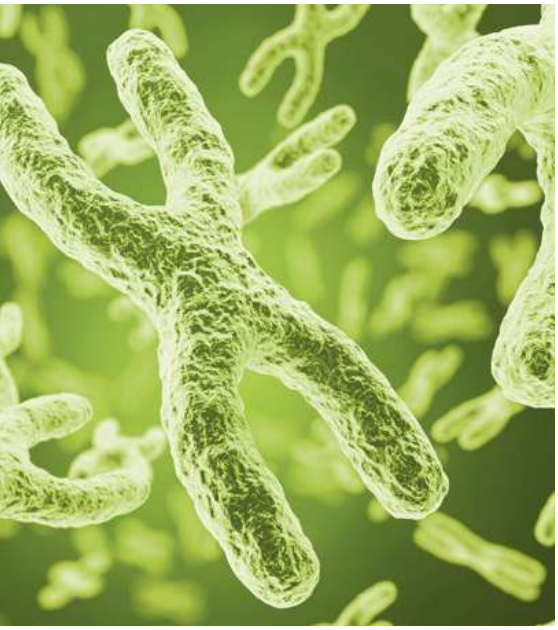
illumina®

Methods Guide





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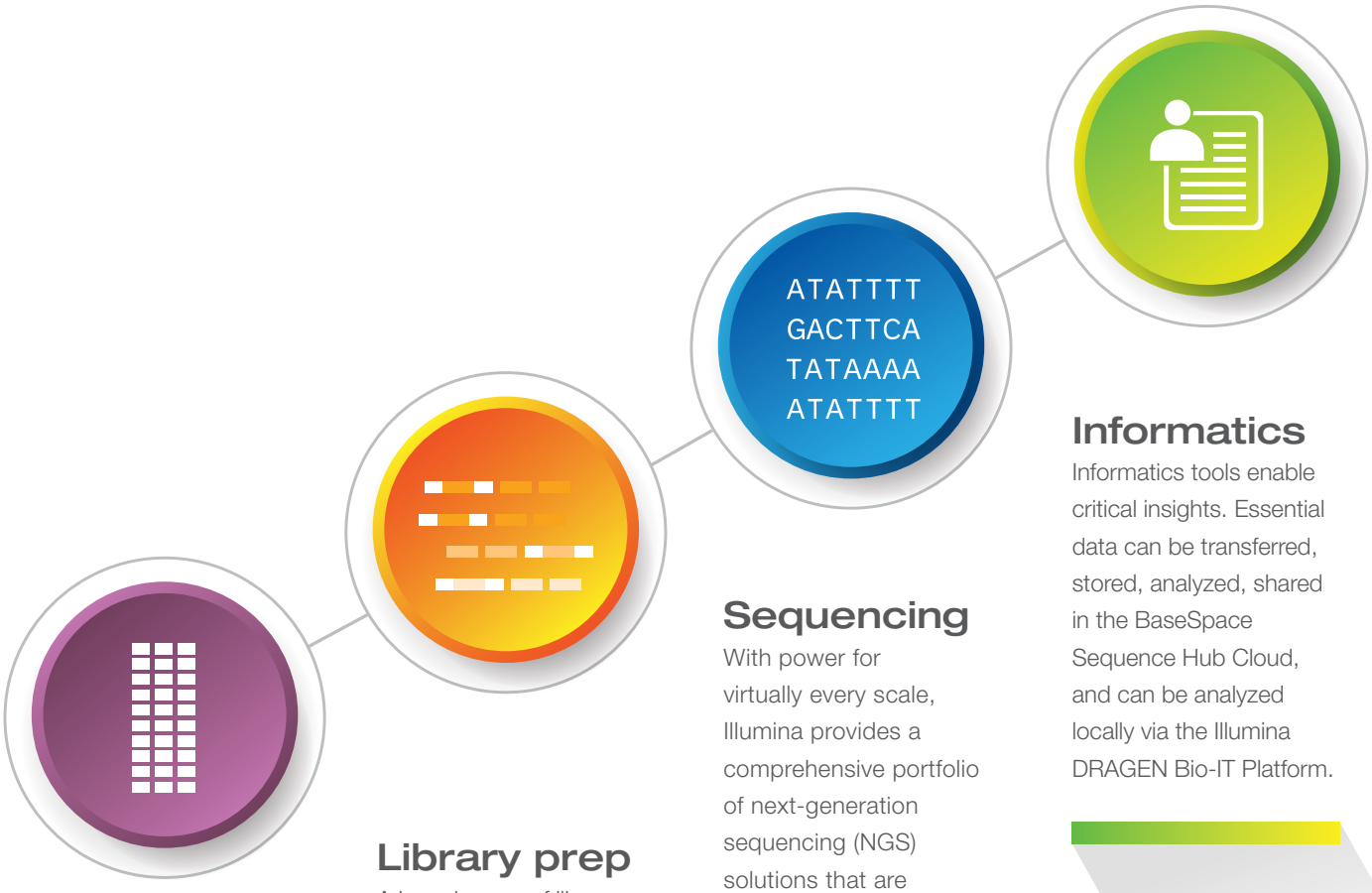

Informatics

Informatics tools enable critical insights. Essential data can be transferred, stored, analyzed, shared in the BaseSpace Sequence Hub Cloud, and can be analyzed locally via the Illumina DRAGEN Bio-IT Platform.

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

RNA

Epigenome

Infinium genotyping systems and BeadChips

INSTRUMENTS

ANALYSIS TOOLS

Research informatics

Translational informatics

SERVICES

Instrument services, training, and consulting

APPLICATION INDEX

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The Highlight 2019

New Technologies

Nextera Flex for Enrichment™

The Nextera Flex for Enrichment solution combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type, input amount, and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors (Table 1). Nextera Flex for Enrichment uses innovative bead-based chemistry with a simplified, single hybridization step (Figure 1). With the Nextera Flex for Enrichment workflow, DNA extraction can be processed directly from fresh blood and saliva samples with the Flex Lysis Reagent Kit and Saliva Lysis Protocol, respectively, for additional time savings.

TruSeq Neurodegeneration™

The TruSeq Neurodegeneration Panel is a targeted sequencing panel that leverages expertise from leading researchers in the neurogenomics community. It was designed with input from scientists studying neurodegenerative disease Informatics.

DRAGEN Bio-IT™

The Illumina DRAGEN (Dynamic Read Analysis for GENomics) Bio-IT Platform provides ultra-rapid secondary analysis of next-generation sequencing (NGS) data.

TruSight Oncology 500™

Assay targeting multiple variant types, including tumor mutational burden (TMB) and microsatellite instability (MSI), even from low-quality samples.

Partnerships

AmpliSeq for Illumina

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 Myeloid™
- AmpliSeq for Illumina Focus™
- AmpliSeq for Illumina Childhood Cance™
- AmpliSeq for Illumina Immune Repertoire Plus, TCR beta™
- AmpliSeq for Illumina TCR beta-SR™

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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

Myeloid
Targeted panel to investigate 40 DNA genes, 29 RNA fusion driver genes, and 5 gene expression levels associated with myeloid cancers

Focus
Targeted DNA and RNA research panel investigating 52 genes with known relevance to solid tumors

Childhood Cancer Research
Targeted panel for investigating 203 genes associated with cancer in children and young adults

Immune Repertoire Plus, TCR beta
Targeted RNA research panel to investigate T-cell diversity and clonal expansion by sequencing T-cell receptor beta chain rearrangements

AmpliSeq for Illumina TCR beta-SR
FFPE-compatible panel for measuring T-cell diversity and clonal expansion in tumor samples by sequencing T-cell receptor beta chain rearrangements

See page 26 for details.

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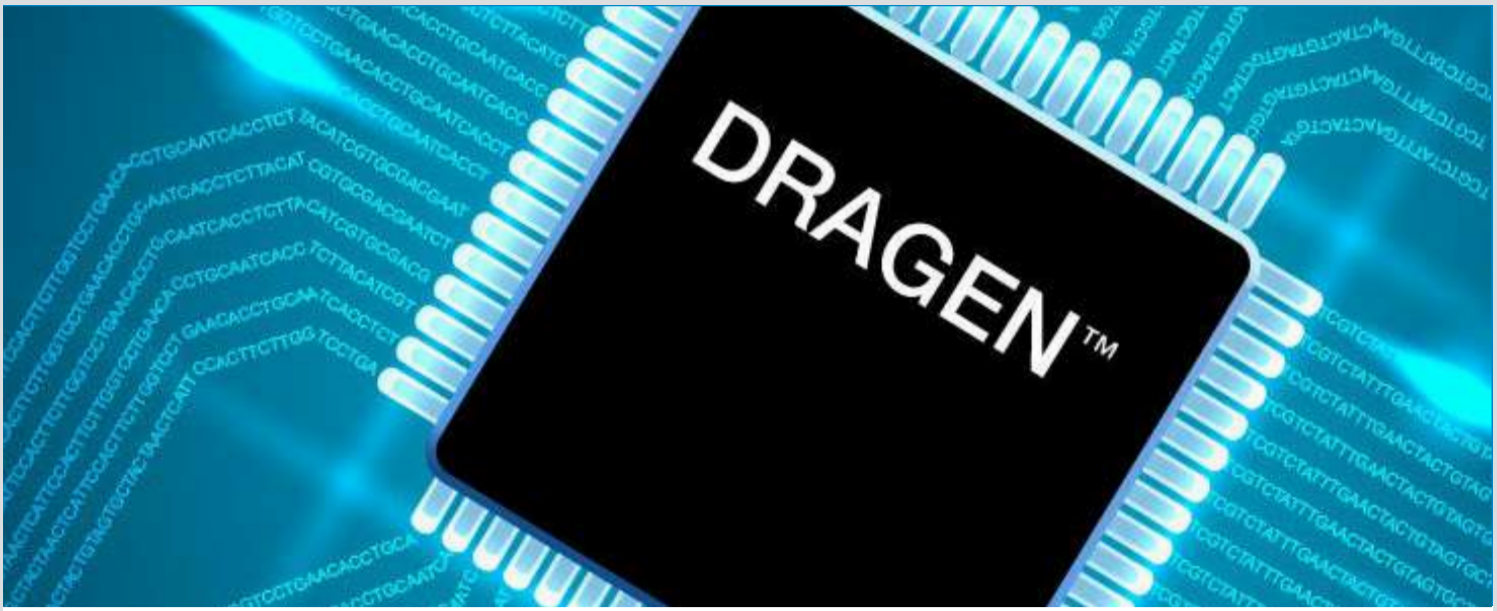

Accurate, Ultra-Rapid Secondary Analysis

Accurate Data
Detects small variants with high analytical sensitivity and specificity

Ultra-Rapid Speeds
Process an entire human genome at 30´ coverage in about 25 minutes on premise

Cost-effective and easy to implement solution
Reduces hardware investments and cloud-based costs, with push-button or command-line options

Robust Applications
Supports a variety of applications and methods both on premise and in BaseSpace™ Sequence Hub

DRAGEN Ultra-Rapid Analysis Pipelines

The robust suite of DRAGEN pipelines are available both on-premise and in the cloud via BaseSpace Sequence Hub (BSSH).

Name	Description	Availability
Illumina DRAGEN Germline Pipeline	The DRAGEN Germline Pipeline provides end-to-end (BCL → VCF) NGS analysis, including advanced error model calibration for increased accuracy, and repeat expansion detection and genotyping through Illumina Expansion Hunter.	On-Premise On BSSH
Illumina DRAGEN Somatic Pipeline	The DRAGEN Somatic Pipeline includes tumor-only and tumor–normal modes, designed for detecting somatic variants in tumor samples	On-Premise On BSSH
DRAGEN Reference Builder	This app accepts FASTA files, and builds the proprietary reference used by the DRAGEN apps.	On-Premise On BSSH
Illumina DRAGEN RNA Pipeline	The DRAGEN RNA Pipeline performs transcriptome analysis starting with splice junction discovery and alignment, followed by rapid alignment and splice junction mapping, quantification, and fusion detection.	On-Premise On BSSH
Illumina DRAGEN Joint Genotyping/Population Pipeline	The DRAGEN Joint Genotyping/Population Pipeline calls variants jointly across multiple genomes and scales to large cohorts of samples at expedited speeds with uncompromising accuracy.	On-Premise On BSSH
Illumina DRAGEN CNV Pipeline	The DRAGEN CNV Pipeline performs copy number variant (CNV) analysis for germline and somatic exomes and genomes. Various levels of filtering can be applied to mitigate false positives before emitting the final calls.	On-Premise On BSSH
Illumina DRAGEN Methylation Pipeline	The DRAGEN Methylation Pipeline offers multiple operating modes, including reference-only alignment and annotation-assisted.	On-Premise On BSSH
Illumina DRAGEN Map + Align Pipeline	The DRAGEN Map + Align pipeline is capable of ultra-rapid mapping and aligning DNA and RNA for both exomes and genomes.	On-Premise On BSSH

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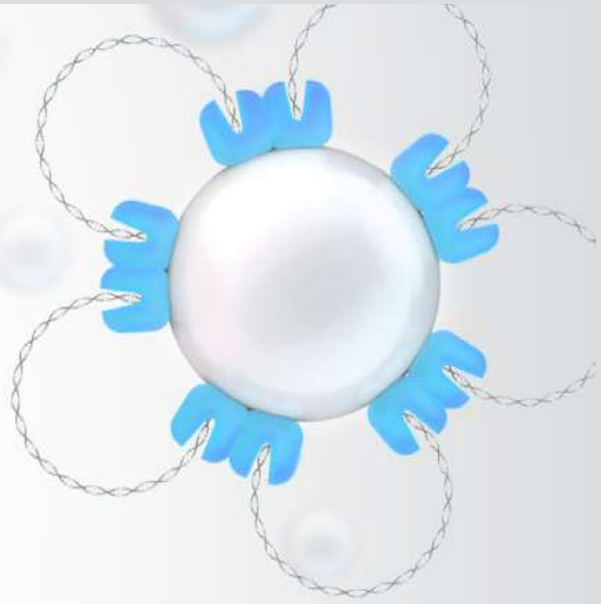
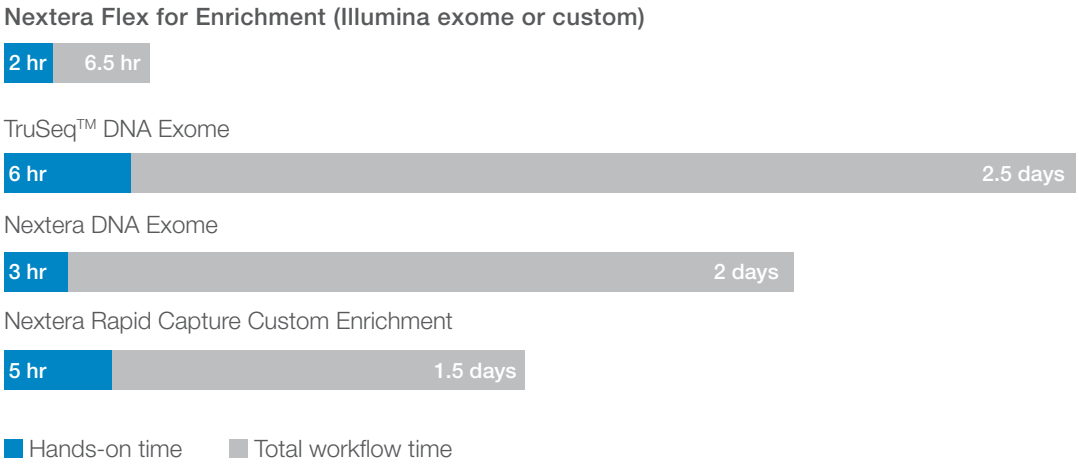
Introducing Nextera Flex for Enrichment

A fast, flexible targeted sequencing solution for DNA

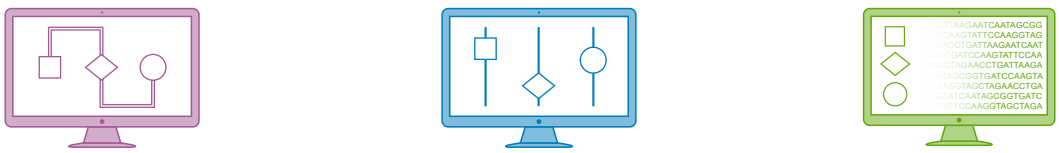


Nextera Flex for Enrichment Workflow



Nextera Flex for Enrichment delivers the fastest Illumina enrichment workflow – Workflow times are based on processing 12 samples at 12-plex enrichment. Times may vary depending on equipment used, number of samples processed, automation procedures, or user experience.

Broad range of applications with Nextera Flex for Enrichment



Fixed panels

- TruSight™ Cancer
- TruSight One
- TruSight One Expanded
- TruSight Cardio
- Illumina Exome

Custom

- Transition existing Nextera Rapid Capture Custom Enrichment panels to a faster workflow
- Develop new custom panels in DesignStudio using Nextera Flex for Enrichment and the Illumina Custom Enrichment Panel probes
- Use Illumina or third-party panels between 500 and 675,000 single- or double-stranded biotinylated probes
- Use DesignStudio to order custom panels between 2000 and 675,000 probes

Whole-exome

- Compatible with 80-mer or 120-mer oligos
- Data sets are available for the Illumina Exome Panel and third-party exome panels in BaseSpace Sequence Hub

Broad range of applications with Nextera Flex for Enrichment – Nextera Flex for Enrichment can be used to support a broad range of application including fixed panels, custom panels, and whole-exome sequencing from Illumina or 3rd party vendors.

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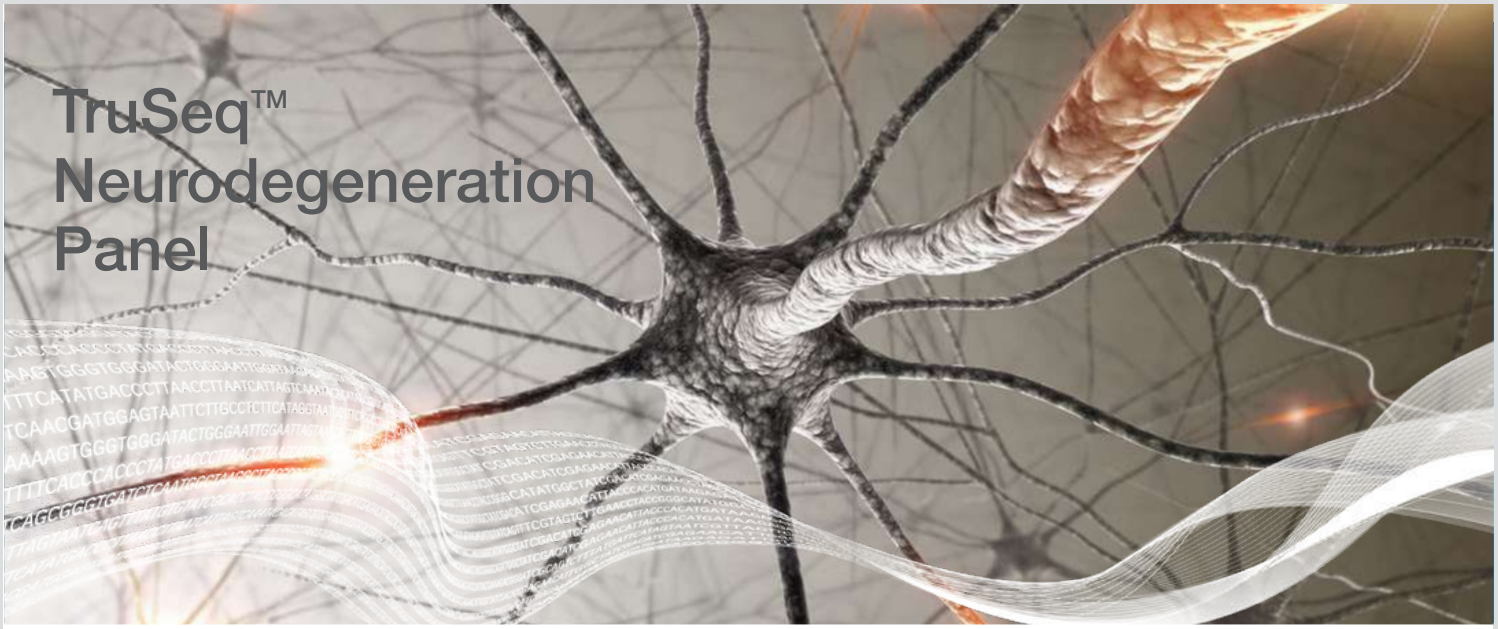
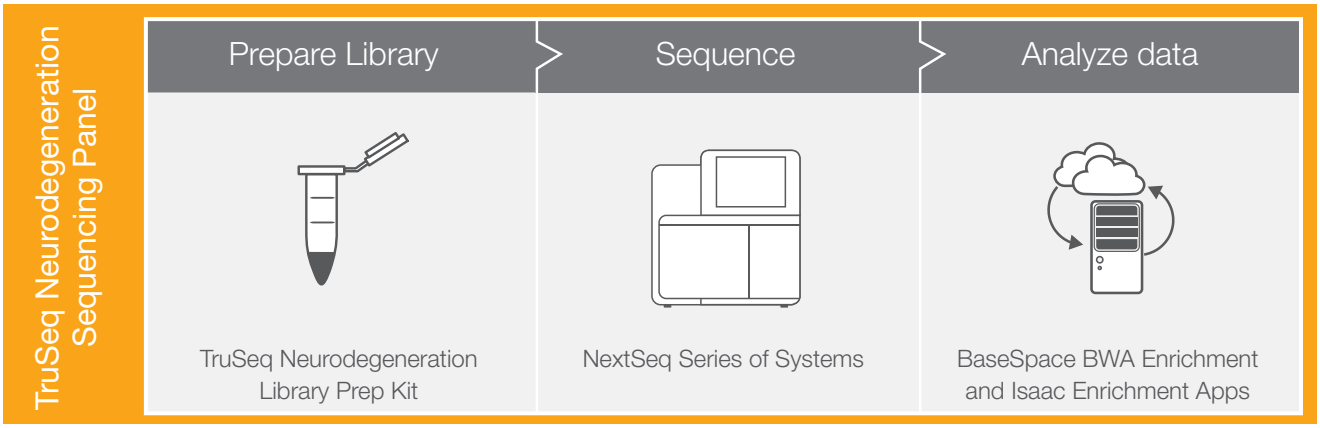


TruSeq™ Neurodegeneration Panel

TruSeq Neurodegeneration Workflow



Overview of the TruSeq Neurodegeneration Sequencing Panel workflow
The TruSeq Neurodegeneration Sequencing Panel is part of an integrated, streamlined workflow that includes library preparation, sequencing, and data analysis.

Product Highlights

Customer validated content
- 118 full genes: exons, introns, UTRs, and promoter regions

Comprehensive panel including:
- Alzheimer's disease
- Parkinson's disease
- Amyotrophic lateral sclerosis (ALS)
- Frontotemporal dementia
- Dementia with Lewy Body
- Dystonia
- Early onset dementia

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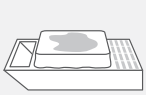
TruSight Oncology 500

TruSight Oncology 500 Workflow

FFPE specimens	Sample	Library prep and enrichment	Sequence	Analysis
				
Supports multiple tissue types	Commercial DNA/RNA extraction kits	TruSight Oncology UMI kit DNA Probes (523 genes) RNA Probes (55 genes) Automatable workflow	NextSeq™ System	TruSight Oncology 500 Local App

TruSight Oncology 500 workflow

TruSight Oncology 500 integrates into current lab workflows, going from nucleic acids to a variant calls in 3–4 days.

Product Highlights

- Save Time, Money, and Samples through Test Consolidation
- Pan-cancer content aligned with key guidelines
- DNA + RNA* assay targeting 523 genes for assessment of small variants, TMB, MSI, splice variants, and fusions
- Identify relevant alterations while achieving highly accurate results
- Achieve Highly Confident Results
- Software filters artifacts for accurate variant calling
- Enrichment chemistry inclusive of unique molecular indexes (UMIs) for high sensitivity in variant detection
- Based on proven Illumina platform
- Unlock Immuno-Oncology
- Panel contains immuno-oncology biomarkers TMB and MSI
- Tumor-only workflow for simplicity and efficiency
- TMB calling performance similar to whole-exome sequencing (WES) panels
- Large 1.94 Mb panel and sophisticated algorithm for accurate TMB score

* The product to evaluate DNA & RNA variants is the TSO500 DNA/RNA bundle.

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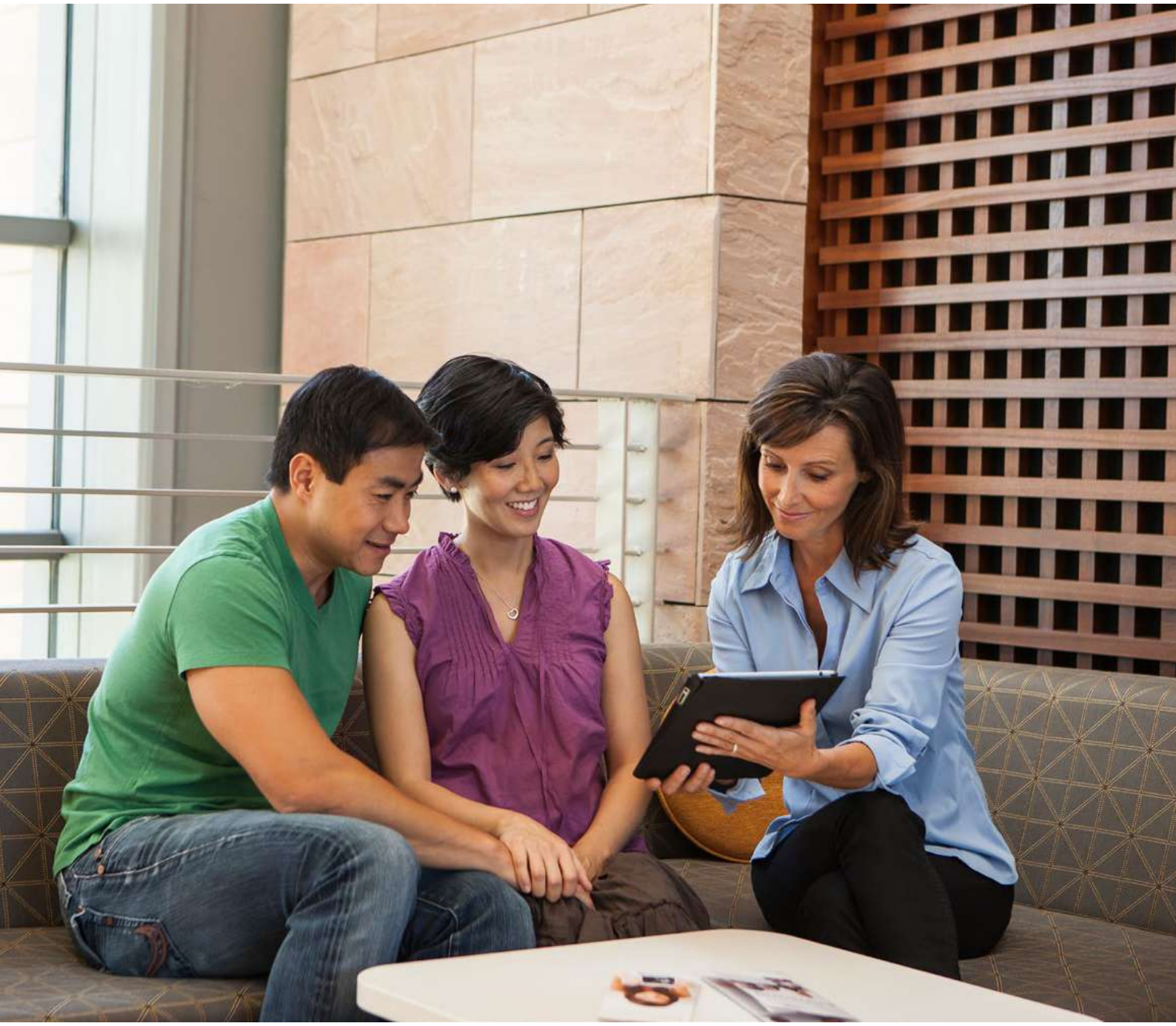
Methods

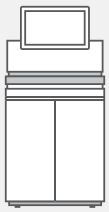


Whole-genome sequencing (large genomes)

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** **NovaSeq 6000 System**	**BaseSpace Sequence Hub** Resequencing Oncology paired analysis Whole-genome phasing Illumina DRAGEN Germline Pipeline
	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 NovaSeq 6000 System	**Workflow Design and Evaluation 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 https://www.illumina.com/techniques/sequencing/dna-sequencing/whole-genome-sequencing/large-genomes.html for more details.

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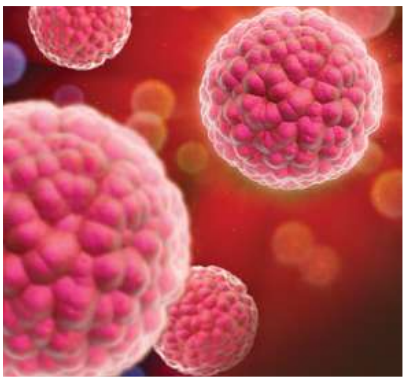
Innovation. Discovery. Application.



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)
Genome-Wide Association Studies (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	12.5 hrs	14.5 hrs	3.5 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 sequencing system


Product	NextSeq 550 System		NovaSeq 6000 System			
Most important to me	Convenience, ease of use, rapid turnaround time, and instrument affordability for large-genome sequencing applications		Scalable throughput and flexibility for virtually any genome, sequencing method, and scale of project			
Run mode/kit type	Mid-output	High-output	SP Xp	S1 Xp	S2 Xp	S4 Xp
Samples/flow cell[a]	1	1	2	4	10	24

a. Assuming a 30× human genome

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STEP 3
Obtain results

Cancer research paired analysis ▼

Resequencing ▼

1 Align/Call variants

DRAGEN Germline Pipeline
- Consumes raw or aligned reads and performs mapping and alignment, position sorting, and variant calling

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

Align/Call variants

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, integrative genomic data sets

3 Interpret/Report

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

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 and powerful tools for cohort analysis and group comparisons of public and proprietary data

B BaseSpace Sequence Hub

Visit https://www.illumina.com/techniques/sequencing/dna-sequencing/whole-genome-sequencing/large-genomes.html for more details.

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Library prep ordering information

Nextera Flex for Enrichment Kit

The Nextera Flex for Enrichment Kit combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type and amount and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors.

The Nextera Flex for Enrichment 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			
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents		96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents		16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents		96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents		16	20025519
Panel		**No. of Enrichment Reactions**	
Illumina Exome Panel–Enrichment Oligos only		8 or 12	20020183
TruSight Cancer – Enrichment Oligos only		4 or 8	FC-121-0202
TruSight One – Enrichment Oligos only		6	20029227
TruSight One Expanded – Enrichment Oligos		6	20029226
TruSight Cardio – Enrichment Oligos only		8	20029229
TruSeq Neurodegeneration–Enrichment Oligos only		8	20029550
Index Adapter Component			
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	96	20027215
IDT for Illumina Nextera DNA UD Indexes Set D	96	96	20027216

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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 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) and multiple DNA input types, and is 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.

The Nextera DNA Flex 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.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Flex Lysis Reagent Kit (96 reactions)			20018706
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

Workflow Design and Evaluation Service

Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can bring new capabilities to your laboratory. Receive in-person consultation to establish needs and goals, sequencing of your samples at the Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	20016091

Whole-genome sequencing (small genomes)

Sequence virtually any small genome and make new discoveries with scalable throughput

Key strengths

- Sequence multiple 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 Flex for Enrichment Library Prep Kit **Nextera DNA Flex Library Prep Kit**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Local Run Manager Software** Resequencing **BaseSpace Sequence Hub** DRAGEN for Resequencing *De novo* assembly
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

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

Visit https://www.illumina.com/techniques/sequencing/dna-sequencing/whole-genome-sequencing/small-genomes.html for more details.

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Innovation. Discovery. Application.

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, fungal, and viral diversity and detect the abundance of microbes in various environments, including unculturable microorganisms. In addition to gene identification, shotgun metagenomics provides gene function information which is key to understanding complex microbial communities.

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. Shotgun metagenomics also enables detection of anti-microbial resistance genes and virulence genes that impact human health.

STEP 1
Choose a library prep method

Product	Nextera DNA Flex Library Prep Kit	Nextera XT DNA Library Prep Kit
Most important to me	Simple and versatile sample-to-data workflow	Less hands on time and longer insert protocol
Least important to me	PCR-free workflow	Amplicons < 300 bp in length
Input quantity	1–500 ng DNA	1 ng DNA
Multiplexing	96 samples	384 samples
Assay time	3–4 hrs	90 min
Hands-on time	1.5 hrs[a]	15 min
Target insert size	~350 bp	300 bp–1.5 kb
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 sequencing system

Product	iSeq 100 System	MiniSeq System	MiSeq System	NextSeq 550 System	NovaSeq 6000 System (SP and S1 flow cells)
Most important to me	Affordability and efficiency	Simplicity and instrument affordability	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**	1–16	1–96	1–96	1–384	96–768[b]
Run mode/kit type	Standard	Mid-output/ High-output	Research mode and Dx	Mid-output/ High-output	SP and S1
Flow cells processed/ run	1	1	1	1	1 or 2

a. Comparison among Illumina sequencing portfolio

b. The *E. coli* genome samples processed per flow cell number is limited by the number of indexes

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STEP 3
Obtain results

Resequencing ▼

1

Align/Call variants

BWA Aligner [B] and Pisces Variant Caller [B]
- Allows for read alignment and mutation detection for any genome with a known reference sequence
- Analyzes bacteria, fungi, or viruses after *de novo* assembly, if a reference sequence is unavailable

SRST2
- Reports the presence of sequence types from the MLST database and/or reference genes from a database of sequences for virulence genes, resistance genes, and plasmid replicons

DRAGEN Germline Pipeline
- Aligns and optionally variant calls FASTQ files, outputting a BAM, VCF, or both

***De novo* assembly ▼**

Assemble

Assembly workflow [L] [B]
- 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

Bionumerics (third-party commercial)
- Combines information from various genomic and phenotypic sources into one global database and conducts combined analyses

Chun Labs BacID (third-party commercial)
- Identifies > 10,000 species for microbial applications in research, applied, and industrial settings

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

2

Analyze

One Codex
- Provides a fast and easy-to-use analysis platform for viral metagenomics from the curated One Codex genome database

DeepCheck (HIV-1, HCV, HBV)
- Performs deep sequencing analyses and reports on subtyping, genotyping, and inferred levels of resistance

Annotate/Filter

Prokka Genome Annotation [B]
- Annotates genes and identifies coding sequences in prokaryotic genomes
- Does not annotate eukaryotic genomes rapidly

Bacterial Analysis Pipeline
- Identifies species, multilocus sequence type, plasmids, virulence, and antimicrobial resistance genes in bacteria

3

Visualize

The Integrative Genomics Viewer (IGV) [B]
- Displays alignments and variants from multiple samples for performing complex variant analysis

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

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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
Nextera XT Index Kit	96	384	FC-131-1002
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
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) and multiple DNA input types, and is 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.

The Nextera DNA Flex 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.
Nextera DNA Flex Library Prep Kit		24	20018704
Nextera DNA Flex Library Prep Kit		96	20018705
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
Flex Lysis Reagent Kit		96	20018706
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

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Workflow Design and Evaluation Service

The Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can bring new capabilities to your laboratory. Receive in-person consultation to establish needs and goals, sequencing of your samples at the Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	20023613
MiniSeq Workflow Design and Evaluation Service	20003924
MiSeq Workflow Design and Evaluation Service	SP-801-1002
NextSeq Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	20016091

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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

TruSight Sequencing
Cancer Panel
Tumor 15 Panel
Myeloid Panel
RNA Pan-Cancer Panel
Tumor 170 Panel[a]
TruSight RNA Fusion
AmpliSeq for Illumina
BRCA Panel
Cancer Hotspot Panel v2
Comprehensive Cancer Panel
Comprehensive Panel v3
Focus Panel
Immune Response Panel
Myeloid Panel
Childhood Cancer Panel
TCR beta-SR Panel

Sequence

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

Analyze and Report

Available in Local Run Manager (MiniSeq, MiSeq, NextSeq)

Available in BaseSpace Sequence Hub

Illumina DRAGEN Somatic Pipeline

Illumina DRAGEN RNA Gene Fusion Detection Pipeline

Illumina Informatics Solutions

BaseSpace Sequence Hub
Run management | Analysis | Reporting

Training

Customer site training
TruSight Tumor 15 Panel
TruSight Tumor 170 Panel
TruSight Cancer Panel
TruSight Myeloid Panel
AmpliSeq Library Prep

Service contracts

Tiered service plans
iSeq 100 System
MiniSeq System
MiSeq System
NextSeq 550 System
NovaSeq 6000 System

Professional services

Workflow Design and Evaluation Service
Standard application functional testing with your samples
Cancer Research Bioinformatics Service

Validation Services
IQ/OQ/IPV[b]

a. Illumina verifies that assays can run on Illumina instruments, which may be a subset of the Illumina sequencing systems. TruSight Tumor 170 has not been verified on the NovaSeq 6000 System.

b. 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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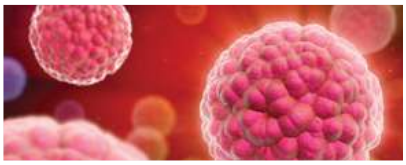
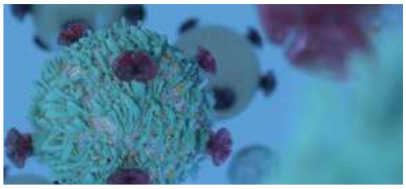
Innovation. Discovery. Application.



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.

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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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AMPLISEQ FOR ILLUMINA PANELS—AMPLICON-BASED LIBRARY PREP KITS

Product	AmpliSeq for Illumina Myeloid Panel	AmpliSeq for Illumina Childhood Cancer Panel	AmpliSeq for Illumina TCR beta-SR Panel	AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel	AmpliSeq for Illumina Transcriptome Human Gene Expression Panel
Key use	Analysis of DNA genes, RNA fusion driver genes, and gene expression levels associated with Myeloid cancers	Targeted panel for investigating 203 genes associated with cancer in children and young adults.	FFPE-compatible panel for measuring T-cell diversity and clonal expansion in tumor samples by sequencing T-cell receptor beta chain rearrangements.	Targeted RNA research panel to investigate T-cell diversity and clonal expansion by sequencing T-cell receptor beta chain rearrangements.	Targeted panel that measures expression levels of >20,000 human RefSeq genes.
Input	DNA: 20 ng RNA: 10 ng	DNA: 10 ng RNA: 10 ng	DNA: 10-1000 ng RNA: 10-1000 ng	RNA: 10-1000 ng	RNA: 1-100 ng
FFPE compatibility	No	No	Yes	No	Yes
Genomic content	54 genes (568 amplicons)	203 genes (3069 DNA and 1701 RNA)	Varies	RNA evaluation of TCRß chain rearrangements, including CDR1, CDR2, and CDR3 (with up to 400 bp read-length amplicons)	> 20,000 genes (20,802 amplicons)
Read length	2 x 150 bp	2 x 150 bp	2 x 150 bp	2 x 150 bp	2 x 150 bp
Estimated Data Required per Sample	Coverage: 1000x DNA: 2M clusters RNA: .25M clusters	Coverage: 500x	Varies	Varies	RNA: 10M clusters
Ideal instrument	MiniSeq System, MiSeq System, MiSeqDx System in Research Mode, NextSeq 550 System	MiniSeq System, MiSeq System, MiSeqDx System in Research Mode, NextSeq 550 System	MiniSeq System, NextSeq 550	MiniSeq System, MiSeq System, NextSeq 550 System	NextSeq 550 System

TRUSIGHT PANELS—ENRICHMENT-BASED SEQUENCING KITS

Product	TruSight Cancer Panel[a]	TruSight RNA Pan-Cancer Panel[a]	TruSight RNA Fusion Panel[a]	TruSight Tumor 170[a]
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,[b] HiSeq 2500 System[c]
UMI Tool Kit compatible	–	–	–	●

a. Decoupled oligo content can be combined with Nextera Flex for Enrichment
b. The TruSight Tumor 170 Panel has not been validated by Illumina on the NovaSeq 6000 System
c. The HiSeq 2500 System is no longer available for purchase; consult with your local service lab regarding system availability

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STEP 2
Choose a sequencer





Product	iSeq 100 System	MiniSeq System		MiSeq System		NextSeq 550 System		NovaSeq 6000 System			
Most important to me	Accessibility, 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	i1	Mid-output	High-output	Nano[c]	High-output	Mid-output	High-output	SP	S1[d]	S2[d]	S4[d]
Number of panels processed per flow cell											
TruSight Tumor 170 Panel	–	–	–	–	–	–	8	–	32[e]	32[e]	32[e]
TruSight Tumor 15 Panel	4	8	2	–	8	24	–	–	–	–	–
TruSight Cancer Panel	6	12	24	12	24	96	96	–	–	–	–
TruSight RNA Pan-Cancer Panel	4	8	24	–	8	24	24	96	48[e]	–	–
TruSight Myeloid Panel	1	2	8	–	8	40	96	–	–	–	–
TruSight RNA Fusion Panel	–	–	8	–	8	24	–	24	48[e]	–	–
AmpliSeq for Illumina *BRCA* Panel	96 germline[e] 12 somatic	96 germline[e] 24 somatic	–	–	96 germline[e] 80 somatic	–	–	–	–	–	–
AmpliSeq for Illumina Cancer Hotspot Panel v2	16	32	–	96	–	–	–	–	–	–	–
AmpliSeq for Illumina Comprehensive Cancer Panel	–	–	–	–	–	12	–	30	52	104	312
AmpliSeq for Illumina Comprehensive Panel v3	–	1	–	3	–	48[e,f]	–	100	192[e,f]	–	–
AmpliSeq for Illumina Focus Panel	8[f]	16	–	48[f]	–	–	–	–	–	–	–
AmpliSeq for Illumina Immune Response Panel	–	8	24	–	24	96	–	192	192	–	–

a. See page 129 in the Instruments chapter to learn more
b. Comparisons among Illumina sequencing portfolio
c. See iSeq 100 System for additional run modes and kit type options
d. Values supplied represent sequencing performed using the NovaSeq Xp workflow; the standard workflow will have lower throughput values
e. Limited by indexing
f. Combined DNA and RNA libraries

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STEP 3
Obtain results

Targeted resequencing ▼

1 Align/Call variants

Illumina DRAGEN Somatic Pipeline
- Builds on the DRAGEN Germline Pipeline and consumes a tumor and normal (optional) samples and calls somatic SNVs and INDELs
- Accepts multiple Tumor-only or Tumor/Normal pairs, which will be launched in independent analyses

Illumina DRAGEN RNA Gene Fusion Detection Pipeline
- Performs transcriptome analysis starting with splice junction discovery and alignment, followed by gene fusion detection

Local Run Manager (LRM) RNA Fusion Module (RNA Fusion)
- Analyzes RNA Fusions on any Illumina benchtop instrument except iSeq 100

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

Nextera Flex for Enrichment Kit

The Nextera Flex for Enrichment solution combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type, input amount, and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples		96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples		16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 96 samples		96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 16 samples		16	20025519
Panel		**No. of Enrichment Reactions**	
Illumina Exome Panel - Enrichment Oligos only		8 or 12	20020183
TruSight Cancer – Enrichment Oligos only		4 or 8	FC-121-0202
TruSight One – Enrichment Oligos only		6	20029227
TruSight One Expanded – Enrichment Oligos		6	20029226
TruSight Cardio – Enrichment Oligos only		8	20029229
TruSeq Neurodegeneration - Enrichment Oligos only		8	20029550
Index Adapter Component			
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	96	20027215
IDT for Illumina Nextera DNA UD Indexes Set D	96	96	20027216

TruSight Tumor 170

TruSight Tumor 170, an NGS assay covering 170 genes associated with common solid tumors, is an enrichment-based targeted panel that simultaneously analyzes DNA and RNA across 170 genes and variant types. The comprehensive nature provides laboratories with a deep view into the genetics of cancer. Libraries prepared with the TruSight Tumor 170 Panel are compatible with the NextSeq 500 System and the NextSeq 550 System.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
TruSight Tumor 170 Kit, With NextSeq v2.5 Reagents (24 Samples)	16	24	20028821
TruSight Tumor 170 Library Prep Kit	16	24	OP-101-1004

See Services section on page 147 for Illumina instrument service plan options.

Select Illumina reagents and consumables are available with product attributes that might benefit clinical research laboratories. See page 95 for details.

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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. Libraries prepared with the TruSight RNA Fusion Panel are compatible with the MiniSeq System, the MiSeq System, the MiSeqDx System in Research Mode, the NextSeq 500 System, and the NextSeq 550 System.

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 System high-throughput sequencing reagents			MS-102-3003

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. 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

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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[a]		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

[a]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
Nextera Flex for Enrichment	12		98	20025524
	1		16	20025523
	12		96	20025520
	1		16	20025519
Training				
TruSight Rapid Capture Library Prep Panel training at customer site				TR-204-0016

a. Requires the purchase of a TruSight Rapid Capture Kit

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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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

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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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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 UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

AmpliSeq for Illumina Myeloid Panel

The Myeloid Panel enables concurrent analysis of both DNA and RNA from blood and bone marrow samples in a single assay to study biomarkers associated with hematologic malignancies. The panel covers relevant targets for these major myeloid disorders: acute myeloid leukemia (AML), myeloid dysplastic syndrome (MDS), myeloproliferative neoplasms (MPN), chronic myeloid leukemia (CML), chronic myelomonocytic leukemia (CMML), and juvenile myelomonocytic leukemia (JMML).

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Myeloid Panel		24	20024478
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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AmpliSeq for Illumina Childhood Cancer Panel

The AmpliSeq Childhood Cancer Panel for Illumina provides a targeted resequencing solution for comprehensive evaluation of somatic variants associated with childhood and young adult cancers.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Childhood Cancer Panel		24	20028446
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

AmpliSeq for Illumina TCR beta-SR Panel

AmpliSeq for Illumina TCR beta-SR (Short Read) Panel is a highly multiplexed targeted resequencing panel intended for use with multiple sample types, including formalin-fixed paraffin-embedded (FFPE) tumor samples. It is designed to measure T-cell diversity and clonal expansion by sequencing T-cell receptor (TCR) beta chain rearrangements. You can use a single pool of multiplex PCR primers, library reagents, and sample barcodes to generate libraries from DNA/RNA extracted from FFPE, whole blood, fresh-frozen tissue, or fluorescence activated cell sorting (FACS)-sorted cells.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina TCR beta-SR Panel		24	20031675
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel

AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel is a highly multiplexed targeted resequencing panel to measure T-cell diversity and clonal expansion by sequencing T-cell receptor (TCR) beta chain rearrangements. Use a single pool of multiplex PCR primers, library reagents, and sample barcodes, to generate libraries from RNA extracted from whole blood, fresh-frozen tissue, or FACS-sorted cells.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel		24	20024479
Companion Kits			
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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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 150 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	Sequence	Analyze and Report
		
AmpliSeq for Illumina Panels Custom Panels Community Panels On-Demand Panels **Nextera Flex for Enrichment + TruSight Sequencing Panels** TruSight One Panel TruSight One Expanded TruSight Cardio Panel TruSeq Neurodegeneration Panel	**iSeq 100 System** **MiniSeq System** **MiSeq System** **MiSeqDx System** **NextSeq 550 System** **NovaSeq 6000 System** See page 42 to learn how many samples can be run on each sequencing platform	**Available on Local Run Manager Software** **BaseSpace Sequence Hub** **Illumina DRAGEN Joint Genotyping/Population Pipeline**

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

Training	Service contracts	Professional services
Customer site training Library prep with MiSeq System Nextera Flex for Enrichment	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Workflow Design and Evaluation Process** **Validation Services** IQ/OQ/PQ[a]

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

Visit https://emea.illumina.com/clinical/reproductive-genetic-health/genetic-health.html for more details.

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Innovation. Discovery. Application.

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.

TruSight One Sequencing Panels provide research labs with an affordable solution for managing a diverse assay portfolio. The TruSight One Sequencing panels provides comprehensive coverage of > 4800 disease-associated genes, while the TruSight One Expanded Sequencing Panel targets ~1900 additional genes with disease associations as reported in recent scientific publications.

Cardiac research (genetic conditions research)

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

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.

Neurodegeneration Research

Complex neurological diseases include Alzheimer's and Parkinson's disease, neuromuscular disorders such as amyotrophic lateral sclerosis (ALS), and psychiatric conditions including depressive disorders and schizophrenia, which are all the result of multifactorial interactions.

NGS studies reveal the full genomic complexity of neurological diseases. The interplay between heritable and nonheritable mutations, epigenetics, and other factors requires NGS-level analyses to increase our understanding.

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STEP 1
Choose a sequencing panel

Product		Maximum number of samples per flow cell at optimal kit configuration							
	Genomic content covered	iSeq 100	MiniSeq System mid-output	MiniSeq System high-output	MiSeq System v2 chemistry[a]	MiSeq System v2 nano/micro	miSeq System v3 chemistry	NextSeq 550 System mid-output	NextSeq 550 System high-output
TruSight One Panel[b]	12 Mb	Not recommended	Not recommended	2	1	0/0	2	12	36
TruSight One Expanded Panel[b]	16.5 Mb	Not recommended	Not recommended	1	0	0/0	1	7	24
TruSight Cancer Panel[b]	255 kb	4	8	24	12	1/4	24	96	384
TruSight Cardio Panel[b]	244 kb	4	8	24	12	1/4	24	96	384

Contact your local account representative for information on AmpliSeq for Illumina Custom Panels and AmpliSeq for Illumina On-Demand Panels

a. Limited by indexes

b. This kit and system combination can be used with Nextera Flex for Enrichment. The recommended read length for this product is 2 x 150 bp.

STEP 2
Choose a sequencing system





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[a] bench-top instrument sample throughput with Dx option	Highest[a] sample throughput benchtop simplicity, and low cost per sample
On-board informatics	●	●	●	–	–
Run mode/kit type	Standard	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	SP, S1, S2, S4
Flow cells processed/ run	1	1	1	1	1 or 2

a. Comparisons among Illumina sequencing portfolio

Visit https://emea.illumina.com/clinical/reproductive-genetic-health/genetic-health.html for more details.

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STEP 3
Obtain results

Targeted resequencing ▼

1

Align/Call variants

DNA Enrichment workflow
- Enables analysis of panels using capture-based methods, such as the Nextera Flex for Enrichment, 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
- 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
- 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

Illumina DRAGEN Joint Genotyping Pipeline
- Implements gVCF and combine gVCF and joint calling in a multi step approach.
- Supports an SV Denovo mode, taking BAMs or CRAMs as input

Illumina DRAGEN Germline Pipeline
- Processes an entire human genome at 30× coverage in about 25 minutes
- Detects small variants with high analytical sensitivity and specificity
- Enables data analysis onsite or in the cloud
- Analyzes sequencing data from whole-genome, whole exome, and targeted panels

2

Interpret/Report

BaseSpace Variant Interpreter
- Enables rapid, rich annotation, filtering, and interpretation of genomic data, customizes workflows and summarize 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 expedite interpretation of variants into meaningful data with confidence

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

BaseSpace Sequence Hub 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 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 compatible with all Illumina systems.

Ordering information

Product		No. of samples	Catalog no.
AmpliSeq Custom DNA Panel for Illumina			20020495
AmpliSeq Custom DNA Large Panel for Illumina			20020497
AmpliSeq for Illumina Custom RNA Panel			20020496
AmpliSeq Custom RNA Fusion Panel for Illumina			20032798
Companion Kits	**No. of indexes**	**No. of samples**	**Catalog No.**
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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

AmpliSeq for Illumina Community Panels

Community panels contain content selected and designed with input from leading researchers. These panels provide an easy, fast way to start targeted resequencing studies in a specific disease area. For maximum flexibility, the panels are fully customizable. Community panels are made to order and available in large pack sizes.

Ordering information

Product	No. of samples	Catalog no.
AmpliSeq for Illumina Cardiac Arrhythmias and Cardiomyopathy Research Panel		20020495
AmpliSeq for Illumina Deafness Research Panel v2		20020495
AmpliSeq for Illumina Autism Research Panel		20020497
AmpliSeq for Illumina BRCA Plus, Extended Hereditary Breast and Ovarian Research Panel		20020495
AmpliSeq for Illumina Cardiovascular Research Panel		20020497
AmpliSeq for Illumina Colon and Lung Research Panel v2		20020495
AmpliSeq for Illumina Dementia Research Gene Panel		20020495
AmpliSeq for Illumina Dermatology Research Panel v2		20020495
AmpliSeq for Illumina Dysmorphia-Dysplasia Research Panel v2		20020497
AmpliSeq for Illumina Gastrointestinal Research Panel v2		20020495
AmpliSeq for Illumina Hearing Loss Research Panel v1		20020495
AmpliSeq for Illumina Hematology Research Panel		20020497
AmpliSeq for Illumina Inborn Errors of Metabolism Research Panel v2		20020497
AmpliSeq for Illumina Inherited Cancer Research Panel		20020495
AmpliSeq for Illumina Long Non-Coding RNA Research Panel		20020496
AmpliSeq for Illumina Neurological Research Panel		20020497
AmpliSeq for Illumina Noonan Research Panel		20020495
AmpliSeq for Illumina Ophthalmic Research Panel		20020497
AmpliSeq for Illumina Primary Immune Deficiency Research Panel v2		20020497
AmpliSeq for Illumina Pulmonary Research Panel v2		20020495
AmpliSeq for Illumina RNA Inflammation Response Research Panel		20020496
AmpliSeq for Illumina Endocrine Research Panel v2		20020497
AmpliSeq for Illumina Epilepsy Research Panel		20020497

Companion Kits	No. of indexes	No. of samples	Catalog No.
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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

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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 compatible with all Illumina systems.

Ordering information

Product		No. of reactions	Catalog no.
AmpliSeq™ for Illumina On-Demand Panel (1-50 Genes)		24	20023977
AmpliSeq™ for Illumina On-Demand Panel (51-300 Genes)		24	20023983
AmpliSeq™ for Illumina On-Demand Panel (301-500 Genes)		24	20023978
AmpliSeq™ for Illumina On-Demand Panel (1-50 Genes)		96	20023979
AmpliSeq™ for Illumina On-Demand Panel (51-300 Genes)		96	20023980
AmpliSeq™ for Illumina On-Demand Panel (301-500 Genes)		96	20023981
Companion Kits	**No. of indexes**	**No. of samples**	**Catalog No.**
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

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 compatible with the MiniSeq System, MiSeq System, NextSeq 550 System, and NovaSeq 6000 System.

Ordering information

Bundled Solutions	No. of samples	Catalog no.
TruSight One Sequencing Panel	9	FC-141-1006
TruSight One Sequencing Panel	36	FC-141-1007
Enrichment Panels		
TruSight One Enrichment Oligos only (6 enrichment reactions)		20029227
Accessory Products		
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples (8, 12-plex enrichment reactions)	96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples (16, 1-plex enrichment reactions)	16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents	96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents	16	20025519
IDT® for Illumina Nextera DNA UD Indexes Set A (96 indexes, 96 samples)	96	20027213

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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 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

Bundled Solutions	No. of samples	Catalog no.
TruSight One Expanded Sequencing Panel	36	FC-141-2007
Enrichment Panels		
TruSight One Expanded – Enrichment Oligos only (6 enrichment reactions)		20029226
Accessory Products		
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples (8, 12-plex enrichment reactions)	96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples (16, 1-plex enrichment reactions)	16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents	96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents	16	20025519
IDT® for Illumina Nextera DNA UD Indexes Set A (96 indexes, 96 samples)	96	20027213

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 Systems	1	12	FC-141-1010
TruSight Cardio Sequencing Kit for NextSeq and NextSeq 550 Dx Systems	4	48	20035190
ruSight Cardio - Enrichment Oligos only	8		20029229

TruSeq Neurodegeneration Panel

The TruSeq Neurodegeneration Sequencing Panel enables systematic investigation of neurodegenerative disease–associated genes. The TruSeq Neurodegeneration Sequencing Panel facilitates fine-mapping validation and identification of rare, putative functional alleles and additional risk alleles. It offers the ability to survey both protein-coding and noncoding regulatory regions of candidate genes for efficient, cost-effective investigation of major neurodegenerative diseases.

Ordering information

Product	No. of enrichments	No. of samples	Catalog no.
TruSeq Neurodegeneration - Enrichment Oligos	8		20029550

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Library prep ordering information

Nextera Flex for Enrichment Kit

The Nextera Flex for Enrichment Kit combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type and amount, and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples		96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples		16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 96 samples		96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 16 samples		16	20025519
Panel		**No. of Enrichment Reactions**	
Illumina Exome Panel - Enrichment Oligos only		8 or 12	20020183
TruSight Cancer – Enrichment Oligos only		4 or 8	FC-121-0202
TruSight One – Enrichment Oligos only		6	20029227
TruSight One Expanded – Enrichment Oligos		6	20029226
TruSight Cardio – Enrichment Oligos only		8	20029229
TruSeq Neurodegeneration - Enrichment Oligos only		8	20029550
Index Adapter Component			
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	96	20027215
IDT for Illumina Nextera DNA UD Indexes Set D	96	96	20027216

Workflow Design and Evaluation Service

The Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can increase the capabilities of your laboratory. Receive in-person consultation to establish needs and goals, sequencing of your samples at the Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	20016091
iSeq 100 Workflow Design and Evaluation Service	20023613
MiniSeq Workflow Design and Evaluation Service	20003924
MiSeq Workflow Design and Evaluation Service	SP-801-1002
NextSeq Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	20016091

See page 150 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 95.

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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
		
Nextera Flex for Enrichment + exome content from Illumina or third-party oligo vendors	**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 Flex for Enrichment + Illumina Exome Panel	**Tiered service plans** NextSeq 550 System NovaSeq 6000 System	**Workflow Design and Evaluation** Standard application functional testing with your samples **Validation Services** IQ/OQ/PQ[a]

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

Visit https://emea.illumina.com/techniques/sequencing/dna-sequencing/targeted-resequencing/exome-sequencing.html for more details.

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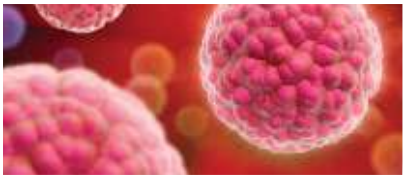
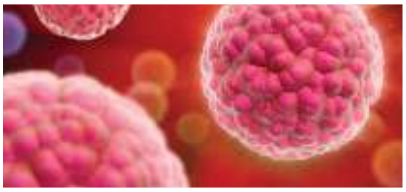


Cancer exome (cancer research)
Sequencing only 1% to 2% of the genome makes it possible to sequence to higher depth more economically, allowing researchers to focus on coding regions only. By performing tumor–normal exome sequencing, researchers can focus on the changes most likely to be tumorigenic.

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	Nextera Flex for Enrichment + Illumina Exome	AmpliSeq for Illumina Exome Panel[a]
Most important to me	Ease of use, FFPE, large panels up to and including exome, direct sample input for blood and saliva	Low input requirement, use with degraded samples, ease and speed of workflow
Least important to me	Very low input	DNA input amount
Target region size	21,415 genes	~59 Mb
Target region description	98.3% RefSeq	97% CCDS
On-target percent	–	> 95%
Recommended read length	2 × 100 bp	2 × 150 bp
Kit configurations	8 enrichment reactions	8 samples

a. Decoupled oligo content can be combined with Nextera Flex for Enrichment

STEP 2
Choose a sequencing system

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

BaseSpace Enrichment App
- Provides rapid alignment and variant detection for targeted sequencing data
- Maps reads using the Isaac aligner; performs small, structural, and copy number variant calling; annotates variants; and calculates enrichment metrics

Isaac Enrichment
- 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
- 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
- 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 identify disease-associated variants in data sets rapidly

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 expedite interpretation of variants into meaningful data with confidence

4 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

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

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Library prep ordering information

Nextera Flex for Enrichment Kit

The Nextera Flex for Enrichment Kit combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type and amount, and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Library Prep Component			
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples		96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples		16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 96 samples		96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 16 samples		16	20025519
Panel		**No. of enrichment reactions**	
Illumina Exome Panel - Enrichment Oligos only		8 or 12	20020183
TruSight Cancer – Enrichment Oligos only		4 or 8	FC-121-0202
TruSight One – Enrichment Oligos only		6	20029227
TruSight One Expanded – Enrichment Oligos		6	20029226
TruSight Cardio – Enrichment Oligos only		8	20029229
TruSeq Neurodegeneration - Enrichment Oligos only		8	20029550
Index Adapter Component			
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	96	20027215
IDT for Illumina Nextera DNA UD Indexes Set D	96	96	20027216

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

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Workflow Design and Evaluation Service

The Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can increase your laboratory capabilities. The service includes in-person consultation to establish needs and goals, sequencing of your samples at the Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	20016091

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Concierge service for your custom panel

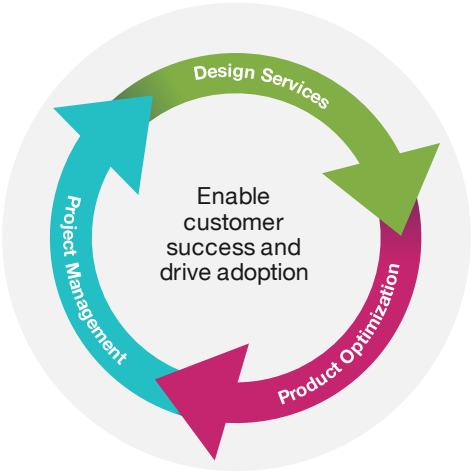
A dedicated expert from design to delivery

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

Concierge services offer design and product optimization assistance for custom array and sequencing products. Our scientists provide customers with innovative design features to maximize performance.

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



Who We Are

We are an experienced team of scientists and project managers that enables customer success and drives adoption of Illumina technology through custom design and product optimization of targeted sequencing and array products.

Our Mission

- Provide expert design support to achieve optimal designs and product performance
- Enable new design and product features to create solutions for innovative genomics applications

What we do

We create custom and personalized solutions through:

- **Design Services** to enable and optimize custom product designs
- **Functional Product Testing** and **Optimization** to achieve high coverage uniformity across target regions
- **Project Management** support to achieve complex goals and desired product features

Applications

- Direct-to-consumer genomic products
- Agrigenomics
- Targeted panels for detection of germline and somatic variants
- RNA expression and epigenetics
- Virology and microbiology

Supported Products

- Custom Infinium iSelect
- Custom Infinium Methylation
- AmpliSeq for Illumina
- Nextera Flex for Enrichment
- Nextera Rapid Capture
- TruSeq Genotype Ne
- TruSeq Targeted RNA Expression

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Shotgun metagenomics

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

Key strengths

- Enables culture-free identification and classification of complex microbial communities
- Accelerates discovery of new markers that translate to virulence or antibiotic resistance
- Promotes accurate evaluation of microbial biodiversity and abundance within and between samples
- Facilitates *de novo* discovery and characterization of novel species and assembly of new genomes

Prepare library	Sequence	Analyze and Report
		
TruSeq DNA PCR-Free Library Prep Kit **ITS Metagenomic Sequencing Protocol** **Nextera DNA Flex Library Prep Kit**	**iSeq 100 System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on BaseSpace Sequence Hub** 16S Metagenomics Kraken2 MetaPhlAn
	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	**Tiered service Plans** iSeq 100 System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Workflow Design and Evaluation Service**

Visit https://emea.illumina.com/areas-of-interest/microbiology/environmental-metagenomics.html for more details.

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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 can evaluate bacterial, fungal, and viral diversity to detect the abundance of microbes in various environments, including unculturable microorganisms.

Human microbiome research

The human gut microbiome varies widely among individuals and populations, and can affect health status. Leveraging shotgun metagenomics, scientists can identify the impact of human genetics on variations in the gut microbiome. Shotgun metagenomics also enables detection of anti microbial resistance genes and virulence genes that can impact human health.

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STEP 1
Choose a library prep method

Product	Nextera DNA Flex Library Prep Kit	TruSeq DNA PCR-Free Library Prep Kit
Most important to me	Simple and versatile sample-to-data workflow	Lowest[a] bias to achieve highest[a] coverage
Least important to me	PCR-free workflow	Low DNA input quantity
Input quantity	1–500 ng DNA	1 µg DNA
Assay time	3–4 hrs	5 hrs
Diversity	≥ 1B unique fragments	≥ 1B unique fragments
Genome coverage	Complete	Complete
Fragmentation process	Enzymatic	Mechanical

a. Comparisons are within the Illumina targeted sequencing portfolio

STEP 2
Choose a sequencing system

Product	iSeq 100 System	MiSeq Series
Most important to me	Ease of use and accessibility	Scalable throughput
16S or ITS metagenomic samples per flow cell at 100K[a] clusters per sample	40	625[b]
Run mode/kit type	Standard	Mid-output/High-output
Flow cells processed/run	1	1

a. Demonstrated protocol for a range of clusters per sample

b. When used with the MiSeq Reagent Kit v3 (600-cycle)

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STEP 3
Obtain results

Shotgun metagenomics ▼

Add biological context

Kraken2 Metagenomics B
- Assigns taxonomic labels to short DNA sequences with high sensitivity and speed using exact k-mer alignments and a novel classification algorithm
- Uses RefSeq reference genomes as the database for classification

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

CosmosID B
- Provides an easy-to-use, third-party application that performs microbial identification at the species, subspecies, or strain level
- Features a private, curated database

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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 supports use with small (bacteria, archaea, viruses, 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		96	20018706
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	96	20027215
Training			
Nextera DNA Flex Library Prep Kit training at customer site			20022900

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

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Targeted metagenomics

Sequence bacterial or fungal communities to identify and classify microbial populations

Key strengths

- Enables bacteria and fungus identification and comparison within a mixed population sample
- Promotes accurate evaluation of microbial biodiversity and abundance within and between samples

Prepare library	Sequence	Analyze and Report
		
16S Metagenomic Sequencing Protocol **ITS Metagenomic Sequencing Protocol**	**iSeq 100 System** **MiSeq System**	**Available on Local Run Manager Software, BaseSpace Sequence Hub** 16S Metagenomics **Available on BaseSpace Sequence Hub** 16S Metagenomics ITS Metagenomics
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Visit www.illumina.com/areas-of-interest/microbiology/environmental-metagenomics.html for more details.

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Innovation. Discovery. Application.

Microbiome

16S and ITS sequencing are established methods for achieving genus-level surveillance of bacterial and fungal populations, respectively, in human, animal, and environmental microbiome studies.

Human microbiome research

The human gut microbiome varies widely among individuals and populations, and can affect health status. Leveraging targeted metagenomics, scientists can identify the impact of human genetics on variations within the gut microbiome.

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STEP 1
Choose a library prep method

Product	16S Metagenomic Sequencing Protocol[a]	ITS Metagenomic Sequencing Protocol[a]
Input quantity	10-15 ng DNA	10-15 ng DNA
Assay time	~7 hrs	~6 hrs
Multiplexing capacity	96[b]	384[b]
Genome coverage	V3 and V4 regions of the 16S rRNA gene	ITS1 region of rRNA cistron
Sequencing method	Amplicon sequencing	Amplicon sequencing
View protocol	16S protocol	ITS protocol

a. Demonstrated protocol

b. Multiplexing capacity is determined by number of indexes available and instrument capacity

STEP 2
Choose a sequencing system

Product	iSeq 100 System	MiniSeq System	MiSeq System
Most important to me	Affordability and efficiency	Simplicity and instrument affordability	Longest read lengths in Illumina portfolio, well-suited for microbiology applications
16S metagenomic samples per flow cell at 100K clusters per sample[a]	40	250	250
ITS metagenomic samples per flow cell at 100K clusters per sample[a]	40	250	250
Recommended read length for targeted metagenomics	2 x 150 bp	2 x 150 bp	2 x 250 bp
Run mode/kit type	Standard	Mid-output/ High-output	Research mode and Dx
Max output per flow cell	1.2 Gb	2.4 Gb (Mid-output) 7.5 Gb (High-output)	0.5 Gb (Nano v2) 1.2 Gb (Micro v2) 7.5 Gb (Standard v2) 15 Gb (Standard v3)
Flow cells processed/run	1	1	1

a. Refer to demonstrated protocol for range of clusters per sample

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Library prep ordering information

16S Metagenomic Sequencing Protocol (Demonstrated Protocol)

Variable regions of 16S rRNA are frequently used in phylogenetic classifications, such as genus or species, in diverse microbial populations. This protocol describes a method for preparing samples for sequencing the variable V3 and V4 regions of the 16S rRNA gene. When combined with a benchtop sequencing system, on-board primary analysis, and secondary analysis using MiSeq Reporter or BaseSpace Sequence Hub, this protocol provides a comprehensive workflow for 16S rRNA amplicon sequencing.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
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
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
PhiX Control Kit v3			FC-110-3001

ITS Metagenomic Sequencing Protocol (Demonstrated Protocol)

The ITS Metagenomic Sequencing Protocol describes a method for preparing libraries that allows researchers to sequence the Internal Transcribed Spacer (ITS) Region 1 of the fungal rRNA cistron. The ITS region of the rRNA cistron is a useful DNA barcode for characterizing fungal and diverse environmental samples such as stool, sputum, skin, soil, and water. This multiplex PCR protocol details a workflow for the amplification and analysis of the ITS1 region to provide accurate classification resolution for a broad range of taxa.

Ordering information

Product	No. of indexes	No. of samples	Catalog no.
Nextera DNA CD Indexes	24	24	20018707
Nextera DNA CD Indexes	96	96	20018708
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
IDT for Illumina Nextera DNA UD Indexes Set A	96	96	20027213
PhiX Control Kit v3			FC-110-3001

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Custom sequencing

Target 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 1 kb exome
  - Combine with Illumina Custom Enrichment Panel (ICEP) or third-party oligos
  - 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 research, population genetics, and genetic disease research

Prepare library	Sequence	Analyze and Report
		
AmpliSeq for Illumina **Nextera Flex for Enrichment + Illumina Custom Enrichment Panel or third-party oligos** **Illumina Custom Enrichment Panel**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software, BaseSpace Sequence Hub** Resequencing
	Illumina Informatics Solutions BaseSpace Sequence Hub Run management | Analysis | Reporting	

Training	Service contracts	Professional services
Customer site training AmpliSeq for Illumina Nextera Flex for Enrichment + ICEP or third-party oligos Illumina Custom Enrichment Panel	**Tiered service plans** iSeq 100 System MiniSeq System MiSeq System NextSeq 550 System NovaSeq 6000 System	**Workflow Design and Evaluation Service** Standard application functional testing with your samples Concierge services

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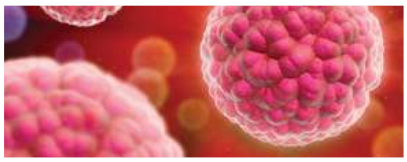
Antibacterial drug resistance (public health and epidemiology)
Drug resistance develops during treatment for certain viral infections as the proportion of naturally occurring drug-resistant variants in the body increases. Public health experts track the development of bacterial drug resistance with targeted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

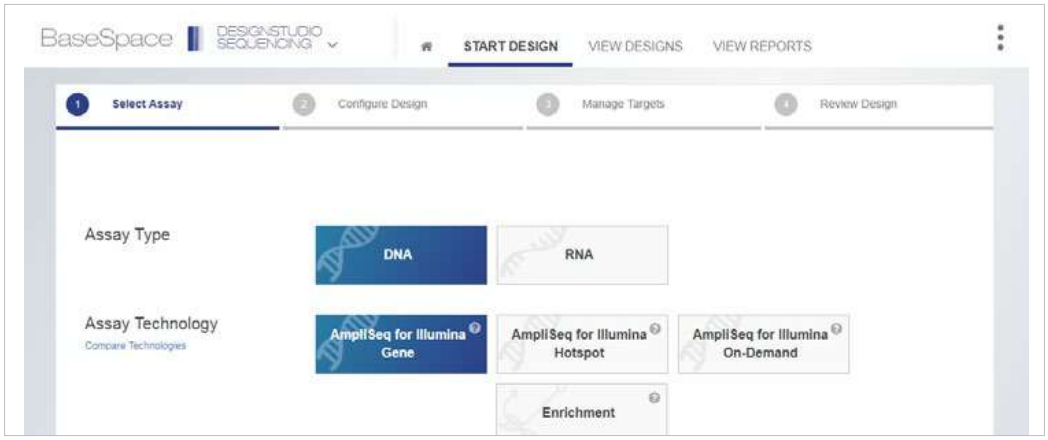
Variant identification (complex disease)
Rare genetic variants sometimes contribute to complex diseases, such as neuropsychiatric disorders. Targeted resequencing enables focused assessment of candidate pathogenic genes and the identification of rare genetic variants that contribute to disease pathogenesis.

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:

- AmpliSeq for Illumina
- Nextera Flex for Enrichment + Illumina Custom Enrichment Panel or third-party oligos
- TruSeq Targeted RNA Expression Kits



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

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STEP 1
Choose a library prep method

Product	AmpliSeq for Illumina Custom Panel	Nextera Flex for Enrichment Library Prep + Illumina Custom Enrichment Panel or third-party oligos
Most important to me	FFPE, low input, easy to use	Ease of use, FFPE, large panels up to and including exome, direct sample input for blood and saliva
Least important to me	Large genomic regions	Very low input
Input quantity	10 ng/pool	50ng–1000 ng without normalization
Assay time	5–7 hrs	6.5 hrs
Hands-on time	1.5 hrs	2 hrs
FFPE compatibility	Yes	Yes
Design	Customers use DesignStudio Software to design probes targeting their regions of interest	
Fragmentation method	None	None

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 price per sample[a]
Onboard informatics	●	●	●	–	–
Run mode/kit type	Standard	Mid-output/ High-output	Mid-output/ High-output	Mid-output/ High-output	SP, 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

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STEP 3
Obtain results

Targeted resequencing ▼

1 Align/Call variants

DNA Amplicon 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 expedite interpretation of variants into meaningful data with confidence

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

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Library prep ordering information

Nextera Flex for Enrichment Kit

The Nextera Flex for Enrichment Kit combines versatile, simple, and fast library prep and enrichment functionality for targeted enrichment and exome sequencing applications. It offers extraordinary flexibility for input type, input amount, and a wide range of supported enrichment sequencing applications encompassing custom panels, fixed panels, and whole-exome sequencing from Illumina or third-party vendors.

Ordering information

Product	No. of samples	Catalog no.
Library Prep Component		
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 96 samples	96	20025524
Nextera DNA Flex Pre-Enrichment Library Prep and Enrichment Reagents - 16 samples	16	20025523
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 96 samples	96	20025520
Nextera DNA Flex Pre-Enrichment Library Prep Reagents - 16 samples	16	20025519
Panel	**No. of enrichment reactions**	
Illumina Custom Enrichment Panel	8	20025371
Index Adapter Component		
IDT for Illumina Nextera DNA UD Indexes Set A	96	20027213
IDT for Illumina Nextera DNA UD Indexes Set B	96	20027214
IDT for Illumina Nextera DNA UD Indexes Set C	96	20027215
IDT for Illumina Nextera DNA UD Indexes Set D	96	20027216

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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 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	20019105
Training			
AmpliSeq for Illumina DNA Library Preparation Kit			20023392
Companion Kits	**No. of indexes**	**No. of samples**	**Catalog No.**
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

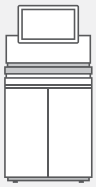
a. A number of ready-to-use AmpliSeq for Illumina panels are available. Visit https://emea.illumina.com/products/by-brand/ampliseq/ready-to-use-panels.html to learn more

Whole-transcriptome and stranded sequencing

Use strand information to analyze coding and multiple forms of noncoding RNA for a more complete picture of the transcriptome

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 RNA Exome**	**NextSeq 550 System** **NovaSeq 6000 System**	**Available on BaseSpace Sequence Hub and RNA-Seq** RNA Amplicon
	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	**Workflow Design and Evaluation Service** Standard application functional testing with your samples

Visit https://emea.illumina.com/techniques/sequencing/rna-sequencing/total-rna-seq.html for more details.

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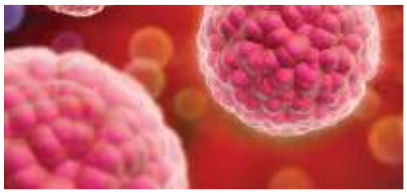

Cancer gene expression (cancer research)
Cancers accumulate numerous genetic changes, but typically, only a few changes actually drive tumor progression. Monitoring changes in expression of both coding and noncoding RNA can inform research questions about cancer classification and progression.

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,** Associate Professor; Director, WorldQuant Initiative for Quantitative Prediction Physiology and Biophysics/Feil Family Brain and Mind Institute/Institute for Computational Biomedicine, Weill Cornell Medicine

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STEP 1
Choose a library prep method

Product	TruSeq Stranded Total RNA with Ribo-Zero Kit	TruSeq RNA Exome
Most important to me	Capture coding and noncoding transcriptome	Capture coding transcriptome and RNA exome
FFPE compatibility	Yes	Yes
RNA input	0.1–1 µg[a]	10 ng from fresh or frozen sample, or 20 ng total RNA from FFPE samples
Assay time	1.5 days	2 days
Multiplexing	12–96 samples	Up to 24 single, 96 combinatorial (CD) dual
Available organisms	Human, mouse, rat, blood, plant	Human

a. Demonstrated performance at lower input quantities

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	SP, 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 ▼

Gene expression profiling ▼

1 Align/Call variants

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

Align/Count

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

2 Assemble/Count

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

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 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

BaseSpace Sequence Hub

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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 RNA and FFPE tissues, 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

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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			20020189
TruSeq RNA Enrichment (12 enrichments)			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, enrichment kit, an exome panel, and an index kit

Workflow Design and Evaluation Service

The Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can expand the capabilities of you laboratory. Receive in-person consultation to establish Workflow Design and Evaluation Service needs and goals, sequencing services of your samples at our Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation 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 Apps

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	**Workflow Design and Evaluation Service** Standard application functional testing with your samples

Visit https://www.illumina.com/techniques/sequencing/rna-sequencing/mrna-seq.html for more details.

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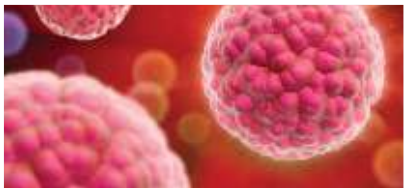
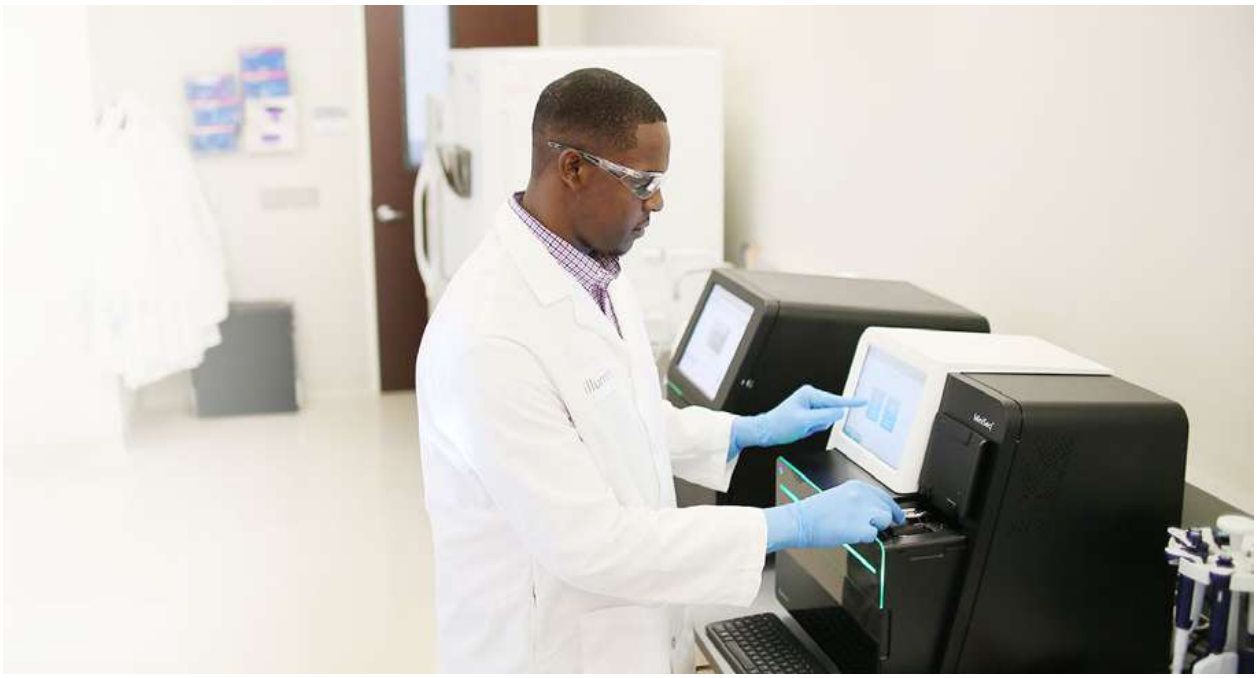


Cancer classification and progression (cancer research)
Cancers accumulate numerous genetic changes, but typically, only a few changes actually drive tumor progression. Monitoring cancer gene expression and transcriptome changes with RNA-Seq can help answer research questions on disease classification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

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

Gene expression profiling ▼

Align/Count

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

2 Assemble/Count

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

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 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

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		48	20020189
Enrichment Component			
TruSeq RNA Enrichment			20020490
Probe Panel			
Exome Panel (45 Mb)			20020183
Index Component			
TruSeq RNA Single Indexes Set A	12	48	20020492
TruSeq RNA Single Indexes Set B	12	48	20020493

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Workflow Design and Evaluation Service

The Workflow Design and Evaluation 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 to establish Workflow Design and Evaluation Service needs and goals, sequencing services of your samples at our Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	20003924
MiSeq Workflow Design and Evaluation Service	SP-801-1002
NextSeq Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation 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 86 for a complete list of targeted RNA expression kits **AmpliSeq for Illumina Transcriptome Human Gene Expression Panel** **Custom Targeted RNA panels**	**iSeq 100 System** **MiniSeq System** **MiSeq System** **NextSeq 550 System** **NovaSeq 6000 System**	**Available on Local Run Manager Software and BaseSpace Sequence Hub Platform** RNA Amplicon

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	**Workflow Design and Evaluation Service** Standard application functional testing with your samples

Visit https://www.illumina.com/products/by-type/sequencing-kits/library-prep-kits/truseq-targeted-rna.html for more details.

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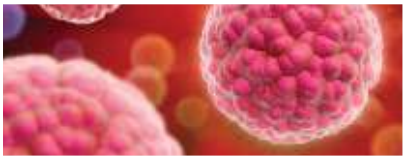

Disease progression (cancer research)
Mapping gene expression and transcriptome changes in cancer is important for understanding tumor classification and progression. Use cancer RNA-Seq to help determine which variants are expressed in cancer samples.

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
Choose a library prep method

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 sequencing system





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

RNA Amplicon workflow L
and TruSeq Targeted RNA

- 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

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 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 Custom RNA		1125 panel for 12-1200 amplicons	
AmpliSeq for Illumina cDNA Synthesis		96	20022654
AmpliSeq for Illumina Library PLUS		24	20019101
AmpliSeq for Illumina Library PLUS		96	20019102
AmpliSeq for Illumina Library PLUS		384	20019103
AmpliSeq UD Indexes for Illumina	24	24	20019104
AmpliSeq for Illumina CD Indexes Set A	96	96	20019105
AmpliSeq for Illumina CD Indexes Set B	96	96	20019106
AmpliSeq for Illumina CD Indexes Set C	96	96	20019107
AmpliSeq for Illumina CD Indexes Set D	96	96	20019167
AmpliSeq for Illumina CD Indexes Set A-D	384	384	20031676

Workflow Design and Evaluation Service

The Workflow Design and Evaluation Service will run a subset of your samples with your desired system and application in a real-world demonstration of how Illumina NGS can expand your laboratory capabilities. Receive in-person consultation to establish needs and goals, sequencing services of your samples at the Illumina Service Lab, and in-person consultation 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 Workflow Design and Evaluation Service	20023613
MiniSeq Workflow Design and Evaluation Service	20003924
MiSeq Workflow Design and Evaluation Service	SP-801-1002
NextSeq Workflow Design and Evaluation Service	SP-801-1003
NovaSeq Workflow Design and Evaluation Service	2001691

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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) transcriptomes

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** Small RNA
	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	**Workflow Design and Evaluation** Standard application functional testing with your samples

Visit https://emea.illumina.com/techniques/sequencing/rna-sequencing/small-rna-seq.html for more details.

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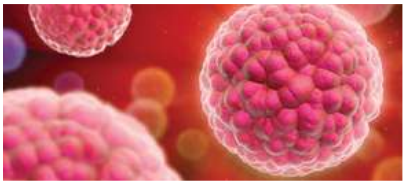

Mutation detection (cancer research)
Researchers often want to analyze precious tumor samples with high sensitivity to detect mutation levels as low as a single copy per cell. miRNA-Seq enables high-throughput profiling or deep-coverage discovery studies of the cancer miRNA transcriptome.

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 App B and Small RNA Analysis Module 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 153 for Workflow Design and Evaluation Service information.

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B BaseSpace Sequence Hub L Local Run Manager Software

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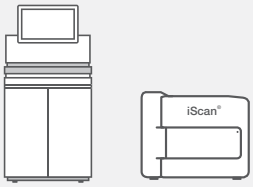
DNA methylation

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** **GenomeStudio Methylation Module** **Illumina DRAGEN Methylation Pipeline**
	Illumina Informatics Solutions BaseSpace Sequence Hub GenomeStudio	

Training	Service Contracts	Professional Services
Customer site training Coming soon	**Tiered service plans** NextSeq 550 System NovaSeq 6000 System iScan System	**Workflow Design and Evaluation** Standard application functional testing with your samples

* NextSeq System enables scanning of Infinium Methylation EPIC BeadChip

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

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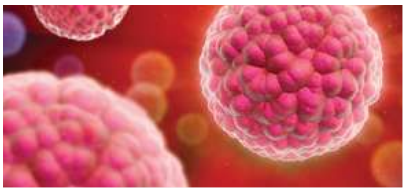

Cancer epigenetics (cancer research)

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.

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.

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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	3.3M
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 platform



Product	iScan System	NextSeq 550 System		NovaSeq 6000 System			
Most important to me	High-throughput processing of Infinium MethylationEPIC BeadChips	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	SP Xp	S1 Xp[a]	S2 Xp[a]	S4 Xp[a]
Whole-genome bisulfite sequencing Human methylation samples sequenced at > 30× coverage per flow cell	All BeadChips	1	1	---	8	8	24
Flow cells processed/run	---	1	1	1 or 2			
Scan time	5 min/sample	40 min/BeadChip		---	---	---	---

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

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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

Illumina DRAGEN Methylation Pipeline
- Analyzes whole-genome and targeted bisulfite DNA sequence data rapidly
- Performs alignment and methyl calling and calculates alignment and methylation metrics
- Supports libraries prepared using TruSeq DNA Methylation and TruSeq Methyl Capture library prep kits

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, compares samples, and integrates gene expression within one viewer

2 Visualize

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

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	No. of samples	Catalog no.
TruSeq Methyl Capture EPIC Library Prep Kit	12	FC-151-1002
TruSeq Methyl Capture EPIC Library Prep Kit	48	FC-151-1003

B BaseSpace Sequence Hub

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Infinium MethylationEPIC Kit

The unique combination of the comprehensive, expert-selected coverage, high sample throughput capacity, and affordable pricing make the Infinium Human MethylationEPIC BeadChip Kit an ideal solution for large sample-size genome–wide DNA methylation studies. Libraries prepared with the kit are compatible with the 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 153 for Workflow Design and Evaluation Service information.

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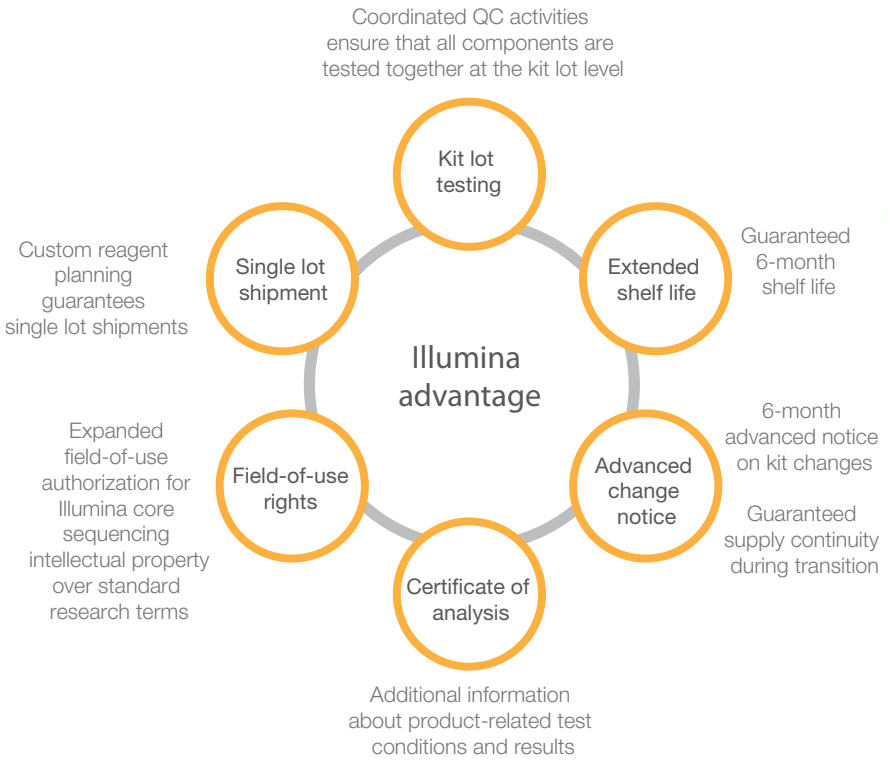
Illumina advantage

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**
	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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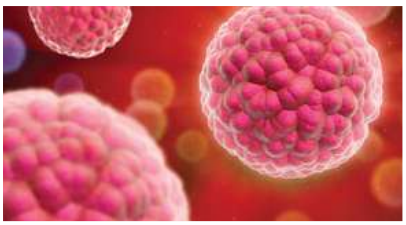

Tumor development and progression (cancer research)

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

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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
- Developed by the Broad Institute and Harvard and integrated into the BaseSpace platform by Illumina

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 153 for Workflow Design and Evaluation Service information.

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B BaseSpace Sequence Hub BSO BaseSpace Sequence Hub Onsite

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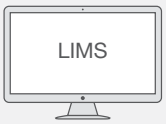
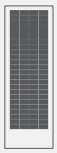
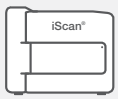
Infinium BeadChip technology

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

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

- Reproducible, high-quality data
- 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 turnaround time from DNA to genotype calls
  - Streamlined workflow
- Intuitive analysis tools to visualize and analyze data
- Flexible genotype plexity ranging from 500–5M probes
- Simultaneous multi-sample processing

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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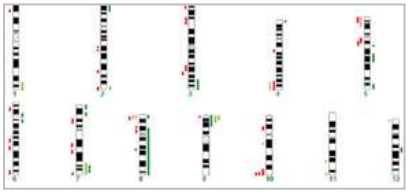
Infinium Human Genotyping Products

Population genetics

Illumina human genotyping arrays contain content supporting whole-genome, targeted genome, and exome analyses. Powered by Infinium assay technologies, Illumina arrays 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 (GWAS) and phenome-wide association studies (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 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.

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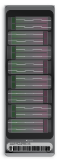
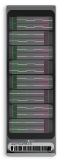
Human BeadChip Portfolio

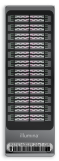
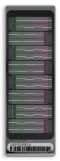
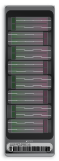
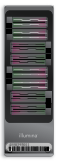
Genome-wide human genotyping arrays




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 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 Chinese populations	High genome-wide coverage, including rare variants	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	Not available	500K custom add-on
Assay type	HTS	HD Super	LCG	LCG Quad
Content Source	HapMap, 1000 Genomes phase 3	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 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

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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. Because 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.

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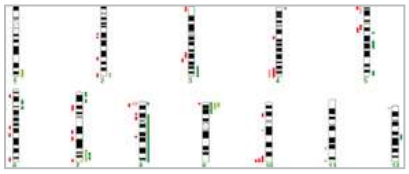
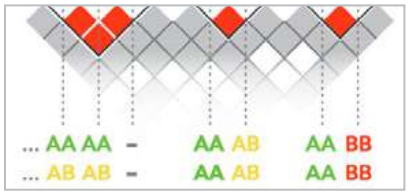


Fine mapping and candidate gene region genotyping

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

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.

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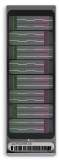
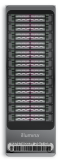
Non-Human Array Portfolio

Standard nonhuman genotyping arrays


Product	GGP BovineLD	BovineHD	BovineLD	BovineSNP50	OvineSNP50
Description	Genome-wide bovine genotyping	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 SNPs for major cattle breeds	50K array containing highly informative SNP for major ovine 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 ovine
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 analysis	Mapping essentially derived varieties	Genomic selection
Most important to me	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 that is easily customizable to support other applications	Cost-effective whole-genome studies
Number of loci	~135K	~75K	~170K	~3K	~56K
Add-on availability	–	–	–	70K	–
Assay type	HTS	HD Ultra	HD Ultra	HD	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
Number of loci	~6.5K	~65K	~65K	~10K	143K
Add on availability	80K	25K	–	–	–
Assay Type	HD	HD Ultra	HD Ultra	HD	HD

Product	GGP Equine
Description	Identification of genes and polymorphisms that contribute to traits of interest in all major horse breeds
Key applications	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

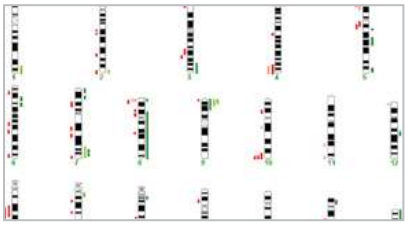
Custom genotyping is the ideal solution for screening large sample sets against novel or targeted content. With custom designs, researchers can target regions of the genome relevant to their specific research interests.

Fully custom arrays can be developed with up to 700K markers targeting any loci across the genome for any species, including novel organisms. Illumina also flexible throughput options and 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 predesigned products

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.

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Custom and Infinium XT Portfolio

Scalable and flexible arrays




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 3000–700K SNPs, increasingly popular for high-density custom panel designs that support high-throughput processing	Supporting 700,001–2.5M custom assays, unique for 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	3000–700K attempted assays	700,001–2.5M attempted assays
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

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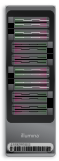
Infinium Cytogenetics

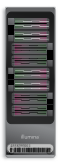
Cost-effective and streamlined screening



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, FANTOM5

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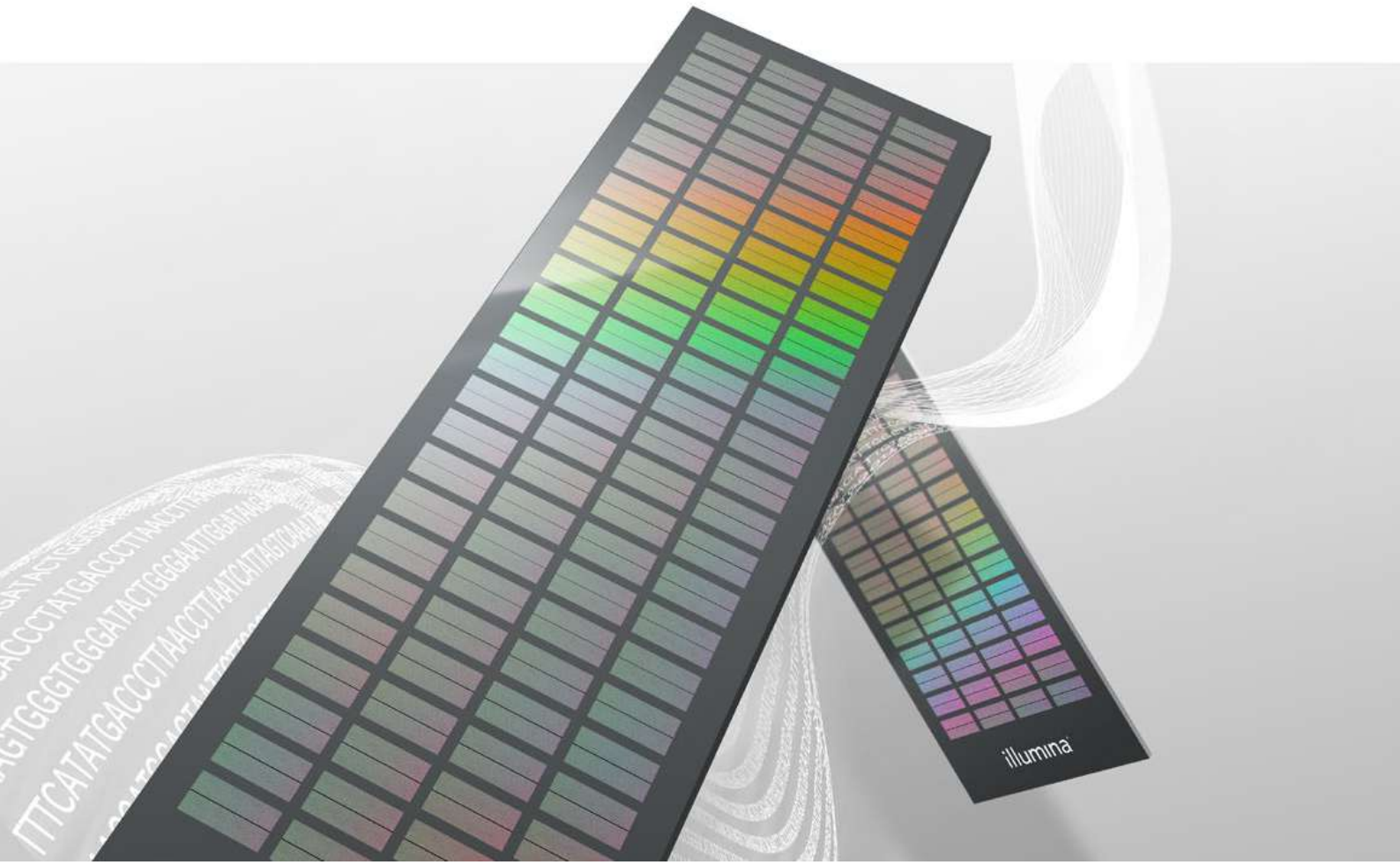
Infinium BeadChip systems

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 multiple millions of 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 an account manager can also help narrow down the options depending on your needs and expected throughput.



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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 (24 BeadChip) 110V	20028878
	Infinium HD Starter Kit (24 BeadChip) 220V	20028879
	Infinium HD Starter Kit (8 BeadChip) 110V	20028872
	Infinium HD Starter Kit (8 BeadChip) 220V	20028873
Infinium HTS Starter Kits	Infinium HTS Starter Kit (24 BeadChip) 110V	20028876
	Infinium HTS Starter Kit (24 BeadChip) 220V	20028877
	Infinium HTS Starter Kit (8 BeadChip) 110V	20028874
	Infinium HTS Starter Kit (8 BeadChip) 220V	20028875
Infinium XT Kit options	Infinium XT Starter Kit (24 BeadChip) 110V	20031992
	Infinium XT Starter Kit (48 BeadChip) 110V	20031993
	Infinium XT Starter Kit (24 BeadChip) 220V	20031915
	Infinium XT Starter Kit (48 BeadChip) 220V	20031916
Tip guides Required when automating the Infinium workflow or when using 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
Te-Flow accessories		
Infinium HD	Infinium HD Te-Flow Chamber	WG-10-202
	Infinium HD Spacers (500)	WG-10-203
Infinium LCG	Infinium LCG Te-Flow Chamber (10)	WG-100-1001
	Infinium LCG Spacers (500)	WG-100-1002
Infinium XT	Infinium XT Te-Flow Chamber (8)	20012129
	Infinium XT Glass Back Plates (8)	20011756
Universal	Infinium Te-Flow Clips (16)	20011758
	Infinium Te-Flow 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 instrument control computer • 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 instrument control computer • 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 the Illumina 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
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
Te-Flow rack, water re-circulator, tubing, and connections	1	1	1	1	1	1
Te-Flow chambers (HTS/LCG specific)	10	30	–	–	–	–
Te-Flow chamber spacers (HTS/LCG specific)	500	500	–	–	–	–
Te-Flow chambers (HD specific)	–	–	10	30	–	–
Te-Flow chamber spacers (HD specific)	–	–	500	500	–	–
Te-Flow chambers (XT specific)	–	–	–	–	24	48
Te-Flow chamber spacers (XT specific)	–	–	–	–	500	500
Te-Flow 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
Te-Flow chamber dismantling tool	1	2	1	2	2	2
Dissecting forceps	1	1	1	1	1	1

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	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 Te-Flow 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 Asian Screening Array-24	30	6048
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.

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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 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.

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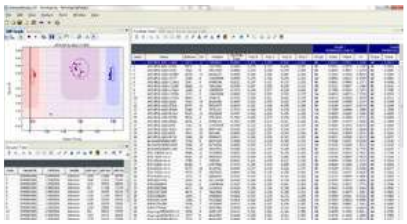

GenomeStudio Genotyping Module

The GenomeStudio Genotyping (GT) Module supports 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 millions of markers
- Estimate LogR 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 CNV 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

GenomeStudio Methylation Module

The GenomeStudio Methylation (M) Module supports 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 users to view CpG island methylation status across the genome with the Illumina Genome Browser and Illumina Chromosome Browser.

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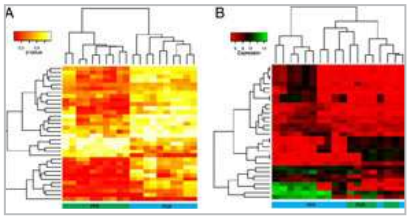
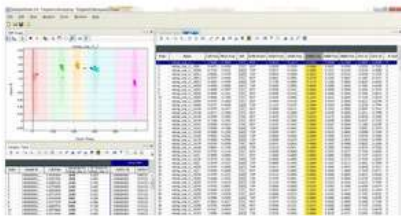


Visualize single-site resolution data line plots, bar graphs, scatter plots, histograms, dendrograms, box plots, or heat maps. The Methylation Module enables users to combine methylation data with gene expression profiling data within the same GenomeStudio project for correlation between levels of methylated 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

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

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

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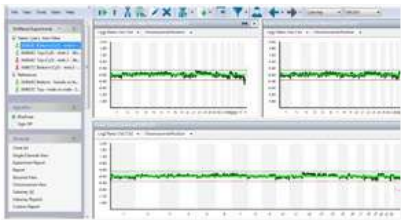
Beeline™ Software

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 interrogate clusters visually increases. Beeline Software addresses this potential bottleneck by enabling prefiltering of large data sets before importing into GenomeStudio Software.

Beeline Software offers flexible filtering capabilities to reduce experimental array data size. The software prefilters large data sets, reducing the overall data volume by eliminating any poorly performing loci (such as those with low signal intensity) and nonrelevant 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 easy integration
- Works directly with Infinium data output and creates projects for direct import into GenomeStudio Software



BlueFuse™ Multi Software

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, CytoChip arrays, and 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

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Instruments

MiniSeq System
Simplicity and accessibility

iSeq 100 System
Accessible targeted sequencing

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
The NextSeq 550Dx is a registered *in vitro* diagnostics NGS instrument in the US, Europe, and other international markets.

FDA-regulated and CE-marked for *In Vitro* Diagnostic (IVD) Use

NovaSeq 6000 System
The next era in sequencing

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Accessible sequencing solutions

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




Product	iSeq 100 System	MiniSeq System	MiSeq System	MiSeqDx System
Description	Most accessible Illumina 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, targeted sequencing, and library QC	Targeted DNA and targeted RNA sequencing	Small-genome, amplicon, and targeted gene panel sequencing	Cystic fibrosis assays Praxis Extended RAS Panel
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	9-19 hrs	4–24 hrs	5–55 hrs	
Max clusters/flow cell[b]	4 M	25 M[c]	25 M[d]	
Max read length	2 × 150 bp	2 × 150 bp	2 × 300 bp	

a. Comparisons are within the Illumina portfolio
b. For the MiniSeq System High-Output Kit only
c. Clusters passing filter
d. For MiSeq Reagent Kits v3 only

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	NextSeq 550 System		NextSeq 550Dx System	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	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	Whole-genome, tumor/normal, transcriptome, exome, and targeted resequencing
	Mid-output	High-output	RUO and IVD modes	SP, S1, S2, S4
	1	1	1	1 or 2
	20–39 Gb	30–120 Gb	20–120 Gb	80–3000 Gb
	15–26 hrs	12–30 hrs	15-30 hrs	13–44 hrs
	130 M	400 M	2.5 B	10 B
	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 x 250 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 studies

Sequencing reagent options	
Flow cell type	i1
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 x 150 bp	Nextera DNA Flex Library Prep Kit
		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 15
		AmpliSeq for Illumina Cancer HotSpot Panel v2
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina BRCA Panel
		AmpliSeq for Illumina Immune Response Panel
Genetic conditions sequencing panel	2 × 150 bp	TruSight Cardio Sequencing Panel
Custom sequencing	2 x 150 bp	AmpliSeq for Illumina Custom Panels
	2 x 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits

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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 or 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 DNA Flex Library Prep Kit
		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
		AmpliSeq for Illumina Cancer HotSpot Panel v2
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina BRCA Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel v3
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel
		AmpliSeq for Illumina Immune Response Panel
	2 x 151 bp	AmpliSeq for Illumina Myeloid Panel
		AmpliSeq for Illumina TCR beta-SR Panel
		AmpliSeq for Illumina Exome Panel
		AmpliSeq for Illumina Transcriptome Human Gene Expression Panel
		AmpliSeq for Illumina Childhood Cancer Panel
Genetic conditions sequencing panels	2 × 150 bp	TruSight Cardio Sequencing Panels
		TruSight One Sequencing Panels
		AmpliSeq for Illumina Community Panels
Custom sequencing	2 x 150 bp	AmpliSeq for Illumina On-Demand Panels
	2 x 150 bp	AmpliSeq for Illumina Custom Panels
	2 x 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
microRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Epigenome		
Protein-DNA interactions	2 x 75 bp	TruSeq Chip Library Prep Kit

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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 DNA Flex Library Prep Kit
		Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
Cancer research sequencing panels	2 × 150 bp	TruSight Cancer Hotspot Sequencing Panel
		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
		AmpliSeq for Illumina Cancer HotSpot Panel v2
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina BRCA Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel v3
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel
		AmpliSeq for Illumina Immune Response Panel
	2 x 151 bp	AmpliSeq for Illumina Myeloid Panel
		AmpliSeq for Illumina TCR beta-SR Panel
		AmpliSeq for Illumina Exome Panel
		AmpliSeq for Illumina Transcriptome Human Gene Expression Panel
		AmpliSeq for Illumina Childhood Cancer Panel
Genetic conditions sequencing panel	2 x 150 bp	TruSight Cardio Sequencing Panels
		TruSight One Sequencing Panels
		AmpliSeq for Illumina Community Panels
Custom sequencing	2 x 150 bp	AmpliSeq for Illumina On-Demand Panels
	2 x 150 bp	AmpliSeq for Illumina Custom Panels
	2 x 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Transcriptome		
Targeted RNA sequencing	1 × 50 bp	TruSeq Targeted RNA Expression Panel Kits
mRNA sequencing	2 x 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Exome Library Prep Kit
microRNA sequencing	1 × 50 bp	TruSeq Small RNA Library Prep Kit
Epigenome		
Protein-DNA interactions	2 x 75 bp	TruSeq ChIP Library Prep Kit

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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 v2.5		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.

FDA-regulated and CE-marked for in *In Vitro* Diagnostic (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 Flex for Enrichment Reagents
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
		Nextera DNA Flex Library Prep Kit
Cancer research sequencing panels	2 x 101 bp	TruSight Oncology 500 Kit
	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 Tumor 170 Sequencing Panel
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina BRCA Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel v3
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel
		AmpliSeq for Illumina Immune Response Panel
	2 x 151 bp	AmpliSeq for Illumina Myeloid Panel
		AmpliSeq for Illumina TCR beta-SR Panel
		AmpliSeq for Illumina Exome Panel
		AmpliSeq for Illumina Transcriptome Human Gene Expression Panel
		AmpliSeq for Illumina Childhood Cancer Panel
Genetic conditions sequencing panel	2 x 150 bp	TruSight Cardio Sequencing Panels
		TruSight One Sequencing Panels
		TruSight One Expanded Sequencing Panel
		AmpliSeq for Illumina Community Panels
Custom Sequencing	2 x 150 bp	AmpliSeq for Illumina On-Demand Panels
	2 x 150 bp	AmpliSeq for Illumina Custom Panels
	2 x 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Exome sequencing	2 x 75 bp	Nextera DNA Exome Kit
	2 x 75 bp	TruSeq DNA Exome Kit
	2 x 100 bp	Nextera Flex for Enrichment Reagents
Transcriptome		
Whole-transcriptome sequencing	2 x 75 bp	TruSeq Stranded Total RNA with Ribo-Zero Library Prep Kits
Targeted RNA sequencing	1 x 50 bp	TruSeq Targeted RNA Expression Panel Kits
mRNA sequencing	2 x 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Exome Library Prep Kit
microRNA sequencing	1 x 50 bp	TruSeq Small RNA Library Prep Kit
Epigenome		
Protein/DNA interactions	2 × 75 bp	TruSeq ChIP 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
Epigenetics	**Supported array**
MethylationEPIC BeadChip Kit	Infinium MethylationEPIC BeadChip Kit

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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.

Flow cell type	SP			S1			S2			S4	
Sequencing reagent options											
Bases (Gb)/flow cell	80	250	400	160	320	480	333	667	1000	3000	
Clusters (M)/flow cell	800	800	800	1600	1600	1600	3333	3333	3333	10,000	
Cycles	100	300	500	100	200	300	100	200	300	300	
Sequencing output per flow cell											
2 x 50 bp	65–80 Gb			134–167 Gb			333–417 Gb			N/A ‡	
2 × 100 bp	N/A ‡			266–333 Gb			667–833 Gb			1600–2000 Gb	
2 × 150 bp	200–250 Gb			400–500 Gb			1000–1250 Gb			2400–3000 Gb	
2 x 250 bp	325-400 Gb			N/A ‡			N/A ‡			N/A ‡	
Clusters per flow cell											
Single-end Reads	650–800 M			1.3–1.6 B			3.3 B–4.1 B			8-10 B	
Paired-end Reads	1.3–1.6 B			2.6–3.2 B			6.6–8.2 B			16–20 B	
Cycles per flow cell											
Cycles	500	300	100	300	200	100	300	200	100	300	200

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 Mate Pair Library Prep Kit
		Nextera Flex for Enrichment Reagents
WGS (small genomes)	2 × 150 bp	Nextera XT DNA Library Prep Kit
		Nextera Mate Pair Library Prep Kit
		Nextera DNA Flex Library Prep Kit
Cancer research sequencing panels	2 x 150 bp	TruSight Cancer Sequencing Panel
		TruSight RNA Pan-Cancer Sequencing Panel
		TruSight Tumor 170 Sequencing Panel
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina BRCA Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel
		AmpliSeq for Illumina Comprehensive Cancer Panel v3
		AmpliSeq for Illumina Focus Panel
		AmpliSeq for Illumina Immune Repertoire Plus, TCR beta Panel
		AmpliSeq for Illumina Immune Response Panel
	2 x 151 bp	AmpliSeq for Illumina Myeloid Panel
		AmpliSeq for Illumina TCR beta-SR Panel
		AmpliSeq for Illumina Exome Panel
		AmpliSeq for Illumina Transcriptome Human Gene Expression Panel
		AmpliSeq for Illumina Childhood Cancer Panel
	2 x 75 bp	TruSight RNA Fusion
Genetic conditions sequencing panels	2 × 150 bp	TruSight Cardio Sequencing Panels
		TruSight One Sequencing Panels
		TruSight One Expanded Sequencing Panel
		AmpliSeq for Illumina Community Panels
Metagenomics	2 × 250 bp	TruSeq DNA PCR-Free Library Prep Kit
		Nextera DNA Flex Library Prep Kit
Custom sequencing	2 x 150 bp	AmpliSeq for Illumina On-Demand Panels
	2 x 150 bp	AmpliSeq for Illumina Custom Panels
	2 x 75 bp	Nextera Rapid Capture Custom Enrichment Kit
Exome sequencing	2 x 75 bp	Nextera DNA Exome Kit
	2 x 75 bp	TruSeq DNA Exome Kit
	2 x 100 bp	Nextera Flex for Enrichment Reagents
Transcriptome		
Whole-transcriptome sequencing	2 x 75 bp	TruSeq Stranded Total RNA with Ribo-Zero Library Prep Kits
Targeted RNA sequencing	1 x 50 bp	TruSeq Targeted RNA Expression Panel Kits
mRNA sequencing	2 × 75 bp	TruSeq Stranded mRNA Library Prep Kit
		TruSeq RNA Access Library Prep Kit
microRNA sequencing	1 x 50 bp	TruSeq Small RNA Library Prep Kit
Epigenome		
Protein-DNA interactions	2 x 75 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
AmpliSeq for Illumina		✓	✓	●	✓	
Nextera Flex for Enrichment			●	✓	●	●
Nextera DNA Flex	●	✓	✓	✓		●
Nextera XT DNA	●	✓	✓	✓	✓	✓
TruSight Tumor 15		✓	✓		✓	
TruSight Tumor 170		✓		✓		
TruSeq DNA PCR-Free	●	✓	✓	●	✓	✓
TruSeq Nano DNA	●	✓	✓	✓	✓	✓
TruSeq RNA Exome	●	✓	✓	✓	✓	✓
TruSeq Stranded mRNA	●	✓	✓	✓	✓	✓
TruSeq Stranded Total RNA	●	✓	✓	✓	✓	✓
16S rRNA			●	✓		

✓ 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/automation for the most up-to-date information on available methods. Qualified methods not available for all partner systems, please check website for specific partner system information. 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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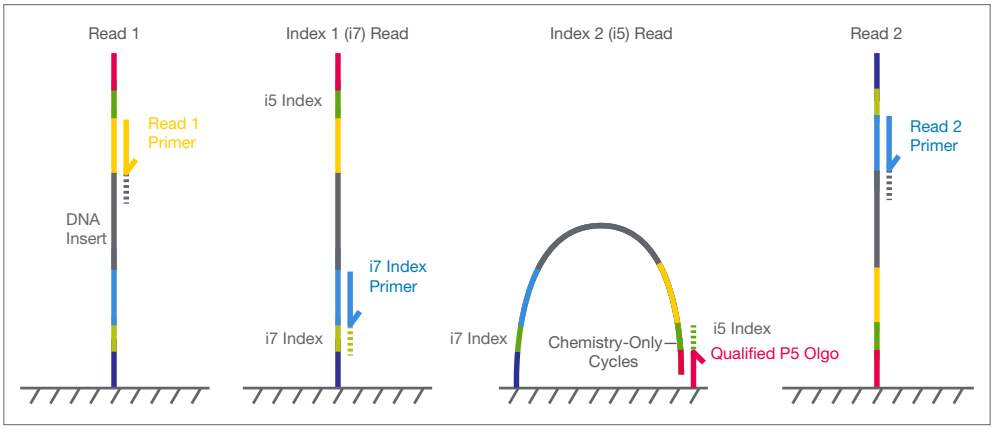
Indexing technology quick reference

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 indexed 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, meaning that multiple indexed samples can be loaded into a sequencing run.



Dual Indexed Sequencing on a Paired-End Flow Cell

1. **Read 1**—Read 1 follows the standard Read 1 sequencing protocol using SBS reagents. The Read 1 sequencing primer is annealed to the template strand during the cluster generation step.
2. **Index Read preparation**—The Read 1 product is removed and the Index 1 (i7) sequencing primer is annealed to the same template strand.
3. **Index 1 (i7) Read**—Following Index Read preparation, the Index 1 (i7) Read performs up to 20 cycles of sequencing. NOTE: The number of cycles in each Index Read depends on the system and run parameters.
4. **Index 2 (i5) Read**—The Index 1 (i7) Read product is removed and the template anneals to the grafted P5 primer on the surface of the flow cell. The run proceeds through an additional seven chemistry-only cycles (no imaging occurs), followed by up to 20 cycles of sequencing.
5. **Read 2 resynthesis**—The Index Read product is removed and the original template strand is used to regenerate the complementary strand. The original template strand is then removed to allow hybridization of the Read 2 sequencing primer.
6. **Read 2**—Read 2 follows the standard paired-end sequencing protocol using SBS reagents.

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 Nextera DNA UD Indexes	Human WGS, amplicons, plasmids, and microbial species	96 (Set A)	20027213
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 (Set A) 96 (Set B) 96 (Set C) 96 (Set D) 96 (Set A-D)	20019105 20019106 20019107 20019167 20031676
AmpliSeq for Illumina UD Indexes	Use with AmpliSeq for Illumina Panels	24	20019104
TruSeq Targeted RNA Indexes	Using targeted RNA panels	96 of 384	15033977

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Analysis tools

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Research informatics

Gain insights from a growing body of complex data with informatics tools that can help manage, analyze, and interpret the data

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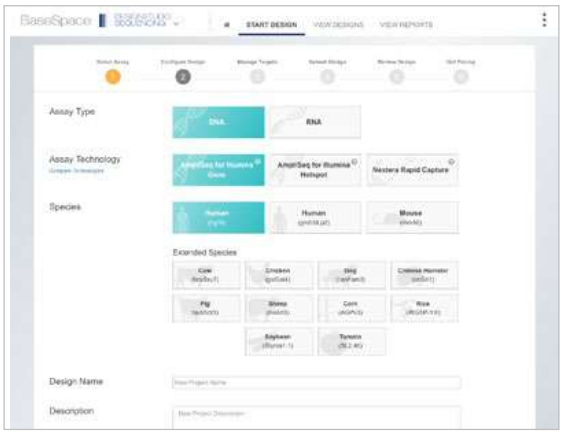
Sequence assay design

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
- 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 genomic, 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. These tools 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.

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Sample tracking and experiment design

Set up and run sequencing experiments easily

BaseSpace Sequence Hub

Users can plan entire sequencing workflows for the MiniSeq System and NextSeq 550 System with BaseSpace Sequencing Hub Prep tab. 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 starting the run and saving time
- Add libraries and pools to planned runs with just a couple clicks
- Set up and track libraries, pools, and flow cells, simplifying complex library preparation
- 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 minimize or eliminate errors in setting up sample sheets
- Flexible, easy, streamlined management of sample sheets and plate layouts
- Desktop support for sample sheet setup for the MiSeq System, NextSeq 550 System, and NovaSeq 6000 System

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Data analysis and management

Spend more time doing research and less time setting up software infrastructure

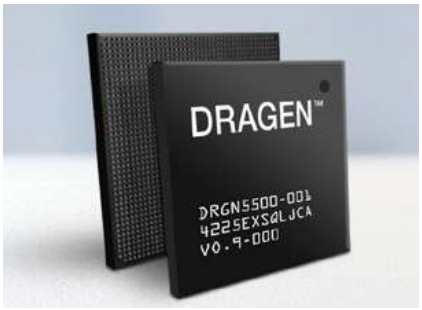
BaseSpace Sequence Hub

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, real-time 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 analyze data generated by common methods such as RNA-Seq, MethylSeq, exome, enrichment, amplicon, WGS, *de novo* assembly, metagenomics, and more.

BaseSpace Sequence Hub apps

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



DRAGEN Bio-IT Platform

The Illumina DRAGEN (Dynamic Read Analysis for GENomics) Bio-IT Platform provides ultra-rapid secondary analysis of NGS data. The DRAGEN platform uses highly reconfigurable field-programmable gate array technology (FPGA) to provide hardware-accelerated implementations of genome analysis algorithms, such as BCL conversion, mapping, alignment, sorting, duplicate marking, and haplotype variant calling. Fundamental features of the DRAGEN platform address common challenges in genomic analysis, such as lengthy compute times and massive volumes of data. Without compromising accuracy, the DRAGEN platform delivers quickness, flexibility, and cost efficiency. The reprogrammable nature of the DRAGEN platform enables Illumina to develop custom algorithms and allows for improvements to accommodate future applications.

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Biological data interpretation

Turn genomic information into biological insight

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

Scientists can quickly extract biological meaning from their, or public, genomic data through the user-friendly, intuitive BaseSpace Correlation Engine, originally developed under the NextBio Research Platform. Illumina has spent over a decade normalizing the raw data from whole-genome studies across platforms and data types and ingesting into BaseSpace Correlation Engine to create the world's largest continuously growing curated database. BaseSpace Correlation Engine routinely identifies thousands of studies by correlation analysis and multivariate testing of harmonized data (140,000 genomic signatures from 24,000 studies as of March 2019) for 1000s of genes that have few or no results in PubMed.

- Hypothesis-free data exploration with scientific tools such as Body Atlas, Disease Atlas, Pharmaco Atlas, Knockdown Atlas, Genetic Markers, Pathway enrichment, and Meta-analysis
- Context-rich 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

For investigators specializing in the analysis of large cohorts of individual subjects, learn about BaseSpace Cohort Analyzer in the Translational Informatics section on page 145.

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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 analyze complex genomic data in novel ways. These bioinformatics tools turn data into insights. 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 is a platform for analyzing complex human data for translational research applications, hosting more than 11,500 records with over 1000 unique phenotypic attributes. These records include public subjects from high-profile studies like the Cancer Genome Atlas (TCGA) and NCI TARGET studies. 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 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 enables researchers to view their data across a broad spectrum of data types and platforms in the context of nearly 22,000 public studies.

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Pharma and biotech analysis

Powerful analytical platforms 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 data analysis for preclinical and clinical trials. These 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 regarding 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 therapeutic areas, including cancers and autoimmune diseases. BaseSpace Correlation Engine allows users to securely integrate their private data with the more than 22,000 public studies. Daily updates to the public studies ensures a robust and growing repository of study signatures, providing data-driven answers for future discoveries.

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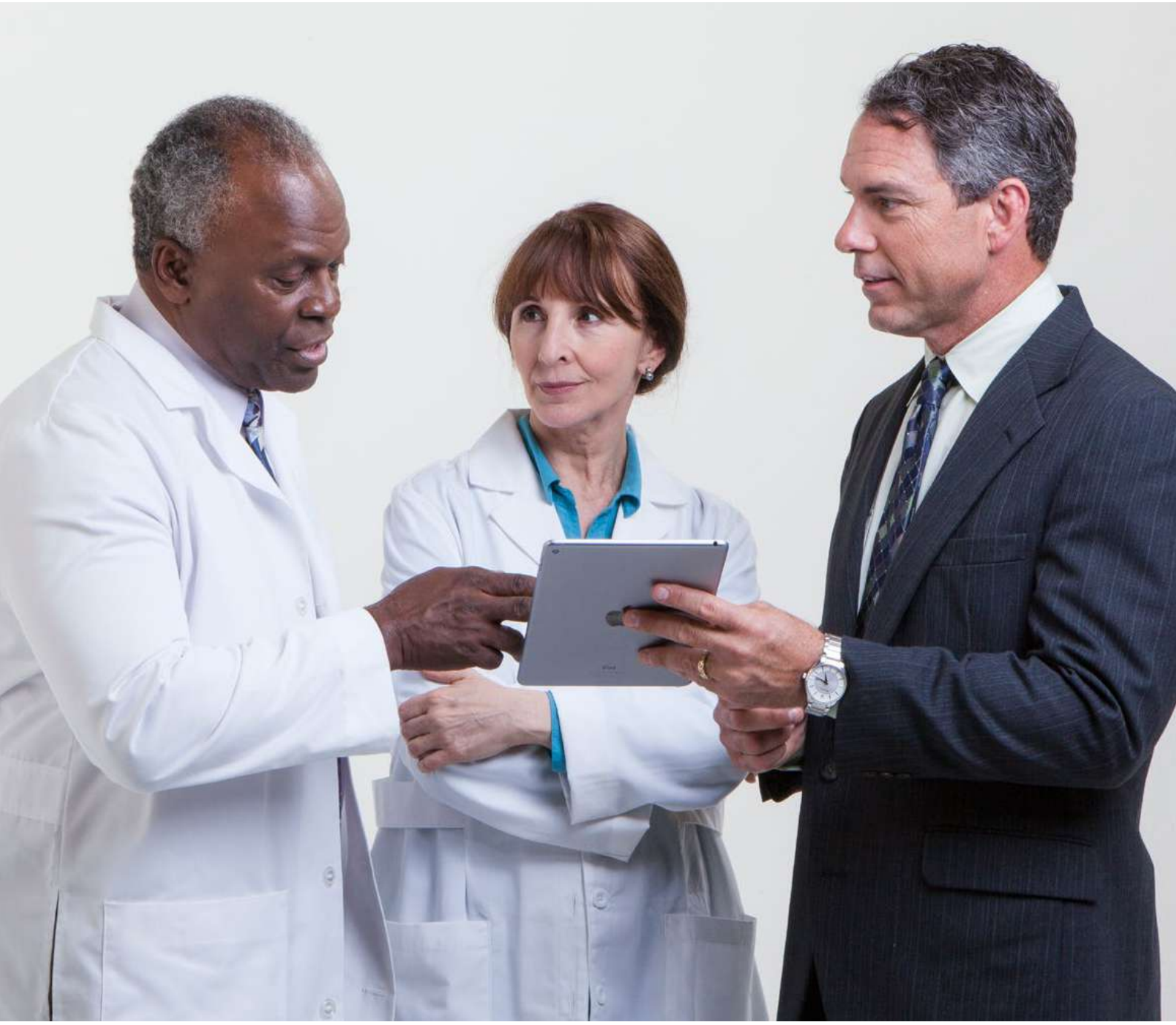
Services



Instrument services, training, and consulting

We provide solutions so you can focus on the big discoveries

Illumina offers integrated service solutions that combine product support with opportunities for training and consulting to help boost your lab's efficiency and productivity, and optimize your workflow. With our flexible offerings, 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 instrument purchase, Illumina offers instrument maintenance, repair, and qualification solutions. From basic to 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 offer 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

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For all 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.

Bronze

Bronze is our most affordable repair-only service plan that protects against unpredictable costly failures. It is designed to maintain instrument performance on a limited budget.

Silver

Silver is our most popular full-service plan. It is designed to maximize performance while minimizing downtime and assisting regulatory compliance with routine documented preventive maintenance. It is a premium full-service plan, balancing performance, productivity, and cost. This plan is best suited for a wide range of research, translation, and applied genomics customers with medium to high sample volume, small to medium fleet size, and requiring high instrument availability with redundant back-up capacity.

Gold

Gold is our most extensive plan. It is designed to maximize performance and uptime for mission-critical systems with fast sample-to-data turnaround needs. This plan is best suited for laboratories in an environment requiring the fastest on-site response plus peace of mind in adherence to stringent guidelines.

Dedicated On-Site Plan

Dedicated On-Site is our highest service level plan with a dedicated full-time on-site Field Service Engineer and immediate response time. It is designed to enable the highest productivity, uptime, and peak performance for sites with a large fleet of instruments. This plan is best suited for large production laboratories with ten or more high-throughput instruments per site requiring the highest level of service and lowest cost of operation.

Illumina Product Support Service Plan comparison

	BRONZE	SILVER	GOLD	DEDICATED ONSITE
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 preventive maintenances for HiSeq X and NovaSeq 6000 System services, and 1 preventive maintenance for all other instruments
d. Operational Qualification at preventive maintenance and qualified repair visits

Visit https://emea.illumina.com/services/instrument-services-training/product-support-services.html 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 are 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 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 Oncology 500 DNA	20031667
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
MiniSeq System Training–Customer Site	20005637

Visit https://emea.illumina.com/services/instrument-services-training/product-support-services.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 NGS or BeadChip (microarray) technology, scientists frequently navigate through many decisions regarding choice of instrumentation, applications, throughput requirements, and time from DNA to analysis.

Workflow Design and Evaluation Service

Thinking of purchasing an Illumina NGS instrument, microarray, or library prep solution? Try the Workflow Design and Evaluation Service with your own samples or libraries on a sequencing instrument at the Illumina Application Lab.

Step 1 — Pre-Library Prep and/or Sequencing Consultation
- Determine the appropriate Illumina NGS/BeadChip solution
- Use Illumina Concierge for custom designs

Step 2 — Library Preparation
- Prepare libraries from customer samples
- Develop a customized DNA-to-data user guide
- Generate sample and library QC data

Step 3 — Cluster Generation, Sequencing, and Data Analysis
- Generate full sequence data set
- Analyze data with BaseSpace Sequence Hub or Illumina genotyping software

Step 4 — Post-Library Prep and/or Sequencing Consultation
- Review personalized sample-to-data workflow
- Review sample data with Illumina team

Illumina Concierge Services

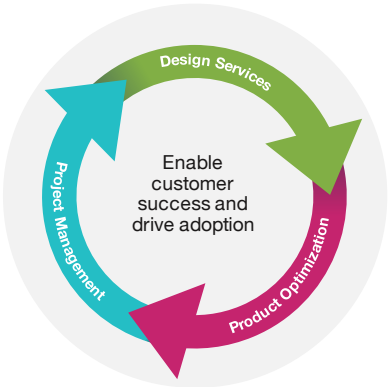
Concierge Services provides design and product optimization services for custom array and sequencing products. Our scientists provide customers with innovative design features to maximize performance.

Concierge Design Services

Concierge Design Services cover the creation and optimization of a custom targeted design using design strategies that can be aimed to improve *in silico* coverage or can provide customization of design configurations beyond design parameters and species supported through DesignStudio Software. The customer will work with a dedicated scientist from the Concierge team to discuss and implement design requirements to create the custom design.

Concierge Wetlab Testing and Optimization Services

Concierge Wetlab Testing and Optimization Services offer wetlab testing and oligo pool enhancement aimed to improve coverage uniformity of custom targeted DNA sequencing panels. A dedicated Concierge project manager will be assigned as the point of contact to manage testing, oligo synthesis, and communication with the customer. This service is offered for new and existing (reordered) custom panels, and applies to panels targeting human DNA or nonhuman species.



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, delivered by experts with deep scientific and product knowledge, 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

Consideration	Concierge Custom Design Service Workflow Design and Evaluation Genomics IT Consulting
Installation	Installation Qualification (IQ) Operational Qualification (OQ) Workflow Training Geonomics IT Consulting
Optimization	Gene Curator Operational Qualification (OQ) Performance Qualification (PQ) Workflow Training Bioinformatics Consulting Geonomics IT Consulting
Data Analysis	Bioinformatics Consulting Geonomics IT Consulting Custom App Custom Informatic

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.

Initial Consultation	Assess and review current lab genomic IT infrastructure needs
Design Recommendation	Provide detailed hardware recommendations and integrated architecture designs
Procurement	Customer obtains hardware and software from Illumina-qualified vendor or vendor of their choice
Implementation	Test and confirm successful installation of all software and hardware
Support	Provide support for a broad range of NGS needs from initial instrument installation to final data analysis

Illumina Genomics IT Consulting Services

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Clinical and Lab Consulting Services

Clinical and Laboratory Consulting Services provide support to customers when implementing Illumina sequencing and array technologies. Analytical evaluation services facilitate adoption in regulated laboratories, and consulting services for sequencing and array customers facilitate adoption in a high-throughput testing environment.

Illumina Sequencing Consulting Service

Illumina Sequencing Consulting Services provide customized support to customers when implementing Illumina sequencing technologies to facilitate adoption in a high-throughput testing environment.

Illumina ArrayLab Consulting Service

Illumina ArrayLab Consulting Services provide customized support to customers when implementing Illumina array technologies to facilitate adoption in a high-throughput testing environment.

TruSight Oncology 500 Analytical Evaluation Service

TruSight Oncology 500 Analytical Evaluation Service is a set of tools intended to guide customers to align with the latest CAP/AMP/European standards and industry best practices for the analytical evaluation of the TruSight 500 Panel. The tools provided with this service enable customers to perform an analytical evaluation of TruSight 500 Panels quickly, potentially reducing implementation time in laboratories down to 2-4 months.

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

Application index

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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.

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

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

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