
Most important to me	Cost-effective solution for whole-genome studies in beef and dairy cattle	Most comprehensive genome-wide genotyping array	Economical imputation tool including parentage markers	Cost-effective solution for whole-genome studies in beef and dairy cattle	Cost-effective solution for whole-genome studies in porcine
Number of loci	~30K	~770K	~8K	~54K	~54K
Add-on availability	–	–	80K	600K	–
Assay type	HTS	HD Super	HD Ultra	HTS	HD





Product	GGP Bovine 150K	GGP Bos Indicus HD	CanineHD	MaizeLD	MaizeSNP50
Description	Ideal array for testing for elite seedstock, donor dams, AI studs, and high-impact cattle	Economical array for genomic selection for entire bovine herd focused on Bos indicus	Array enabling interrogation of genetic variation in any domestic dog breed	Economical array for essentially derived varieties testing in maize	50K array containing highly informative SNP for maize
Key applications	Genomic selection, and parentage	Genomic selection, imputation, and parentage	Genetic mapping and CNV	Mapping essentially derived varieties	Genomic selection
Most important to me	Ideal for testing for elite seedstock, donor dams, AI studs, and high-impact cattle	Economical imputation, including parentage markers focused on Bos indicus	Interrogation of genetic variation in any domestic dog breed	Economical array for essentially derived varieties testing in maize, easily customizable to support other applications	Cost-effective solution for whole-genome studies
Number of loci	~135K	~75K	~170K	~3K	~56K
Add-on availability	–	–	–	70K	–
Assay type	HTS	HD Ultra	HD Ultra	HTS	HD

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Product	ShrimpLD-24	PorcineSNP60 v2	GGP PorcineHD	GGP PorcineLD	GGP GIGA-MUGA
Description	Economical genomic selection tool for Pacific white shrimp	Economical genomic selection tool for porcine	Comprehensive solutions available for genome-wide genotyping in Porcine	Economical imputation tool for porcine	Economical tool for genetic mapping and prediction
Key applications	Genomic selection	Genomic selection and genetic mapping	Genomic selection, genetic mapping, and prediction	Imputation	Genetic mapping, prediction, CNV
Most important to me	Economical genomic selection tool for Pacific white shrimp	Economical genomic selection tool for porcine	Comprehensive solutions available for genome-wide genotyping in Porcine	Economical imputation tool for porcine	Economical tool for genetic mapping and prediction
Number of loci	~6.5K	~65K	~65K	~10K	143K
Add on availability	80K	25K	–	–	–
Assay Type	HTS	HD Ultra	HD Ultra	HTS	HD

Product	GGP Equine
Description	Array enabling identification of genes and polymorphisms that contribute to traits of interest in all major horse breed
Key applications	Genetic mapping and parentage
Most important to me	Genetic mapping and parentage
Number of loci	65K
Add-on availability	–
Assay type	HD Ultra

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Infinium custom genotyping

Custom arrays for genotyping almost any variant, in virtually any species

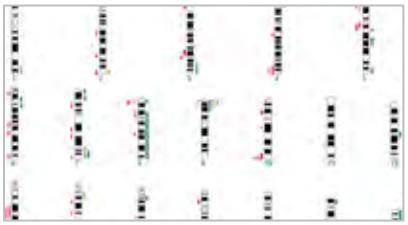
Custom genotyping is the ideal solution for screening large sample sets against novel or targeted content. With custom designs, researchers can target regions of the genome relevant to their specific research interests. Illumina offers flexible options and simple online design tools to maximize success with your customized genotyping assays.

Fully custom arrays can be developed easily with up to 700K markers targeting any loci across the genome, for any species, including novel organisms. Illumina also offers flexible throughput options, as well as the ability to tailor standard array products with add-on content to include novel variants of interest.

Advantages of custom genotyping

- Enables studies of species or populations not supported by standard products
- Allows researchers to focus on genes, variants, and/or genomic regions of interest relevant to certain diseases or traits of interest, but not covered in pre-designed products
- Conserves resources by avoiding irrelevant regions of the genome

Innovation. Discovery. Application.



Fine mapping and candidate gene region genotyping
Genotyping can be used to create fine map traits and/or test a genotype-phenotype hypothesis. QTL analyses can be performed on the results to characterize how differential gene expression might contribute to phenotypic variation.

Custom and Infinium XT Portfolio

Scale and flexibility

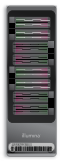



Product	XT	HD	HTS	LCG
Description	Low-plex, ultra-high-throughput product scaling for processing 100K–1M+ samples per year	Targeting 3072–90K SNPs, most popular[a] format for low- to mid-density arrays leveraging low- to high-throughput workflows	Targeting 90,001–700K SNPs, becoming increasing popular for high-density custom panel designs that support high-throughput processing	Supporting 700,001–2.5M custom assays, unique to support strategies that require ultra-high-density assays for maximum coverage across populations
Key applications	Human and agricultural screening within applied markets	Custom design that supports any application, most popular[a] SNP range for agrigenomics applications	Custom design that supports any application, most popular[a] SNP range for healthy human screening applications	Custom design that supports high-density discovery or maximum coverage across human populations
Most important to me	Lowest-cost, highest-throughput[a] array product for focused screening applications	Low-cost options for small panel designs and flexible throughput options (low to high)	Low-cost options for high-density designs used for screening hundreds of thousands to millions of samples	Highest density custom design option in the market[a], requires large sample numbers to be cost effective
Number of loci + add on real estate	200–50K SNPs add-on cannot exceed 50K total SNPs	3072–90K attempted assays	90,001–700K attempted assays	700,001–2.5M attempted assays
Assay type	Infinium iSelect XT	Infinium iSelect HD	Infinium iSelect HTS	Infinium iSelect LCG
Assay order conversion guarantee	200–10K SNPs: 99% > 10K–50K: 95%	80% (higher conversion option available)	80% (higher conversion option available)	80% (higher conversion option available)

a. Comparisons based on the Illumina portfolio

Infinium Cytogenetics

Cost-effective and streamlined screening



Product	CytoSNP-850K	CytoSNP-12	Karyomap-12
Description	Comprehensive coverage for congenital disorders and cancer	**Scalable solution for genome-wide detection of structural variation**	Solution with most informative markers for inheritance of single-gene defects
Key applications	Investigation of genetic disease associated with constitutional and cancer studies	Cytogenetic investigation of prenatal, postnatal, and cancer samples	Preimplantation genetic diagnosis (PGD)
Most important to me	High-resolution cytogenomic analysis for accurate profiling of subtle chromosomal aberrations and structural variants	Cost-effective and scalable cytogenomic studies yielding accurate data and confident results	Rapid PGD solution for single-gene disorders
Number of loci + add-on real estate	850K	300K	300K

Infinium Methylation

High-throughput epigenetic analysis

Product	MethylationEPIC
Description	Comprehensive coverage of CpG islands, RefSeq genes, ENCODE chromatin, ENCODE transcription factor sites, and FAMTOM5 enhancers
Key applications	Methylation status, differential methylation, developmental biology, cancer research, multiomics analysis
Most important to me	Cost-effective, genome-wide methylation analysis at single CpG resolution
Number of loci + add-on real estate	~850K
Assay type	Infinium Methylation HD
Content source	RefSeq, ENCODE, FAMTOM5

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Infinium BeadChip systems

Scalable and streamlined solutions for microarray research

Infinium BeadChip systems are the foundation of the ability to produce data from Infinium BeadChips. Choose from standard and high-throughput systems that enable the processing of genotyping and methylation arrays, or the Infinium XT system, designed specifically for production-scale Infinium processing of 100K to 1M samples per year.

Researchers can configure a system by selecting from option packages of ancillary equipment, liquid-handling automation, the iScan System, scanner loading automation (Autoloader) and Infinium LIMS. A quick discussion with a sales rep can also help narrow down the options depending on your needs and expected throughput.

Hardware Accessories

Hardware accessories required to run Infinium assays are conveniently bundled into starter and upgrade kits. Choose the kit that meets your processing throughput needs.

Accessory package	Product (Quantity)	Catalog no.
Infinium HD Starter Kits	Infinium HD Starter Kit (8 BeadChip batches)	110/220V: 20022217
	Infinium HD Starter Kit (24 BeadChip batches)	110/220V: 20022219
Infinium HTS/LCG Kits	Infinium HTS/LCG Starter Kit (8 BeadChip batches)	110/220V: 20022218
	Infinium HTS/LCG Starter Kit (24 BeadChip batches)	110/220V: 20022220
Infinium XT Kit options	Infinium XT Starter Kit (12 BeadChip batches)	110/220V: 20015525
	Infinium XT Starter Kit (24 BeadChip batches)	110/220V: 20011100
	Infinium XT Starter Kit (48 BeadChip batches)	110/220V: 20011069
	Infinium XT Upgrade Package (12 BeadChip batches)	110/220V: 20015526
	Infinium XT Upgrade Package (24 BeadChip batches)	110/220V: 20011101
Tip guides Required when automating the Infinium workflow or when utilizing the Infinium XT assay, choose the tip guide that matches the assay you are running. For more information about which tip guide to use, reference the relevant support page for your product and/or the Infinium Automation Workflows Tech Note	Infinium HD Super/HD Ultra Assay (12)	SE-104-1005
	Infinium HD Ultra (6)	SE-104-1009
	Infinium HD Super (6)	SE-104-1011
	Infinium LCG (12)	SE-104-1013
	Infinium HTS (8)	SE-104-1015
	Infinium XT (6)	20011102
TeFlow accessories		
Infinium HD	Infinium HD TeFlow Chamber	WG-10-202
	Infinium HD Spacers (500)	WG-10-203
Infinium LCG	Infinium LCG TeFlow Chamber (10)	WG-100-1001
	Infinium LCG Spacers (500)	WG-100-1002
Infinium XT	Infinium XT TeFlow Chamber (8)	20012129
	Infinium XT Glass Back Plates (8)	20011756
Universal	Infinium TeFlow Clips (16)	20011758
	Infinium TeFlow Frames (8)	20011757

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Infinium Automation

Automating the Infinium assay workflow maximizes processing efficiency and minimizes the risk of processing errors. Choose the Infinium Validated Liquid Handling Automation Kit that meets your needs.

Automation platform	Product	Catalog no.	Included
Infinium 8-Tip Robot automation options	Infinium Automation Package – non-LIMS	110V: SC-30-401 220V: SC-30-402	• Illumina customized Tecan EVO 150 Robot with LiHa (not LIMS compatible) • PC workstation • Illumina Robot Control Software • Installation and onsite training • 1-year warranty • Infinium HD Ultra, HD Super, HTS, and LCG Robot tip guides (Infinium XT tip guides sold separately)
	Infinium Automation Package – LIMS ready	110V: SC-30-403 220V: SC-30-404	• Illumina customized Tecan EVO 150 Robot with RoMa (LIMS compatible) • PC workstation • Illumina Robot Control Software • Installation and onsite training • 1-year warranty • Infinium HD Ultra, HD Super, HTS, and LCG Robot tip guides (Infinium XT tip guides sold separately)

Scanner and Scanning Automation

The iScan System provides high-precision, high-throughput array scanning for the Infinium portfolio.

iScan System	Catalog no.
The iScan System supports our broad portfolio of innovative genetic analysis assays for genotyping, CNV analysis, and DNA methylation profiling.	110/220V: SY-101-1001

Integration of the Autoloader 2.x enables 24-hour hands-free scanning of 1 or 2 iScan Systems, depending on your system of choice.

Autoloader 2.x	Product	Catalog no.
Autoloader 2.x options	AutoLoader 2.x, single scanner configuration	(110/220V): SY-202-1002
	AutoLoader 2.x, dual scanner configuration	(110/220V): SY-202-1002

Laboratory Information Management Systems (LIMS)

LIMS provide high-fidelity tracking of important experimental information.

Description	Product	Catalog no.
Integrated LIMS provide a high-speed scalable system that enables positive sample tracking, workflow optimization and enforcement, overall monitoring of sample processing, and laboratory efficiency. Available for Infinium Assays and sequencing systems.	Illumina LIMS Package	20018976
	Illumina LIMS ST Server Upgrade	20018977
	Illumina LIMS HT Server Upgrade	20015563

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Infinium Starter Kits

	Infinium HTS/LCG Assay Starter Kits		Infinium HD Assay Starter Kits		Infinium XT Assay Starter Kits	
Hardware/Accessories	**Quantity**	**Quantity**	**Quantity**	**Quantity**	**Quantity**	**Quantity**
No. of BeadChip batches	8	24	8	24	24	48
Catalog no.	20022218	20022220	20022217	20022219	20011100	20011069
High-speed microplate shaker and accessories	2	2	2	2	2	2
Heat sealer	1	1	1	1	1	1
Heat sealer foils	100	100	100	100	100	100
Staining rack and wash dish	1	2	1	2	1	1
BeadChip wash rack	2	3	2	3	3	6
Glass tray	3	9	3	9	4	8
Hybridization chamber	2	6	2	6	4	8
BeadChip storage box (25 slide capacity)	1	1	1	1	1	3
Hybridization oven with rocker attachment and mat	1	1	1	1	2	4
HybEx heat block incubator	1	2	1	2	6	6
HybEx midi-plate insert	1	2	1	2	6	6
Multi-sample alignment fixture	1	1	1	1	1	1
TeFlow rack, water re-circulator, tubing, and connections	1	1	1	1	1	1
TeFlow chambers (HTS/LCG specific)	10	30	–	–	–	–
TeFlow chamber spacers (HTS/LCG specific)	500	500	–	–	–	–
TeFlow chambers (HD specific)	–	–	10	30	–	–
TeFlow chamber spacers (HD specific)	–	–	500	500	–	–
TeFlow chambers (XT specific)	–	–	–	–	24	48
TeFlow chamber spacers (XT specific)	–	–	–	–	500	500
TeFlow glass back plate plastic drying rack	1	2	1	2	2	4
Vacuum desiccator and accessories	1	3	1	3	2	4
Dissecting scissors	1	2	1	2	1	2
TeFlow chamber dismantling tool	1	2	1	2	2	2
Dissecting forceps	1	1	1	1	1	1

	Infinium XT Upgrade Package (12 BeadChip batches) Catalog no. 20015526	Infinium XT Upgrade Package (24 BeadChip batches) Catalog no. 20011101
Hardware	**Quantity**	**Quantity**
Infinium XT multi-sample BeadChip alignment fixture	1	2
Wash rack	2	3
Glass back plate rack	1	1
Infinium XT TeFlow chambers	16	24
Infinium XT dual hybridization chamber insert and robot baseplate	6	12
Infinium XT tip guides (K,L,M)	6	12
Hybridization chamber	2	4

Notes:
These kits support existing Infinium customers looking to add the Infinium XT Assay workflow to expand their targeted genotyping applications. They include quantities of Infinium XT Assay hardware components to support processing up to 12 or 24 BeadChip X-Stain batches through the Infinium XT Assay workflow.

The iScan System, AutoLoader 2.x, and the automation robot are sold separately.

The tip guide set is included.

BeadChip scan times table

BeadChip	Approximate scan time per BeadChip (minutes)[a]	Maximum iScan throughput per week[b]
Infinium Core-24	30	6048
Infinium CoreExome-24	30	6048
Infinium OmniExpress-24	30	6048
Infinium Global Screening Array-24	30	6048
Infinium PsychArray-24	30	6048
Infinium QC Array-24	30	6048
Infinium iSelect HTS 24-Sample	30	6048
Infinium MethylationEPIC	20	4536
Infinium Multi-Ethnic Global-8	35	1728
Infinium Omni2.5-8	35	1728
Infinium Omni2.5Exome-8	35	1728
Infinium XT iSelect-96	45	16128

a. Scan times are based on configurations associated with iScan System computers installed in 2016 or later with LIMS integration turned off. Scan times may be estimated if scanning times were not available at product launch and inferred from BeadChips of similar plexity.

b. Approximate maximum throughput estimation is based on iScan System scan speeds using the optional AutoLoader. Actual throughput may vary dependent on the level of liquid handling automation, staffing, and working shifts. Contact your local FAS for more information on setting up your lab to meet your throughput goals.

BeadChip informatics

Scalable and streamlined solutions for microarray research

Infinium supporting software

Illumina Laboratory Information Management System (LIMS) package

Array-based LIMS use advanced automation and precise robotic control to facilitate high-throughput microarray processing and sample tracking. Illumina offers a state-of-the-art LIMS that guides sample/data handling and tracking from DNA to genotypes, ensuring reliable, high-quality data outputs and enabling multiple projects to be managed in parallel.

Project managers can easily enter experimental and control samples into the system using familiar sample sheet definitions. The assigned samples are queued automatically for batch processing. This batching system communicates to lab personnel that samples are ready for processing without additional intervention from a project manager.

- 100% sample tracking to prevent sample mix-ups
- Real-time quality metrics from image scanning
- Rapid scale-up options to easily increase sample throughput
- Automatic project archiving
- File management of decoding, image, intensity, and analytical data files

GenomeStudio Software

GenomeStudio Software enables visualization and analysis of microarray data generated on Illumina platforms. The software package is composed of discrete application modules that enable researchers to obtain a comprehensive view of the genome and gene regulation.

- System requirements
  - 8 GB RAM on a 64-bit system
  - Windows Operating System

GenomeStudio Software Genotyping Module

- Analyze SNP and CNV data across up to millions of markers and probes
- Detect sample outliers

The graphical display of genotypes in GenomeStudio Software is a SNP Graph with data points color coded for the call (red = AA, purple = AB, blue = BB). Genotypes are called for each sample (dot) by their signal intensity (norm R) and Allele Frequency (Norm Theta) relative to canonical cluster positions (dark shading) for a given SNP marker.

The GenomeStudio Genotyping (GT) Module supports the analysis of Infinium genotyping array data. This module enables efficient genotyping data normalization, genotype calling, clustering, data intensity analysis, loss of heterozygosity (LOH) calculation, and copy number variation (CNV) analysis. Fully integrated with the Infinium LIMS server, the GT Module allows you to access data and manage projects directly from within GenomeStudio Software.

As in all GenomeStudio modules, the GenomeStudio Framework displays data output in tabular form and enables you to visualize your results quickly and easily using the Illumina Genome Viewer and Illumina Chromosome Browser graphical tools.

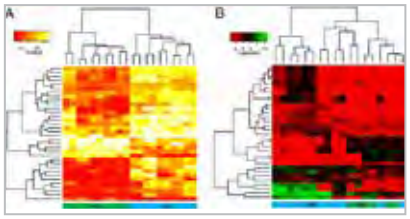
GT Module Highlights

- Analyze SNP and CNV data across up to millions of markers
- Estimate Log R ratio and B-allele frequency for copy number analysis
- Call genotypes, normalize and cluster data, and generate SNP statistics
- Export genotype data to various third party applications; access multiple CNV algorithms and copy number variation analysis tools
- Generate a chromosomal heat map for examining copy number aberrations across the entire genome for multiple samples
- Analyze data from two different product versions within the same project

Methylation Module

- Detect cytosine methylation at single-base resolution
- Identify methylation signatures across the entire genome

The GenomeStudio Methylation (M) Module supports the analysis of Infinium and GoldenGate methylation array data. This module calculates methylation levels (beta values) and analyzes differential methylation levels between experimental groups. It enables you to view CpG island methylation status across the genome with the Illumina Genome Browser and Illumina Chromosome Browser.



Single-site resolution data can be visualized as line plots, bar graphs, scatter plots, histograms, dendrograms, box plots, or heat maps. This module also enables you to combine methylation data with gene expression profiling experiments within the same GenomeStudio project for correlation between levels of methylated sites (beta values) and differential gene expression levels (p values).

Methylation Module Highlights

- Calculate methylation levels and visualize CpG island information
- Analyze differential methylation levels between two experimental groups
- Visualize results as line plots, bar graphs, scatter plots, histograms, dendrograms, box plots, heat maps, or control summary reports
- Merge gene expression profiling data with methylation data in the same project
- Display whole-genome data and beta values within the Illumina Genome Viewer
- Visualize beta values for one or more samples in the Illumina Chromosome Browser

Polyploid Genotyping Module

- Analyze polyploid organism genotyping data

The Polyploid Genotyping Module supports genotyping data analysis of polyploid organisms such as wheat and potato. It uses two density clustering algorithms to assign samples to meaningful clusters: PolyGentrain and Density Based Spatial Clustering of Applications with Noise (DBSCAN).

Polyploid Genotyping Module Highlights

- Call as many clusters as desired, enabling studies of hexaploid or octoploid species
- Automate clustering and genotyping calling for polyploid organisms

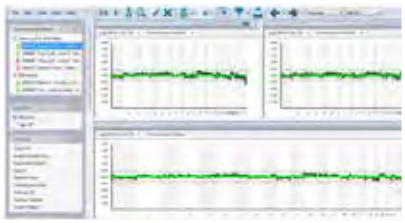
Beeline™ Software

Beeline Software offers a direct path to reduce experimental microarray data size and facilitate data analysis for large experiments. As the size of array data sets increases, the time required to calculate sample statistics and visually interrogate clusters increases. Beeline Software addresses this potential bottleneck by enabling pre-filtering of large data sets prior to import into GenomeStudio Software.

Beeline Software offers flexible filtering capabilities to reduce experimental array data size. The software pre-filters large data sets, reducing the overall data volume by eliminating any poorly performing loci (such as those with low signal intensity) and non-relevant variants across all samples.

Beeline Highlights

- Saves time on analysis
- Enables automatic calculation of sample statistics and allele calls.
- Reduces data size
- Offers flexible user-defined parameters to select/deselect samples and loci for targeted downstream analysis.
- Provides accessible reporting
- Generates text document output of numerous report formats.
- Provides seamless integration
- Works directly with Infinium data output and creates projects for direct import into GenomeStudio Software.



BlueFuse™ Multi Software

Fully automated software for analyzing and managing cytogenomic data. BlueFuse Multi Software combines fully automated data analysis with the ability to manage microarray and NGS data. It combines quality control measures, storage of information related to experiments, and sophisticated “Decision Track” visualization in an easy-to-use software package.

BlueFuse Multi Software is designed to manage, search, and display the wealth of data generated by whole-genome analysis experiments, using all the benefits of sophisticated processing algorithms. BlueFuse Multi Software can create, store, and manage databases of complex scientific information.

BlueFuse Multi Software is supplied at no additional cost with supported applications. These include VeriSeq™ PGS*, 24sure™, 24sure+, HumanKaryomap-12, and CytoChip arrays, as well as Infinium CytoSNP-12 and CytoSNP-850K BeadChips. Details of new features in each release are included in the release notes.

* VeriSeq PGS is a sequencing application

Instruments

iSeq 100 System
Affordable targeted sequencing

MiniSeq System
Simplicity and accessibility

MiSeqDx System
Focused diagnostic power
The MiSeqDx System is a registered *in vitro* diagnostic NGS instrument in the US, Europe, and other international markets.

For *In Vitro* Diagnostic Use
CE marked for IVD use

MiSeq System
Focused power

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

NextSeq 550 System
Flexible power

NextSeq 550Dx System
Next level in diagnostic power
For *In Vitro* Diagnostic Use
CE marked for IVD use

NovaSeq 6000 System
Introducing the next era in sequencing

HiSeq X System
Population power

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

Accessible sequencing solutions

Power tailored for every researcher, application, and scale of study

Product	iSeq 100 System	MiniSeq System	MiSeq System	MiSeqDx System
Description	Affordable and accessible benchtop sequencer	Simplicity and accessibility for targeted sequencing	Simplicity for targeted sequencing and small genomes, with the longest read length and largest range of throughputs[a]	Two run-mode platform: FDA-regulated and CE marked for *in vitro* diagnostic use and standard run mode with all MiSeq System capabilities
Key methods	Small-genome, amplicon, and targeted gene panel sequencing	Targeted DNA and targeted RNA sequencing	Small-genome, amplicon, and targeted gene panel sequencing	Cystic fibrosis assays
Run mode/kit type	Standard	Mid-output/ High-output	v3, v2, micro, and nano	
Flow cells processed/run	1	1	1	
Output/flow cell	1.2 Gb	1.9–7.5 Gb	0.3–15 Gb	
Run time	9–17 hrs	4–24 hrs	5–55 hrs	
Max clusters/flow cell[c]	4 M	25 M[d]	25 M[e]	
Max read length	2 × 150 bp	2 × 150 bp	2 × 300 bp	

a. Comparisons are within the Illumina portfolio
b. Specifications shown for an individual HiSeq X System; the HiSeq X System is available only as part of the HiSeq X Five or HiSeq X Ten System
c. For the MiniSeq System High-Output Kit only
d. Clusters passing filter
e. For MiSeq Reagent Kits v3 only

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	NextSeq 550 System		NextSeq 550Dx System	HiSeq X Five[b]/ HiSeq X Ten Systems[b]		NovaSeq 6000 System
	Power and simplicity for everyday genomics		The power and flexibility of the NextSeq 550 System with the reliability and consistency of an FDA-regulated clinical instrument	High throughput and low cost for production-scale WGS power		Massive throughput and flexibility for a broad range of high-intensity sequencing applications
	Everyday genome, exome, transcriptome, and targeted resequencing		Flexibility to perform research only testing, lab developed tests, and IVD assays	Population-scale WGS		Whole-genome, tumor normal, transcriptome, exome, and large panel targeted resequencing
	Mid-output	High-output	RUO and IVD modes	—	—	S1, S2, S4
	1	1	1	1 or 2		1 or 2
	20–39 Gb	30–120 Gb	20-120 Gb	900–1800 Gb		500Gb–6 Tb
	15–26 hrs	12–30 hrs	15-30 hrs	< 3 days		< 3 days
	130 M	400 M	2.5 B	3 B		10 B
	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp		2 x 150 bp

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iSeq 100 System

Affordable targeted sequencing

iSeq 100 System

With the lowest price, smallest footprint, and fastest run time of any Illumina instrument, the iSeq 100 System lets you expand the scope of your research, without the cost. Sequence microbes, targeted genes, RNA transcripts, and more at the push of a button.

- Affordable to acquire
- Small footprint
- Ideal for targeted gene expression (> 48 targets) studies

Sequencing reagent options	
Flow cell type	Standard
Bases (Gb)/flow cell	1.2
Clusters (M)/flow cell	4
Cycles	300
No. lanes/flow cell	1

www.illumina.com/iseq

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Methods for the iSeq 100 System

Method	Recommended read length	Optimized kit
Genome		
WGS (small genomes)	2 × 150 bp	Nextera Flex DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Cancer research sequencing panels	2 × 150 bp	TruSight Cancer Sequencing Panel
		TruSight Tumor Sequencing Panel 15
Genetic conditions sequencing panel	2 × 150 bp	TruSight Cardio Sequencing Panel
Custom sequencing	2 × 150 bp	AmpliSeq for Illumina Custom Panels
	2 × 150 bp	Nextera XT DNA Library Prep Kit
	2 × 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits

MiniSeq System

Simplicity and accessibility for targeted sequencing

MiniSeq System

Leveraging industry-leading sequencing technology in a simple, small, affordable system, the MiniSeq System supports a broad range of targeted DNA and RNA applications for examining single genes of entire pathways. An intuitive user interface, load-and-go operation, and onboard data analysis make it easy to learn and easy to use.

- Cost efficient to run and affordable to acquire
- Ideal for targeted sequencing

Sequencing reagent options				
Flow cell type	Mid-output		High-output	
Bases (Gb)/flow cell	2.4	1.9	3.8	7.5
Clusters (M)/flow cell	8	25	25	25
Cycles	300	75	150	300

www.illumina.com/miniseq

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Methods for the MiniSeq System

Method	Recommended read length	Optimized kit
Genome		
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Cancer research sequencing panels	2 × 150 bp	TruSight Cancer Sequencing Panel
		TruSight Tumor Sequencing Panel 26
		TruSight Tumor Sequencing Panel 15
		TruSight Myeloid Sequencing Panel
		TruSight RNA Pan-Cancer Sequencing Panel
		TruSight RNA Fusion Sequencing Panel
Genetic conditions sequencing panels	2 × 150 bp	TruSight One Sequencing Panel
		TruSight Cardio Sequencing Panel
Immunology sequencing panel	2 × 150 bp	TruSight HLAv2 Sequencing Panel
Metagenomics	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit
		Nextera DNA Library Prep Kit
		Nextera XT DNA Library Prep Kit
Custom sequencing	2 × 150 bp	AmpliSeq for Illumina Custom Panels
	2 × 150 bp	Nextera XT DNA Library Prep Kit
	2 × 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits
microRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Ribosome profiling	1 × 50 bp	TruSeq Ribo Profile Kit (Yeast)
		TruSeq Ribo Profile Kit (Mammalian)
Epigenome		
Protein–DNA interactions	2 × 75 bp	TruSeq ChIP Library Prep Kit

MiSeq Systems

Speed and simplicity for targeted and small-genome sequencing

MiSeq System

Combining speed, high-quality data, and the longest read lengths from Illumina, the MiSeq System is ideal for sequencing targeted panels, amplicons, and small genomes. A cost-effective alternative to capillary electrophoresis (CE) applications, it can perform rapid sequencing and variant detection for time-critical studies.

- Replaces CE in many applications
- Offers the longest read length of any Illumina benchtop system

www.illumina.com/miseq

Sequencing reagent options								
Flow cell type	Standard v2			Micro v2	Nano v2		Standard v3	
Bases (Gb)/flow cell	0.8	4.5	7.5	1.2	0.3	0.5	3.8	15
Clusters (M)/flow cell	15	15	15	4	1	1	25	25
Cycles	50	300	500	300	300	500	150	600

MiSeqDx System

Designed specifically for clinical laboratories, the MiSeqDx System is a registered *in vitro* diagnostic NGS instrument in the US, Europe, and other international markets.

- Performs screening and diagnostic testing
- Tailors data output to the needs of clinical labs
- Integrates software to enable sample tracking, user traceability, and results interpretation

Intended Use

The MiSeqDx instrument is intended for targeted sequencing of DNA libraries from human genomic DNA extracted from peripheral whole blood or formalin-fixed, paraffin-embedded (FFPE) tissue, when used for *in vitro* diagnostic (IVD) assays performed on the instrument. The MiSeqDx instrument is not intended for whole genome or *de novo* sequencing. The MiSeqDx instrument is to be used with registered and listed, cleared, or approved IVD reagents and analytical software.

For *In Vitro* Diagnostic Use
CE marked for IVD Use

www.illumina.com/miseqdx

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

Methods for the MiSeq System

Method	Recommended read length	Optimized kit
Genome		
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Cancer research sequencing panels	2 × 150 bp	TruSight Cancer Hotspot Panel
		TruSight Cancer Sequencing Panel
		TruSight Tumor Sequencing Panel 26
		TruSight Tumor Sequencing Panel 15
		TruSight Myeloid Sequencing Panel
		TruSight Pan RNA Sequencing Panel
		TruSight RNA Fusion Sequencing Panel
Genetic conditions sequencing panels	2 × 150 bp	TruSight One Sequencing Panel
		TruSight Cardio Sequencing Panel
Immunology sequencing panel	2 × 150 bp	TruSight HLA v2 Sequencing Panel
Metagenomics	2 × 250 bp	16S Metagenomic Sequencing Protocol
	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit
		Nextera DNA Library Prep Kit
		Nextera XT DNA Library Prep Kit
Custom sequencing	2 × 150 bp	AmpliSeq for Illumina Custom Panels
	2 × 150 bp	Nextera XT DNA Library Prep Kit
	2 × 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits *See page 75 for information on specific panels*
microRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Ribosome profiling	1 × 50 bp	TruSeq Ribo Profile Kit (Yeast)
		TruSeq Ribo Profile Kit (Mammalian)
Epigenome		
Protein–DNA interactions	2 × 75 bp	TruSeq ChIP Library Prep Kit

NextSeq 550 Systems

Speed and simplicity for everyday genomics

NextSeq 550 System

The NextSeq 550 System delivers the power of high-throughput sequencing with the simplicity of a benchtop sequencer, transforming exome, transcriptome, and whole-genome sequencing into everyday research tools. High-quality data combined with versatile, streamlined DNA-to-data workflows enables low- and high-throughput studies supporting a range of project sizes and applications.

- Configurable output and fast run time support a wide range of applications and sample sizes
- Broad menu of genomic applications runs on a single platform

www.illumina.com/systems/sequencing-platforms/nextseq.html

Sequencing reagent options					
Flow cell type	Mid-output v2		High-output v2		
Bases (Gb)/flow cell	20	39	30	60	120
Clusters (M)/flow cell	130	130	400	400	400
Cycles	150	300	75	150	300

NextSeq 550Dx System

The NextSeq 550Dx System is FDA-regulated and CE-marked for *in vitro* diagnostic (IVD) use, enabling clinical laboratories to develop and perform a wide range of applications, from NGS IVD assays using targeted panels, to clinical research applications that include methods from targeted panels to whole genomes.

Intended Use (United States)

The NextSeq 550Dx instrument is intended for targeted sequencing of DNA libraries from human genomic DNA extracted from peripheral whole blood or formalin-fixed, paraffin-embedded (FFPE) tissue, when used for *in vitro* diagnostic (IVD) assays performed on the instrument. The NextSeq 550Dx instrument is not intended for whole genome or de novo sequencing. The NextSeq 550Dx instrument is to be used with registered and listed, cleared or approved, IVD reagents and analytical software.

Intended Use (European Union/Other)

The NextSeq 550Dx instrument is intended for sequencing of DNA libraries when used with *in vitro* diagnostic (IVD) assays performed on the instrument. The NextSeq 550Dx instrument is to be used with specific registered, certified or approved IVD reagents and analytical software.

For *In Vitro* Diagnostic Use
CE marked for IVD Use

www.illumina.com/systems/sequencing-platforms/nextseq-dx.html

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Sequencing methods for the NextSeq 550 System

Method	Recommended read length	Optimized kit
Genome		
WGS (large genomes)	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
		Nextera DNA Library Prep Kit
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Cancer research sequencing panels	2 × 150 bp	TruSight Cancer Sequencing Panel
		TruSight Tumor Sequencing Panel 26
		TruSight Tumor Sequencing Panel 15
		TruSight Myeloid Sequencing Panel
		TruSight Pan RNA Sequencing Panel
		TruSight Tumor 170 Sequencing Panel
Genetic conditions sequencing panels	2 × 150 bp	TruSight One Sequencing Panel
		TruSight One Expanded Sequencing Panel
		TruSight Cardio Sequencing Panel
Immunology sequencing panel	2 × 150 bp	TruSight HLA v2 Sequencing Panel
Exome sequencing	2 × 75 bp	TruSeq DNA Exome
	2 × 75 bp	Nextera DNA Exome
Metagenomics	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit
		Nextera DNA Library Prep Kit
		Nextera XT DNA Library Prep Kit
Custom sequencing	2 × 150 bp	AmpliSeq for Illumina Custom Panel
	2 × 150 bp	Nextera XT DNA Library Prep Kit
	2 × 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Whole-transcriptome sequencing	2 × 75 bp	TruSeq Stranded Total RNA with Ribo-Zero Library Prep Kits
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits
miRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Ribosome profiling	1 × 50 bp	TruSeq Ribo Profile Kit (Yeast)
		TruSeq Ribo Profile Kit (Mammalian)
Epigenome		
DNA methylation	2 × 75 bp	TruSeq DNA Methylation Library Prep Kit
Protein/DNA interactions	2 × 75 bp	TruSeq ChIP Library Prep Kit
Targeted methylation	2 × 100 bp	TruSeq Methyl Capture EPIC Library Prep kit

Microarray methods for the NextSeq 550 System

Method	Supported array
Cytogenomics	Infinium CytoSNP-850K BeadChip Kit
	Infinium HumanCytoSNP-12 DNA Analysis BeadChip Kit
Karyomapping	Infinium HumanKaryomap-12 DNA Analysis Kit

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HiSeq X Series

Population power. WGS on an epic scale

HiSeq X Ten System

The HiSeq X Ten System is the first and only sequencing platform to break the $1000 barrier for a human genome at 30× coverage. Comprised of 10 or more individual instruments, the HiSeq X Ten System is ideal for population-scale projects focused on the discovery of genotypic variation. It can rapidly sequence thousands to tens of thousands of genomes at high coverage, delivering a comprehensive catalog of human variation within and outside of coding regions. With its ultra-high-throughput and unprecedented low price per genome, the HiSeq X Ten System makes population-scale human WGS a reality.

- Powerfully sequences thousands to tens of thousands of human whole genomes per year
- Achieves the $1000 human genome milestone, including depreciation, library prep, and labor*
- Enables ultra-high-throughput whole-genome bisulphite sequencing, *de novo* assembly, and whole-genome phasing of human and non-human species

*Depreciation schedules and labor might differ due to differences in institutional accounting and staffing.

> 90% of the world's sequencing data is generated using Illumina SBS technology.†

†Data calculations on file. Illumina, Inc., 2015

www.illumina.com/hiseqxten

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HiSeq X Five System

The HiSeq X Five System, a set of 5 or more individual HiSeq X Series instruments, delivers fast, affordable, production-scale WGS. With a lower initial capital investment than the HiSeq X Ten System, the HiSeq X Five System provides an accessible entry point to human WGS with an upgradeable path to population-scale sequencing and the $1000 genome. Now more researchers can complete large-scale human WGS projects rapidly, in their own labs.

- Powerfully sequences thousands of whole genomes per year
- Offers an affordable price per whole genome

www.illumina.com/systems/sequencing-platforms/hiseq-x.html

Instrument	HiSeq X Five System	HiSeq X Ten System
Bases (Gb)/flow cell	900	900
Clusters (M)/flow cell	3000	3000
Cycles	300	300

Methods for the HiSeq X Series

Method	Recommended read length	Optimized kit
Genome		
WGS (large genomes)	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit

NovaSeq 6000 System

The next era in sequencing starts now

NovaSeq 6000 System

The NovaSeq 6000 System unleashes groundbreaking innovations that build upon proven Illumina SBS technology. Get scalable throughput and flexibility for virtually any sequencing method, genome, and scale of project.

Sequencing reagent options							
Flow cell type		S1			S2		S4
Bases (Gb)/flow cell	160	320	480	333	667	1000	3000
Clusters (M)/flow cell	1600	1600	1600	3333	3333	3333	10,000
Cycles	100	200	300	100	200	300	300

www.illumina.com/systems/sequencing-platforms/novaseq.html

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Methods for the NovaSeq 6000 System

Method	Recommended read length	Optimized kit
Genome		
WGS (large genomes)	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq Nano DNA Library Prep Kit
		Nextera DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Exome sequencing	2 × 100 bp	Nextera DNA Exome
		TruSeq DNA Exome
Cancer research sequencing panels	2 × 150 bp	TruSight Tumor 170
		TruSight Cancer Sequencing Panel
	2 × 75 bp	TruSight RNA Fusion
		TruSight Pan RNA Sequencing Panel
Genetic conditions sequencing panels	2 × 150 bp	TruSight One Sequencing Panel
		TruSight One Expanded Sequencing Panel
		TruSight Cardio Sequencing Panel
Metagenomics	2 × 150 bp	TruSeq DNA PCR-Free Library Prep Kit
		TruSeq DNA Nano Library Prep Kit
		Nextera DNA Library Prep Kit
		Nextera XT DNA Library Prep Kit
Custom sequencing	2 × 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Whole-transcriptome sequencing	2 × 75 bp	TruSeq Stranded Total RNA with Ribo-Zero Library Prep Kits
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits
miRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Ribosome profiling	1 × 50 bp	TruSeq Ribo Profile (Mammalian)
		TruSeq Ribo Profile (Yeast)
Epigenome		
DNA methylation	2 × 75 bp	TruSeq DNA Methylation Library Prep Kit
Protein–DNA interactions	2 × 100 bp	TruSeq ChIP Library Prep Kit
Targeted methylation	2 × 100 bp	TruSeq Methyl Capture EPIC Library Prep Kit

Library prep automation

Partner developed and Illumina qualified

Library prep kits from Illumina can be automated on most general purpose liquid-handling robots. Illumina facilitates kit automation by partnering with leading automation vendors to develop methods for library prep. The resulting Illumina qualified methods significantly decrease the time and expense of method development and optimization, enabling rapid scaling for higher throughput.

	Agilent	Beckman Coulter	Eppendorf	Hamilton	Perkin Elmer	Tecan
16S rRNA				✓●		
Nextera					●	
Nextera Rapid Capture Custom Enrichment	●	✓	✓	●	✓	✓
Nextera XT DNA Library Prep Kit	●	✓	✓	✓	●	✓
AmpliSeq for Illumina Custom Panel						
TruSeq DNA PCR-Free	●	✓	✓	✓	✓	✓
TruSeq DNA Exome		●		●	✓	
TruSeq Nano DNA	●	✓	✓	✓	✓	✓
Nextera DNA Exome		●	✓	●		
TruSeq RNA Access	●	✓	✓	✓	✓	✓
TruSeq RNA v2		●			●	
TruSeq Stranded mRNA	●	✓	✓	✓	✓	✓
TruSeq Stranded Total RNA	●	✓	✓	✓	✓	✓
TruSight Cancer		✓	✓		✓	
TruSight HLA v2		✓	●			
TruSight Tumor 15		✓	✓		✓	
TruSight Tumor 170		✓		✓		

✓ Qualified by Illumina, which indicates our analysis has shown that libraries prepared with the method perform comparable to libraries prepared manually

● The method is available through an automation partner or other public source and is not qualified by Illumina

● Method is available through the automation partner, but not yet qualified by Illumina

Visit the automation partner's web page at www.illumina.com/HTautomation for the most up-to-date information on available methods. Contact the automation vendor directly for information on availability, installation, and support, including user guides and application notes.

www.illumina.com/automation

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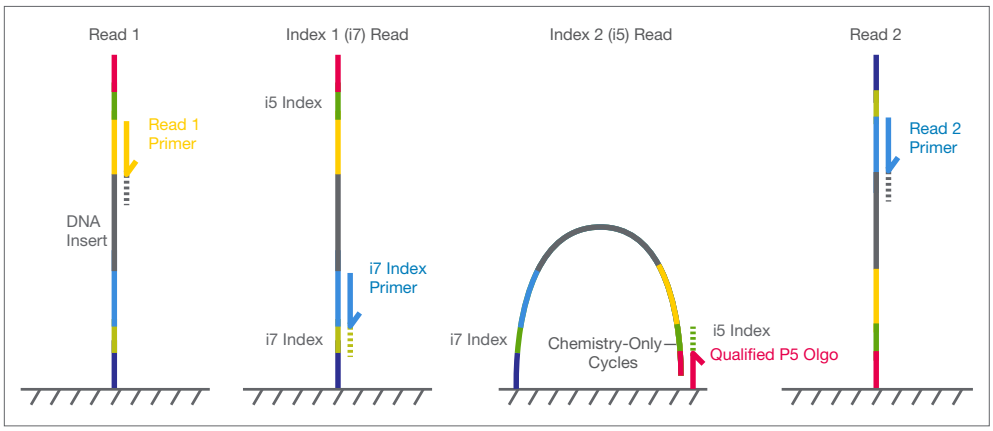
Indexing technology quick reference

Enabling higher throughput sequencing at a lower cost

Indexing, or Indexed sequencing, refers to the addition of short oligonucleotide sequences to libraries from individual samples. Indexed sequencing allows multiple libraries to be pooled and sequenced together, enabling higher throughput, and lowering overall costs per sample.

During index sequencing, the index is sequenced in a separate read, called the Index Read. When libraries are dual-indexed, the sequencing run includes two additional reads, called the Index 1 Read and Index 2 Read. Dual indexing allows for a high number of unique index combinations, and thus, indexed samples that can be loaded into a sequencing run.



Dual Indexed Sequencing on a Paired-End Flow Cell

Illumina offers numerous indexing solutions depending on the number of indexes desired, the type of sample, library prep kit, or instrument. Some kits include indexes. All indexes are compatible with Illumina sequencers. The table below will help determine when to choose indexes that are ordered separately from library prep kits.

	Recommended use	No. of indexes	Catalog no.
IDT for Illumina – TruSeq UD Indexes	WGS, WES, WTS (TruSeq Nano, PCR-Free, Stranded mRNA, Stranded Total RNA, IDT Exome, TruSeq Exome)	24 96	20020590 20022370
UMI Tool Kit	Liquid biopsy or similar samples when rare variants are important to identify	48 (samples)	20024586
TruSeq DNA and RNA Single Indexes (sets A and B)	For less sensitive applications and workflows requiring low level multiplexing	12 of 24 (RNA) 12 of 24 (RNA) 12 of 24 (DNA) 12 of 24 (DNA)	20020492 20020493 20015960 20015961
Nextera DNA CD Indexes	Applications using Nextera chemistry (not compatible with Nextera XT library prep)	24 96	20018707 20018708
AmpliSeq for Illumina CD Indexes	Use with AmpliSeq for Illumina Panels	96 96 (plate format)	20019105 20019108
TruSeq Targeted RNA Indexes	Using targeted RNA panels	96 of 384 96 of 384 96 of 384 96 of 384	15033977

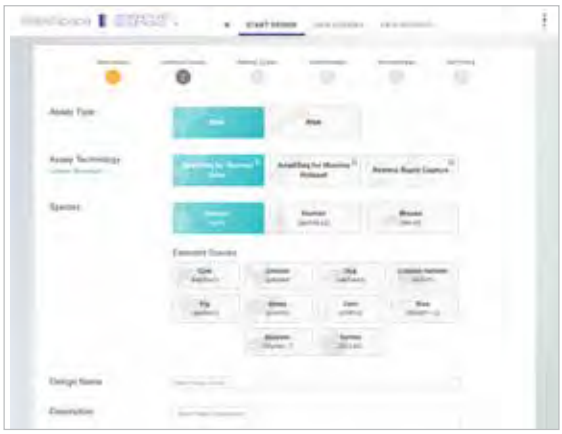
Analysis tools

Research informatics

Extend your research by integrating your data with the world's knowledge

Sequence assay design

Design sequencing experiments rapidly and efficiently



DesignStudio Software

DesignStudio Software, a personalized, easy-to-use, web-based sequencing assay design tool, provides dynamic feedback to optimize target region sequencing coverage, reducing the time required to design custom projects.

- Personalize and optimize content to fit your needs
- Design high-coverage (> 95%) custom targeted panels for your genes of interest
- Report and link to the University of California, Santa Cruz (UCSC) Genome Browser for confidence in your custom panel
- Use DesignStudio Software without bioinformatics expertise

Unified informatics

Simplify and expedite genomic workflows

BaseSpace Informatics Suite

BaseSpace Informatics Suite is a unified portfolio of informatics products:

- BaseSpace Sequence Hub
- BaseSpace Variant Interpreter
- BaseSpace Correlation Engine
- BaseSpace Cohort Analyzer

When used together on the BaseSpace platform, these tools deliver a seamless user experience that supports genomic research and precision medicine workflows. It provides an environment for integrating molecular and clinical data, and permits more people to participate in the discovery process.

BaseSpace Informatics Suite guides you from experiment design and sample tracking to data interpretation and meaningful reports. We make it easier to produce high-quality genomic information and apply the results for research, translational, and future precision medicine applications. Use these BaseSpace Informatics Suite products together, separately, or with the existing solutions in your lab.

Sample tracking and experiment design

BaseSpace Sequence Hub

The Prep feature in BaseSpace Sequence Hub allows users to plan entire sequencing workflows for the MiniSeq System and NextSeq 550 System. It allows users to easily design libraries, track library prep, and store logs of biological samples, libraries, and pools.

- Configure sequencing runs ahead of time, easily getting the run going, to save time
- Add libraries and pools to your planned runs with just a couple clicks
- Set up and track libraries, pools, and flow cells, simplifying complex library preparations
- Plan runs simply and intuitively; BaseSpace Sequence Hub integrates with Illumina instruments and allows easy, cloud-based configuration of MiniSeq System and NextSeq 550 System runs

Illumina Experiment Manager

Illumina Experiment Manager (IEM) is a wizard-driven application that allows researchers to design experiments before an Illumina sequencing run.

- Intuitive creation and setup of sample sheets
- Built-in validation checks that minimize or eliminate any errors in setting up sample sheets
- Flexible, easy, streamlined management of sample sheets and plate layouts
- Support for sample sheet setup for the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System

Data analysis and management

Spend more time doing research and less time setting up software

BaseSpace Sequence Hub

BaseSpace Sequence Hub allows users to store, analyze, and share NGS data. With BaseSpace Sequence Hub, you can securely upload sequencing data directly to the cloud from your instruments, analyze the data with a push of a button through BaseSpace Sequence Hub apps, and easily share data and results with colleagues. There's no need to build out IT and bioinformatics infrastructure, accelerating your research.

BaseSpace Sequence Hub is the only platform that enables direct streaming of sequencing data, monitoring of sequencing runs, and tracking of instrument performance. BaseSpace Sequence Hub apps, developed by Illumina or third parties, enable push-button NGS data analysis. These apps use common data analysis methods such as RNA-Seq, MethylSeq, exome, enrichment, amplicon, WGS, *de novo* assembly, metagenomics, and more.

BaseSpace Sequence Hub apps

- BaseSpace Sequence Hub apps provide the earliest access to cutting-edge algorithms from Illumina and third-party developers
- Share data and results instantaneously with collaborators across the world, with data storage in the Amazon Web Services (AWS) cloud
- Write your own pipeline as an app and make it publically available or run it privately on BaseSpace Sequence Hub

Biological data interpretation

More than data; we empower understanding

BaseSpace Variant Interpreter

BaseSpace Variant Interpreter leverages genomic interpretation content, enabling rapid and rich annotation, filtering, and accelerated interpretation of genomic data. Customize workflows and summarize findings into structured reports. Determine biological significance of genomic variants quickly and confidently within a software framework focused on data security and operational efficiency.

- Seamless integration with BaseSpace Sequence Hub and data transfer with an intuitive user interface enables a user-friendly and efficient variant-to-report workflow
- Comprehensive source of expert-curated content to expedite variant interpretation
- Comprehensive, rapid, and accurate genomic data annotation from a broad range of sources
- Customizable workflows for assays at all scales of throughput, from WGS to targeted sequencing, increase lab efficiency and scalability
- Security features such as access control, audit trail, and test configuration support security and lab compliance

BaseSpace Correlation Engine

Through the user-friendly, intuitive applications originally developed under the NextBio Research Platform, scientists can extract biological meaning from their experimental data at a speed and scale unmatched by other analysis tools. BaseSpace Correlation Engine powers discovery by enabling researchers to view all of their data across a broad spectrum of data types and platforms in the context of nearly 20,000 public studies. As software-as-a-service (SaaS) offering, BaseSpace Correlation Engine does not require servers or complicated software installation. Users can access all of these benefits from any modern web browser.

- Accelerated analysis of large data sets with statistical correlation and multivariate testing of data types across thousands of experiments and platforms
- Hypothesis-free data exploration with a wealth of anatomy, disease, pharmacology, genetic marker, and pathway information
- Hypothesis testing of genetic perturbations that affect a queried gene, sequence region, biogroup, or bioset
- Simplified access to additional tools using Application Programming Interface (API)
- Easy data sharing, annotation, and archiving

Translational and cancer informatics

Turn clinical research and molecular data into meaningful insights with powerful, turnkey informatics tools

BaseSpace Cohort Analyzer and BaseSpace Correlation Engine enable translational and cancer researchers to use complex genomic data in novel ways. These bioinformatics tools turn data into insight. Complex analysis can be executed with a few mouse clicks, empowering biologists and cancer researchers to make important discoveries without specialized bioinformatics skills.

BaseSpace Cohort Analyzer

BaseSpace Cohort Analyzer offers a platform for analyzing complex human subject data for translational research applications, hosting more than 70,000 subject records with over 1700 unique phenotypic attributes. This includes > 45,000 public subjects from high-profile studies like the Cancer Genome Atlas (TCGA). BaseSpace Cohort Analyzer maps and normalizes data, enabling researchers to compare thousands of subjects with a few clicks in real time.

By facilitating access to a large and growing repository of curated public data sets and powerful tools for large cohort analysis and group comparisons of both public and proprietary data, BaseSpace Cohort Analyzer enables transformation of complex data into meaningful insights.

BaseSpace Correlation Engine

Through user-friendly, intuitive applications found within BaseSpace Correlation Engine, oncology researchers can extract biological meaning from their experimental data, at a speed and scale unmatched by other analysis tools. BaseSpace Correlation Engine powers discovery by enabling researchers to view their data across a broad spectrum of data types and platforms in the context of nearly 22,000 public studies.

Pharma and biotech

A powerful analytical platform to inform drug discovery and development

BaseSpace Correlation Engine and BaseSpace Cohort Analyzer

BaseSpace Correlation Engine and BaseSpace Cohort Analyzer accelerate the core steps of drug development, providing tools for drug target discovery, lead identification, and analysis of preclinical and clinical trials. These analysis tools potentially increase clinical trial value by assessing the impact of therapies or drug effects in large patient populations.

BaseSpace Correlation Engine and BaseSpace Cohort Analyzer provide access to a growing library of curated genomic data. These easy-to-use web-based tools mine data and create billions of novel correlations. BaseSpace Correlation Engine and BaseSpace Cohort Analyzer contain a curated knowledge base of approximately 90,000 biomarkers with information about their biological impact. This knowledge base assists with rapid interpretation of data the significance of identified biomarkers.

Private data may be uploaded into BaseSpace Cohort Analyzer for comparison to any of the thousands of public subjects across many therapeutic areas, including cancers, autoimmune diseases, metabolic disorders, cardiovascular diseases, and more. BaseSpace Correlation Engine has more than 22,000 studies with daily updates and provides a large and growing repository of study signatures, providing data-driven answers for future discoveries.

Services

Instrument services, training, and consulting

We provide solutions so you can focus on the big discoveries

Illumina offers an integrated service solution that combines product care with opportunities for training and consulting support to help boost your lab efficiency and productivity, and optimize your workflow. With our flexible offering, you can select the services you need, so you can focus on what matters in your lab.

Illumina Product Support Services

In addition to the one-year basic service warranty included with your new purchase, Illumina offers instrument maintenance, repair, and qualification solutions. From basic to fully dedicated onsite options, we strive to anticipate your needs and exceed your expectations, including reagent replacement if one of our instruments fails during its service plan coverage.

Illumina University

Get high-quality results on Illumina technology even faster. Whether you want to maximize the effectiveness of your Illumina system, train new employees, or learn the latest techniques and best practices, we have a huge assortment of instructor-led, hands-on courses and web-based options for you at Illumina University.

Illumina Consulting

Illumina Consulting offers a suite of workflow, operation, and bioinformatics consulting services that expedite lab startup and maximize NGS and microarray workflow efficiencies so you can find the answers you seek. Harness our global network of expertise in genomics applications, IT/networking, data management, high-throughput operations, and bioinformatics.

Visit www.illumina.com/ProductServices

For your Illumina next-generation solutions from sample prep, library prep, arrays, and sequencing to informatics, we are here to provide you with the resources you need to accelerate progress.

Illumina product support services

Customized service to meet your needs

The Illumina Product Support Service team includes highly qualified field service engineers (FSE), technical applications scientists (TAS), field application scientists (FAS), system support engineers, bioinformaticians, and IT network experts. Deeply familiar with the intricacies of our systems, the team delivers accurate and expedient service and support.

You can upgrade to an Illumina Support Service Plan at any time during your warranty or extend coverage after your warranty. Many labs appreciate both the additional peace of mind and the fast, onsite service we offer with no hidden fees or extra costs.

Illumina Product Support Service Plan comparison

	BRONZE	SILVER	GOLD	DEDICATED ONSITE
	• Most affordable instrument performance maintenance option • Low sample volume • No mission-critical applications or critical turnaround needs	• Most popular full-service option • Designed maximize performance and productivity while helping ensure regulatory compliance with routine, documented Preventive Maintenance (PM) • Medium to high sample volume • Small to medium fleet size with redundant backup capacity required	• Ideal for labs in a regulated environment • Designed to maximize system performance and productivity for labs with rapid turnaround and stringent lab compliance needs • Significant savings on instrument qualification service vs à la carte purchasing	• Premium service plan ideal for large-production labs with > 10 high-throughput instruments per site • Designed to support the highest productivity and peak performance for sites with a large fleet of instruments • Full-time, onsite field service engineer for immediate response • Requires a parts-only plan[a]
Term (years)	1	1	1	2
Replacement parts	Yes	Yes	Yes	Parts-only contract required
Replacement reagents for instrument failure	No	Yes	Yes	Yes
Labor and travel	Yes	Yes	Yes	Yes
Targeted onsite response time (business days)	3	2[b]	Next business day[b]	Immediate
Preventative maintenance	No	1	1	2[c]
Qualification	No	No	Yes[d]	No

a. Excludes PM kits
b. Consult with your local service staff for availability
c. 2 PMs for HiSeq X and NovaSeq 6000 System services, and 1 PM for all other instruments
d. OQ at PM and qualified repair visits

Visit www.illumina.com/serviceplans for more information on Illumina Product Support Service Plans.

Illumina account managers and inside sales team are here to help you determine which plan is best suited for your lab.

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Comprehensive Qualification Services

Helping labs meet compliance requirements

At Illumina, we understand the changing regulatory landscape and strive to provide solutions to help our customers comply with the latest standards and regulations. To maintain compliance, it's important for laboratories to adopt well-documented qualification protocols for their Illumina instruments.

Services Offered

Qualification Service	Service Description	Qualification Recommended Intervals	Event-Specific Service
Installation Qualification (IQ)	Provides documented verification that the instrument is installed according to our specifications and safety regulations. During the IQ, a trained engineer confirms that the latest supported firmware and software versions were installed, verifies instrument setup and accessory logistics, checks that physical and environmental safety conditions are met, and provides a signed audit-ready report.	• After initial installation • After relocation and reinstallation	• Before first-time use • After general changes to lab environment (eg, remodeling, construction, electrical disruptions)
Operational Qualification (OQ)	Follows a comprehensive, well-defined protocol to make sure that the system is functioning according to our preset and validated operational specifications. The OQ protocol was developed and validated in Illumina labs and is updated after each instrument hardware and software release so you receive the most up-to-date service. Critical aspects of the OQ include motion, optics, fluidics, and thermal qualifications.	• After a reactive service, software upgrade, or preventive maintenance • Periodically, according to lab standard operating procedure	• With an IQ to test for baseline level of instrument performance • Before starting a major study or experiments
Performance Qualification (PQ)	Follows a comprehensive, well-defined protocol to make sure that the system is functioning according to preset and validated performance specifications. The PQ protocol was developed and validated in Illumina labs and is updated after each instrument hardware and software release so you receive the most up-to-date service. Critical aspects of the PQ include a PhiX data run (including projected yield total), data quality, and any additional comments.	After any qualified major repair	After maintenance, replacement, or upgrade of selected modules

Illumina University

Hands-on training at your chosen facility

Certified Illumina instructors deliver the following courses at a lab chosen by the requester. Each course can include up to four participants.

Training course ordering information for Illumina research products

Course name	Catalog no.
Genome	
TruSeq Cancer Hotspot Panel	TR-204-0031
AmpliSeq for Illumina DNA Library Prep Kit	20023392
Nextera DNA Flex Library Prep Kit	20022900
Nextera XT Library Prep Kit	TR-204-0009
TruSeq DNA PCR-Free Library Prep Kit	TR-204-0011
Nextera Rapid Capture Enrichment Kit	TR-204-0014
TruSight Tumor Library Prep with MiSeq Kit with MiSeq System	TR-204-0015
TruSight Rapid Capture Library Prep with MiSeq Kit with MiSeq System	TR-204-0016
Transcriptome	
TruSeq Targeted RNA Expression Library Prep with MiSeq Kit with MiSeq System	TR-204-0017
TruSeq Stranded Total RNA Library Prep Kit	TR-204-0012
TruSeq Stranded mRNA Library Prep Kit	TR-204-0013
AmpliSeq for Illumina RNA Library Prep Kit	20023393

Training course ordering information for Illumina diagnostic products

Course name	Catalog no.
MiSeqDx Cystic Fibrosis 139-Variant Assay[a]	TR-204-0018
MiSeqDx TruSeq Custom Amplicon Kit Dx[a]	TR-204-0019
NextSeq 550Dx TruSeq Custom Amplicon Kit Dx[a]	20023953

a. For *in vitro* diagnostic use

System training course information for Illumina products

System training courses	Catalog no.
NovaSeq Sequencing System Training - Customer Site	20016092
MiSeq System Training - Customer Site	20003928
NextSeq 500/550 Sequencing System Training - Customer Site	20003929
HiSeq 2500 Sequencing System Training - Customer Site	20004120
HiSeq 3000/4000 Sequencing System Training - Customer Site	20004121
MiniSeq System Training - Customer Site	20005637

Visit www.illumina.com/services/instrument-services-training/training.html and download the Customer Site Sample Preparation Training PDF for more information.

To ask questions or schedule a training course, call 800.809.4566 in North America or 1.858.202.4566 outside of North America.

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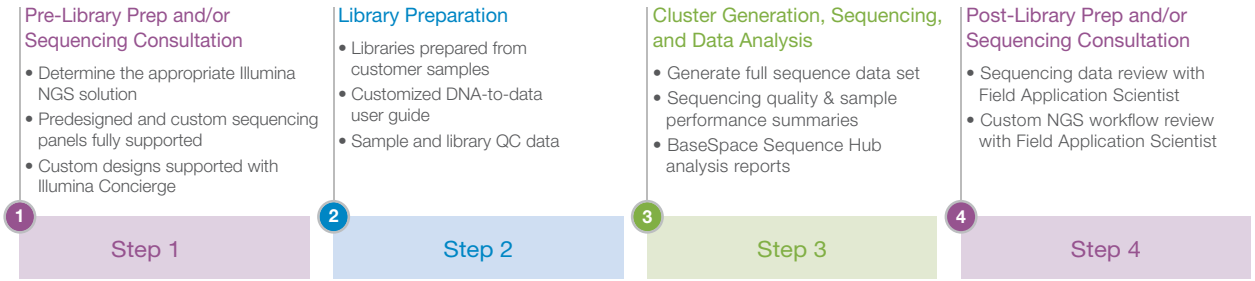
Illumina consulting

Try NGS or BeadChip technology with customer-provided samples using a fully customized, DNA-to-data Illumina service

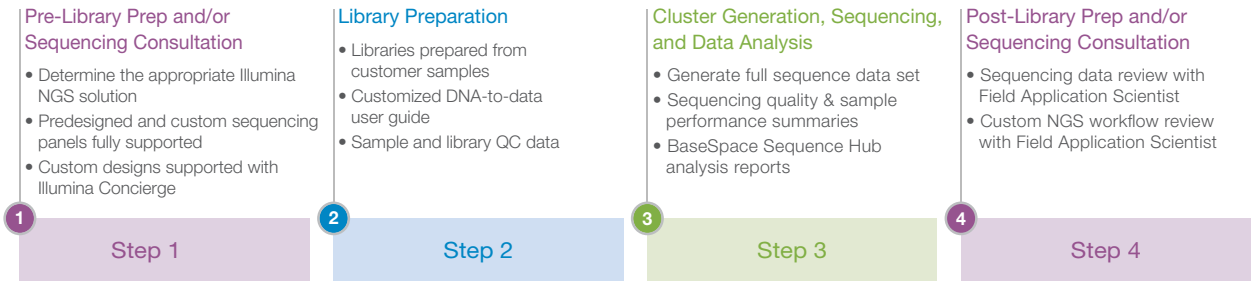
When considering the move to next-generation sequencing (NGS) or BeadChip (microarray) technology, scientists frequently navigate through many decisions regarding choice of instrumentation, applications, throughput requirements, and time from DNA to analysis.

Proof-of-Concept Service

Thinking of purchasing an Illumina NGS instrument, microarray, or library prep solution? Try our Proof-of-Concept (POC) Service with your own samples or libraries on a sequencing or library preparation instrument at the Illumina Application Lab.



NGS Proof-of-Concept Service Workflow—Illumina NGS Proof-of-Concept Service provides rapid delivery of data for any Illumina system or application.

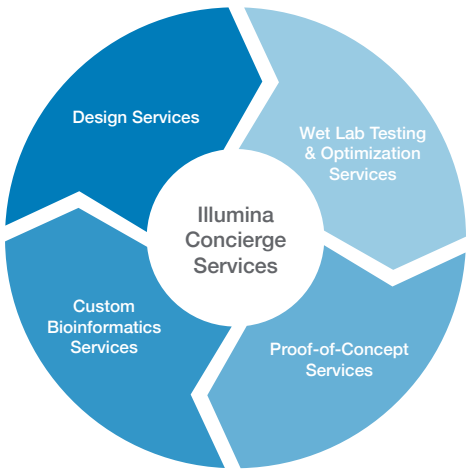


BeadChip Proof-of-Concept Service Workflow—The Illumina BeadChip Proof-of-Concept Service provides a streamlined workflow for rapid delivery of data.

Illumina Concierge Services

Experience comprehensive custom design and wet lab testing and optimization services by working with a dedicated scientist.

To address the challenges scientists can face with creating custom workflows, Illumina offers a wide range of Concierge Services from design assistance and wet lab testing and optimization to custom POC and bioinformatics.



Illumina Concierge Services offer dedicated assistance from experienced Illumina scientists.

Illumina Bioinformatics Professional Services

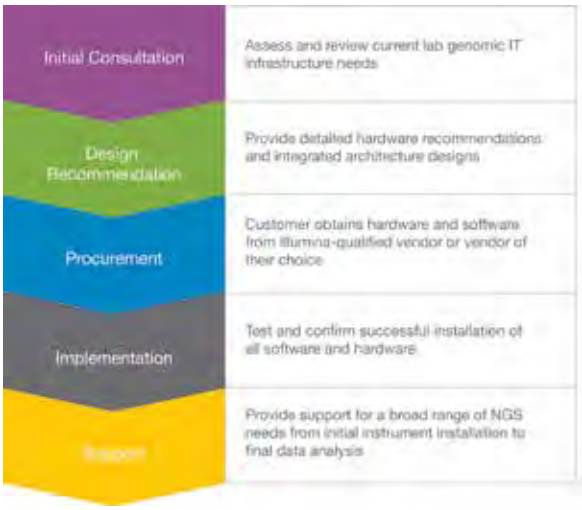
Whether you are a researcher new to bioinformatics or you need to improve your current analytical skills, Illumina can help. Illumina Bioinformatics Professional Services are delivered by experts with deep scientific and product knowledge, who are committed to helping researchers effectively pursue their scientific goals. Illumina experts can support a range of skill levels for both standard and specialized workflows.

Personalized consultations with Illumina Bioinformatics Professional Services

Illumina Bioinformatics Professional Services

Illumina Genomics IT Consulting Services

Illumina consultants leverage the power of their extensive knowledge and experience to deliver professional genomics IT services that can help you assess your facility's unique infrastructure requirements and provide answers scalable to meet tomorrow's data needs. Together with our qualified industry partners, we can help you accurately define and implement the hardware and software to bring NGS technology into your lab.



Illumina Genomics IT Consulting Services

Illumina Sequencing Consulting Service

Do you own, or are you contemplating purchasing multiple sequencing systems? The Illumina Sequencing Consulting Service provides you with customized, accurate, and expedient solutions for system implementation. We assist your facility in maximizing potential and achieving operational excellence at the low cost of ownership. Our comprehensive solutions include capacity planning, fleet management, risk mitigation, performance and QC trending, troubleshooting, bioinformatics, data management, and IT infrastructure.

Maximize efficiency		Achieve operational excellence
• Capacity planning • Risk reduction • Trending and troubleshooting • IT infrastructure	• Lab management • Staffing and communication • Inventory management • Data analysis and delivery	Large-scale sequencing

Illumina ArrayLab Consulting Service

Illumina provides customized support that can help your lab achieve its maximum potential for high-throughput array studies. With the Illumina ArrayLab Consulting Service, you can get customized consulting solutions for high-throughput genotyping production delivered by our specialized consulting professionals. This tailored support includes high-throughput laboratory setup; capacity planning; lab management, staffing, and resource planning; inventory management; data quality, risk management, and troubleshooting; and hardware and data management.

Contact your local account manager for additional information on Illumina consulting services.

Application index

Industry-leading solutions.

A community of support.

From library prep, arrays, and sequencing to informatics, Illumina genomic solutions empower researchers and clinical researchers across the globe to find the answers they seek.

When you join the Illumina community, you become part of a dynamic scientific movement that includes thousands of researchers and industry thought leaders. Throughout the year, we host user group meetings, symposia, consortia, online forums, and other initiatives—all designed to bring the best minds together to share ideas and advance science.

In addition to on-site training, ongoing support, and phone consults, we offer webinars and courses at various Illumina locations. We're here with all the resources you need to accelerate progress.

A global genomics leader, Illumina delivers next-generation sequencing workflow solutions to the basic and translational research communities. More than 90% of the world's sequencing data is generated using Illumina sequencing by synthesis technology.* Through collaborative innovation, Illumina is fueling groundbreaking advancements in the fields of oncology, reproductive health, genetic disease, microbiology, agriculture, and forensic science.

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* Data calculations on file. Illumina, Inc., 2018.

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