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Simplifying NGS for greater accessibility

Scientists across the world are adopting next-generation sequencing (NGS) methods to gain a deeper understanding of biological systems. NGS is a flexible tool that helps answer multiple experimental questions with sequencing readouts. Compared to conventional technologies, NGS offers increased scale and sensitivity, providing more comprehensive results to help address many complex genomic questions. With the ability to interrogate large and small genomes, gene expression, chromatin accessibility, methylation, and more, NGS is a versatile discovery engine.

As NGS systems become more powerful to assist genomic discovery, they are also becoming easier to use and more cost-efficient. Over the last 20 years, the cost of NGS has rapidly decreased and workflows have become streamlined, leading to an increase in usage.^{1,2} Data analysis of large NGS data sets has also become increasingly user-friendly, allowing for automated data interpretation without prior knowledge of bioinformatics.3

Benchtop sequencing systems offer tunable sequencing capabilities with rapid turnaround times, making NGS more accessible to a wider range of users. Illumina benchtop systems combine operational simplicity with high performance, integrating the latest developments in chemistry and data analysis to enhance sequencing capacity and speed.

With output that is scalable from millions of bases (megabases or Mb) to billions of bases (gigabases or Gb), low- and mid-throughput benchtop systems offer flexible sequencing breadth to support a wide range of applications. Low-throughput benchtop systems are best suited for small whole-genome sequencing, microbial metagenomics, and targeted gene sequencing applications or for library quality control (QC) before large-scale studies. Labs focused on data-rich methods like whole-exome and single-cell sequencing should consider a mid-throughput benchtop system. This eBook highlights some of the most popular methods and applications for benchtop sequencing systems, from microbial genomics to multiomics.



Learn more

Choosing a benchtop sequencing system New to NGS? Learn the basics

Illumina benchtop systems and example applications





System	MiSeq i100 Series	NextSeq 1000 and NextSeq 2000 Systems				
System type	Low-throughput benchtop systems	Mid-throughput benchtop systems				
Output	1.5 Gb to 30 Gb	10 Gb to 540 Gb				
Reads per run	5 million to 100 million	100 million to 1.8 billion				
Advantages	Fast workflow for targeted samples or shotgun sequencing Well suited for pilot studies and library QC before larger projects	Flexibility for a wide range of emerging applications like spatial biology and single-cell sequencing Broader output range while maintaining a benchtop footprint				
Large whole-genome sequencing		• •				
Small whole-genome sequencing	•••	•••				
Exome and large panel sequencing		•••				
Targeted gene sequencing	•••	•••				
Single-cell profiling		•••				
Transcriptome sequencing	••	•••				
Targeted gene expression profiling	•••	• •				
miRNA and small RNA analysis	•••	•••				
Chromatin analysis		•••				
Methylation sequencing		• •				
16S metagenomic sequencing	•••	•••				
Shotgun metagenomics	•••	• •				
Metagenomic profiling		•••				
Cell-free sequencing		••				
Library QC	•••	••				

Key applicationSupported application

Fast, culture-free microbial analysis

Microbial genomics provide insights into the impact of microbes on human health and environmental processes. NGS is established as an important tool for analyzing small genomes, including bacteria, viruses, and other microbes. For microbiologists, NGS facilitates the discovery of novel microbes and the characterization of difficult-to-culture organisms. Microbiome sequencing also enables the creation of accurate reference genomes essential for microbial identification and comparative genomic studies across diverse applications. This technology is further instrumental in investigating outbreaks of infectious diseases, tracking antibiotic resistance, and identifying foodborne pathogens.

Microbiome sequencing with benchtop systems

Benchtop sequencing systems are well suited for studying microbial genomics, enhancing the accessibility and depth of microbiome research. Using cost-effective and compact NGS systems, researchers can gain comprehensive insights into microbial communities that surpass traditional culture-based methods. Sequencing reagent kits that can generate 300-base pair (bp) paired-end reads are especially useful for microbial sequencing methods, enabling de novo genome assembly and metagenomic detection of bacterial species.

Popular microbial sequencing methods

Small whole-genome sequencing

NGS-based sequencing of small genomes (≤ 5 Mb in size) allows researchers to sequence hundreds of organisms simultaneously with a simple workflow. NGS can identify low-frequency variants and genome rearrangements that may be missed by other methods.4



Learn more

Microbial whole-genome sequencing

Methods guide: Microbial single-genome sequencing

Application note: Small whole-genome sequencing on NextSeq 1000 and NextSeq 2000 Systems

Application note: Microbial whole-genome sequencing with Illumina DNA PCR-Free Prep, Tagmentation



Small whole-genome sequencing for food safety research

NGS is a key tool for food testing in public health. Two outbreaks of Salmonella food poisoning occurred concurrently in one district of South Africa, one at a daycare center and one at a restaurant. Researchers used small whole-genome sequencing of stool samples and food samples from both outbreaks to find epidemiological links between the two cases. The strains found in the samples from the daycare center and the samples from the restaurant were highly related with fewer than five allele differences between them. The close relationship of the Salmonella strains from this investigation indicated a common contaminated food source, likely eggs.5

16S ribosomal RNA (rRNA) sequencing

The prokaryotic 16S rRNA gene, approximately 1500 bp in length, contains nine variable regions amid conserved sequences. The sequences of key variable regions can serve as effective markers for phylogenetic classification, allowing assessment of microbial diversity and abundance within complex communities.6,7



Learn more

Demonstrated protocol: 16s metagenomic sequencing library preparation

16s RNA sequencing

Methods guide: 16S ribosomal sequencing

Application note: 16S rRNA sequencing on NextSeq 1000 and NextSeq 2000 Systems

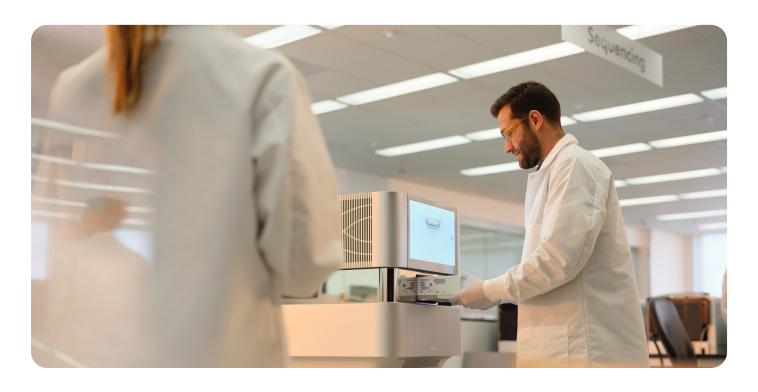
Application note: 16S metagenomics studies with the

MiSeq System



16S rRNA sequencing for human microbiome research

The human microbiome has a large impact on oral health and can inform our understanding of disease mechanisms. Researchers in South Korea used 16S rRNA sequencing to characterize the microbiome in subjects with dental implant infections, which increases the risk for additional bone loss. In comparison to the microbiome of subjects with healthy gums or subjects with severe gum disease, bacterial communities associated with dental implant infections showed higher diversity. Data analysis identified both well-known pathogens associated with gum disease and several bacteria previously unrecognized in gum tissue. Results indicated that dental implant infections cause a more complex inflammatory response that involves more bacteria compared to severe gum disease.8



Shotgun metagenomic sequencing

Shotgun metagenomics involves comprehensive sequencing and identification of all microbes in a complex sample. Sequencing microbial communities helps researchers evaluate bacterial diversity and detect the abundance of microbes in various environments.



Metagenomic sequencing for infectious disease research

As a double-stranded DNA virus with proofreading capabilities, the monkeypox virus (MPXV) generally evolves more slowly when compared to SARS-CoV-2 and other RNA viruses. MPXV is endemic in parts of Africa and typically spreads from animals to humans. However, in 2022, a strain of MPXV, adapted for human-to-human transmission caused several hundred infections across the globe. Researchers in Portugal used shotgun metagenomic sequencing to track the phylogenetic origins of this circulating MPXV strain. The 2022 strain was related to cases from 2018 but had accumulated dozens of mutations at an accelerated rate. Sequencing data showed base changes associated with a family of human virus-fighting enzymes called APOBEC3. These enzymes induce mutations in viral DNA to kill off those viruses, but in this case, they just accelerated the evolution of this MPXV strain to be more virulent.9





Learn more

Shotgun metagenomic sequencing

Application note: A shotgun metagenomics NGS workflow for assessing microbial populations in complex samples

Affordable, targeted sequencing of key genes

Targeted DNA sequencing examines specific genes or genomic regions, enabling researchers to focus studies on the exome or selected genes to accelerate turnaround times and enable deep sequencing for rare variant detection. This approach is more cost-effective than whole-genome sequencing for investigating specific areas of interest, enabling detection of a wide range of genomic alterations, including single nucleotide variants, insertions, deletions, copy number changes, and chromosomal aberrations. Targeted DNA sequencing allows for scalable studies with the ability to sequence multiple samples simultaneously.

Targeted DNA sequencing with benchtop systems

The easy-to-use design and small footprint of benchtop sequencing systems help bring targeted DNA sequencing capabilities to more laboratories, even those in unique locations. For example, the Minderoo Foundation in Australia uses a NextSeq 2000 System on board their marine research vessel. Scientists are collecting environmental DNA from seawater and producing high-quality genomic data in mere hours, revealing which species are present and how climate change is impacting that region. Mid-throughput benchtop systems also allow clinical research labs to bring targeted assays like comprehensive genomic profiling in-house. The search labs to bring targeted assays like comprehensive genomic profiling in-house.



Popular targeted DNA sequencing methods

Amplicon-based targeted DNA sequencing

Deep sequencing of PCR products (amplicons) enables efficient identification and characterization of genetic variants. This approach uses oligonucleotide probes to amplify specific regions of interest, followed by NGS. Multiplexing of hundreds to thousands of amplicons per reaction enables comprehensive coverage, particularly for challenging sequences such as GC-rich regions.

Enrichment-based targeted DNA sequencing

Targeted DNA sequencing through enrichment involves capturing specific genomic regions using biotinylated probes designed for hybridization, followed by magnetic pulldown isolation and NGS. This method is effective for sequencing exomes or large gene sets (> 50 genes) with robust and straightforward workflows.

Exome sequencing

Whole-exome sequencing uses enrichment to focus on the ~2% of the genome that codes for proteins for efficient identification of coding variants across a broad range of applications, including population genetics, genetic disease, and cancer studies. With an accessible combination of turnaround time and price, exome sequencing is a cost-effective alternative to whole-genome sequencing.



Learn more

Amplicon sequencing

Target enrichment

Exome sequencing

Methods guide: Small oncology panels

Application note: NextSeq 1000 and NextSeq 2000 exome

sequencing solution





Exome sequencing for genetic disease research

Endometriosis is a chronic inflammatory disease that shows high heritability, but only 25% of cases are explained by common variants. Researchers in Türkiye performed whole-exome sequencing on three women in the same family with endometriosis. They identified three novel rare variants likely relevant to disease pathogenesis: *TNFRSF1B*, *GEN1*, and *CRABP1*. The *TNFRSF1B* gene codes for a TNF receptor that promotes angiogenesis in the endometrium and regulates apoptosis. The *GEN1* and *CRABP1* genes are tumor suppressors and have been associated with endometrial cancer.¹⁴

Unbiased, comprehensive gene expression analysis

RNA sequencing (RNA-Seq) stands as a powerful tool in modern biology, offering highly sensitive and accurate analysis of gene expression across the transcriptome. Unlike traditional methods that focus on selected transcripts, RNA-Seq provides a comprehensive view of cellular RNA, revealing previously undetectable changes in gene expression and enabling the characterization of diverse noncoding RNA forms. This unbiased approach allows researchers to explore transcriptome architecture in depth, identifying transcript isoforms, gene fusions, single nucleotide variants, and other features crucial for understanding cancer mechanisms and genetic diseases.

RNA-Seg with benchtop systems

Transcriptomics is an accessible entry point for using NGS, given its versatility across use cases and well-established workflows. 15,16 The flexible output of benchtop systems works for multiple RNA analysis methods, from targeted transcripts to whole transcriptomes. Shorter read lengths (eg, 1 × 50 bp) support counting applications like gene expression analysis, while longer read lengths (eg, 2 × 300 bp) enable immune repertoire sequencing.

Popular RNA-Seg methods

Total RNA-Seq

Ribosomal RNA (rRNA) can account for 80% of transcripts. Depletion-based total RNA library preparations remove rRNA transcripts that are not informative and allow for comprehensive transcriptome analysis, covering the coding and noncoding RNA landscape. Total RNA-Seq enables accurate measurement of gene and transcript abundance, and detection of known and novel coding features and multiple forms of noncoding RNA.

mRNA-Seq

PolyA capture-based mRNA library preparations pull down mRNA transcripts with a 3' poly A tail. mRNA-Seq offers sensitive, accurate measurement of gene expression and can identify known and novel isoforms in the coding transcriptome, detect gene fusions, and measure allele-specific expression. Because an intact polyA tail is needed, it is not ideal for degraded samples.

Enrichment-based RNA-Seq

Hybridization-capture RNA library preparations use a probebased approach to target transcripts of interest and enable sensitive detection of splicing events and fusions, even in degraded samples. This method enables cost-effective RNA exome analysis using sequence-specific capture of the coding regions of the transcriptome.



Learn more

Total RNA sequencing mRNA sequencing

RNA exome capture sequencing

Methods guide: RNA sequencing

Application note: NextSeq 1000 and NextSeq 2000 RNA sequencing solution



Enrichment-based RNA-Seq for cancer research

Researchers in the Czech Republic employed targeted RNA-Seq alongside germline DNA sequencing to assess the functional impact of DNA variants on hereditary cancer risk, focusing on a panel of 226 cancer-related genes. This approach facilitated the evaluation of clinically relevant transcripts in easily accessible tissues like blood or nasal swabs. RNA capture-based NGS enabled the detection of low-expression genes and splice variants. The integration of RNA analysis with DNA sequencing proved instrumental in characterizing variants of uncertain significance (VUS), such as the G > A variant in the CHEK2 gene, which was shown to cause aberrant splicing and was reclassified as pathogenic.¹⁷



Immune repertoire sequencing

During an immune response, the repertoire of circulating antigen receptors shifts from a diverse pool to one that is dominated by one or a few expanded clones. Immune repertoire profiling sequences RNA transcripts for antigen receptors to identify unique receptor variants and characterize the adaptive immune response.



Learn more

Immunogenomics

Application note: Full-length V(D)J IR-Seq on the NextSeq 1000 and NextSeq 2000 Systems

Accessible, data-rich epigenetic insights

Genetics alone is not enough to explain complex disease and development. External influences such as environmental conditions, diet, and physical activity can induce epigenetic changes that alter how, where, and when specific genes are expressed.¹⁸ Researchers can use epigenetic NGS methods to quantify and analyze the DNA modifications that help direct cell differentiation or disease progression. Methylation sequencing and chromatin accessibility assays leverage the high quality and sensitivity of NGS to reveal the dynamic epigenomic landscape rapidly and thoroughly.

Epigenetic sequencing with benchtop systems

Mid-throughput benchtop systems provide access to epigenetic sequencing applications, giving researchers more tools to study the roles of chromatin structure and DNA methylation in development and disease. Because of the flexible and scalable output of a mid-throughput benchtop system, laboratories no longer need a production-scale NGS instrument to perform these data-intensive methods.

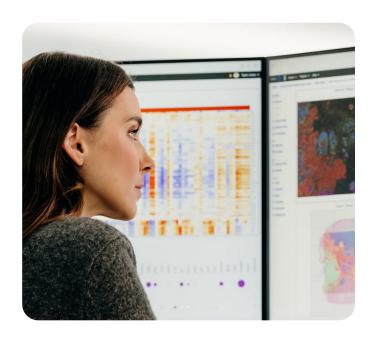
Popular epigenetic sequencing methods

ATAC-Seq for chromatin accessibility

Assay for transposase-accessible chromatin with sequencing (ATAC-Seq) is a rapid, sensitive method for mapping chromatin accessibility throughout the genome. This technique uses a hyperactive transposase enzyme to insert sequencing adapters into exposed DNA regions and generate sequencing libraries that represent open chromatin. ATAC-Seg provides insights into how chromatin packaging impacts gene expression, without prior knowledge of regulatory elements. ATAC-Seq can be used for bulk cell populations or at the single-nucleus level to study heterogeneous cell populations.

Methylation sequencing

Methylation sequencing reveals the methylation status of cytosine-quanine dinucleotides (CpGs) across the genome at a single-nucleotide level. Bisulfite treatment converts cytosine bases to uracil, while methylated cytosines are left unmodified. This method can be performed at the wholegenome level or with targeted methods to enrich for CpG sites or focus on regions of interest.





Learn more

ATAC sequencing chromatin accessibility

Methylation sequencing

Application note: Unify single-cell gene expression and chromatin accessibility

High-resolution, multiomic exploration

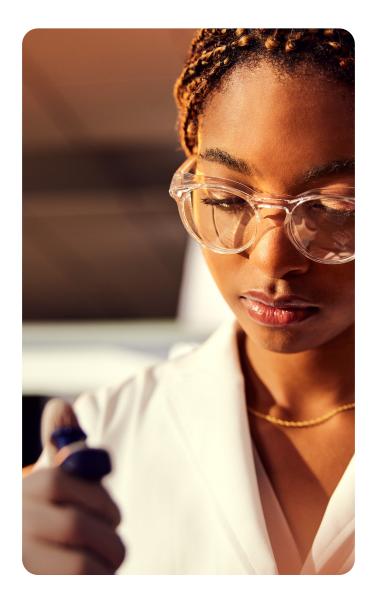
Multiomics provides an integrated approach to power discovery across multiple levels of biology. By combining data from genomics, transcriptomics, epigenetics, and proteomics studies, researchers can achieve a more comprehensive understanding of the molecular changes that contribute to normal development, cellular response, and disease. Integrating these complementary metrics into multiomic data sets brings a fuller picture of cellular phenotypes and helps pull more high-quality information from each sample.

High-resolution methods like single-cell sequencing and spatial sequencing enable deeper insights into complex tissues. Typical NGS methods look at dissociated samples in bulk, masking cellular heterogeneity and losing key spatial information. Profiling gene expression at the single-cell level or with preserved spatial context increases discovery power. Single-cell and spatial sequencing are also compatible with ATAC-Seq, proteomics, and other NGS methods. Multimodal approaches are developed that measure both gene expression and protein levels, or both gene expression and chromatin accessibility, in the same cells.19,20

Multiomic sequencing with benchtop systems

Multiomic insights are more accessible with the power to perform data-intensive applications on your benchtop. With broad application flexibility and output of up to 540 Gb, a mid-throughput benchtop system can be an ideal platform for multiomics. For example, researchers can pair RNA-Seq with exome sequencing to assess whether coding variants impact transcript expression. The capabilities of mid-throughput systems also enable highresolution methods with access to:

- More reads per cell to capture information about lower abundance transcripts
- More cells and samples to empower experimental designs within a given research budget
- Additional conditions, time points, or methods to investigate more complex facets of biology



Popular multiomic sequencing methods

Single-cell RNA-Seq

Single-cell RNA-Seq (scRNA-Seq) can characterize gene expression in hundreds to millions of individual cells from a tissue. This method reveals cellular heterogeneity and provides a more comprehensive understanding of heterogenous cell populations. Using scRNA-Seg facilitates the identification of novel biomarkers and rare cell types that would otherwise be missed with bulk RNA-Seq.^{21,22} Significant advances in single-cell characterization include technologies for cell isolation and new methods and applications for single-cell sequencing. These advances have stimulated the launch of accessible commercial solutions for every step of the single-cell sequencing workflow, from tissue preparation through data analysis.



Learn more

Single-cell RNA sequencing

Application note: NextSeq 1000 and NextSeq 2000 single-cell RNA sequencing solution

Application note: Explore the transcriptome with single-cell resolution

Application note: Unify single-cell gene expression

and chromatin accessibility

Application note: Multiomic interrogation of the immune system at single-cell resolution



Single-cell RNA-Seq for immuno-oncology research

Checkpoint inhibitors show poorer efficacy for cancer therapy in males. Researchers in Oregon conducted scRNA-Seg on metastatic tumor samples from men with metastatic castration-resistant prostate cancer, focusing specifically on CD8 T-cells, which are crucial for checkpoint inhibitor response. The study showed that androgen deprivation slows cancer progression and enhances T-cell activity against tumors. They found distinct CD8 T-cell states associated with response and resistance to PD-1 blockade, revealing that androgen receptor (AR) downregulation in CD8 T-cells correlated with enhanced function. The findings suggest that AR inhibition, by sensitizing hosts to checkpoint blockade, improves anti-tumor immunity and highlights a novel mechanism underlying immunotherapy resistance in prostate cancer.²³

Spatial RNA-Seq

Spatial transcriptomics combines high-throughput imaging and sequencing technologies to show mRNA expression at the cellular level in structurally preserved tissues. Spatial RNA-Seq retains the precise location of biological molecules in morphological context and can help reveal how cells are influenced by neighboring cells, local signaling events, cell-cell interactions, and more. Revealing a tissue's complex mixture of cell types with spatial techniques has already enabled profound new discoveries within the fields of neuroscience, developmental biology, cancer, and more.24



Learn more

Spatial transcriptomics

Application note: Resolve the whole transcriptome within tissue architecture

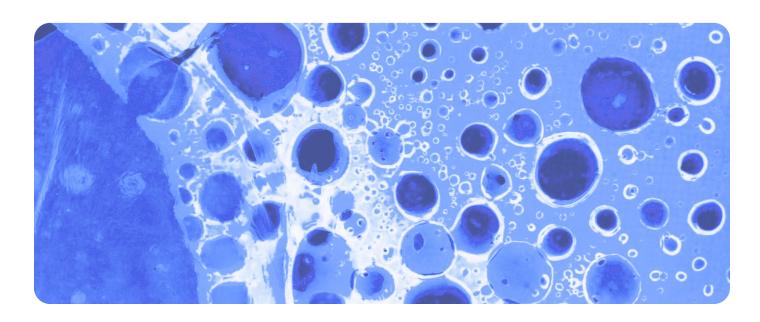
Application note: High-resolution, high-throughput spatial transcriptomics of complex tissues

Application note: High-plex spatial proteogenomics of FFPE tissue sections



Spatial RNA-Seq for human development research

Using spatial transcriptomics, researchers in Sweden delved into the complex development of the human brain, particularly focusing on the period from 5 to 14 weeks postconception. They identified a diverse array of neuronal and glial cell types, amounting to over a thousand distinct types that emerge early in brain development. Mapping the spatial distribution of these cell types revealed intricate patterns organized into hundreds of domains within the adult brain. This study detailed differentiation trajectories of the forebrain and midbrain, during the first trimester of human brain development.²⁵



Featured benchtop sequencing systems from Illumina

MiSeq i100 Series

The MiSeq i100 Series sets the new standard in sequencing simplicity to bring the power of NGS to more labs across the world. These low-throughput systems deliver our fastest run times yet, integrated data analysis, and significant sustainability advancements. As part of a comprehensive NGS solution, the MiSeq i100 Series provides same-day results for various applications, including transcriptomics, microbial genomics, and targeted gene sequencing studies.

The MiSeq i100 Series offers 10 different reagent configurations with read lengths up to 2 × 300 bp that support an output range of 5 million to 100 million reads and 1.5 Gb-30 Gb. This expanded capacity allows researchers to increase sample throughput and perform deeper sequencing than before.



Sample throughput for key applications on the MiSeq i100 Series^a

	Configuration	Reads/output per sample	No. of samples			
Flow cell			5M	25M	50M ^b	100M ^b
Output per flow cell		1.5–3 Gb	2.5–15 Gb	5–30 Gb	10-30 Gb	
Reads per flow cell		5M	25M	50M	100M	
Small whole-genome sequencing	2 × 150 bp 2 × 300 bp	1M	5	25	50	100
16S rRNA sequencing	2 × 300 bp	0.1M	50	250	384°	_
Targeted DNA sequencing (small panels)	2 × 100 bp	2M	2	12	25	50
Targeted gene expression profiling	2 × 50 bp	5M	1	5	10	20
mRNA-Seq	2 × 100 bp	25M	_	1	2	4
Immune repertoire sequencing	2 × 300 bp	5M with 30% PhiX spike in	_	3	7	_

a. Reads per sample and sample throughputs are estimates and highly variable, depending on the panel and desired coverage.

b. 50M and 100M flow cells will be available starting in 2025 for the MiSeg i100 Plus System only.

c. Based on available Illumina indexes; additional indexes can be added.

NextSeq 1000 and NextSeq 2000 Systems

The mid-throughput NextSeq 1000 and NextSeq 2000 Systems are thoughtfully designed to enable more insights and expansive application breadth on your benchtop. With 14 configurations and read lengths from 1 × 50 bp to 2 × 300 bp, these systems efficiently handle benchtop workflows with scalable output, rapid run times, and high data quality. The P4 flow cell on the NextSeq 2000 System enables data-intensive applications like single-cell sequencing and multiomics with up to 1.8B reads and 540 Gb output. These robust and easy-to-use systems are broadening access for regions or institutions that may not be able to afford a high-throughput system.

Sample throughput for key applications on the NextSeq 1000 and NextSeq 2000 Systems with XLEAP-SBS chemistry^a

	Configuration	Reads/output per sample	No. of samples				
Flow cell			P1	P2	P3 ^b	P4 ^b	
Output per flow cell			10-60 Gb	40-240 Gb	120-360 Gb	90-540 Gb	
Reads per flow cell			100M	400M	1.2B	1.8B	
Small whole-genome sequencing	2 × 150 bp 2 × 300 bp	1M	100	384°	384°	384°	
16S rRNA sequencing	2 × 300 bp	0.1M	384°	384°	_	_	
Shotgun metagenomic sequencing	2 × 300 bp	25M	4	16	_	_	
Immune repertoire sequencing	2 × 300 bp	5M with 30% PhiX spike in	14	56	_	_	
Targeted DNA sequencing (mid- to large-size panels)	2 × 100 bp	50M	2	8	24	36	
Exome sequencing	2 × 100 bp	8 Gb 100× coverage	_	10	30	45	
Total RNA-Seq	2 × 100 bp	50M	2	8	24	36	
mRNA-Seq	2 × 100 bp	25M	4	16	48	72	
Enrichment-based RNA-Seq	2 × 100 bp	25M	4	16	48	72	
Large whole-genome sequencing	2 × 150 bp	120 Gb 30× coverage	_	1	3	4	
Targeted methylation sequencing	2 × 150 bp	50M	2	8	24	36	
ATAC-Seq	2 × 50 bp	50M	2	8	24	36	
Single-cell RNA-Seq	2 × 50 bp	200M	_	2	6	9	

a. Reads per sample and sample throughputs are estimates and highly variable, depending on sample type and experimental objective.

b. P3 and P4 flow cells are only available on the NextSeq 2000 System.

c. Based on available Illumina indexes; additional indexes can be added.

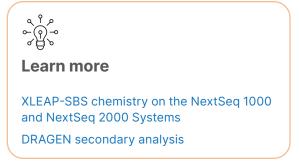
Day-to-day operational simplicity

Illumina believes that genomics should be available to the many, not the few. We are committed to making our technology as affordable and accessible as possible while setting the highest standard for data quality and security. Our benchtop systems are designed to be especially easy to operate, from run setup through analysis. The simple workflows require fewer touchpoints and fewer steps than other NGS systems, reducing the learning curve and the chance of user error.

Rapid and highly accurate data generation

Illumina benchtop sequencing systems integrate the latest technology advancements, including XLEAP-SBS™ chemistry and onboard DRAGEN™ secondary analysis. XLEAP-SBS chemistry, built on the foundation of proven standard Illumina sequencing by synthesis (SBS) chemistry, enables faster, more economical, and higher quality sequencing than ever before. XLEAP-SBS reagents are optimized for stability, performance, and speed, delivering higher confidence in generated data and expediting project completion.

DRAGEN secondary analysis is included on board the MiSeq i100 Series and NextSeq 1000 and NextSeq 2000 benchtop systems, offering automated bioinformatics pipelines with exceptional accuracy for variant calling.²⁶ Analyze whole genomes, exomes, methylomes, and transcriptomes with a single solution that replaces up to 30 open-source tools. Preconfigured workflows reduce time and expense for developing analysis pipelines, meeting researchers where their data and expertise are. DRAGEN secondary analysis is available through an on-premises server, in the cloud, or directly onboard the MiSeq i100 Series and the NextSeq 1000 and NextSeq 2000 Systems.



What's interesting and exciting is the versatility of NGS approaches. It's unbelievable the amount of discovery that can come from just a single piece of equipment, like a sequencer, and how many different questions you could use that one tool to approach in industry and academia. It's unbelievably cool.

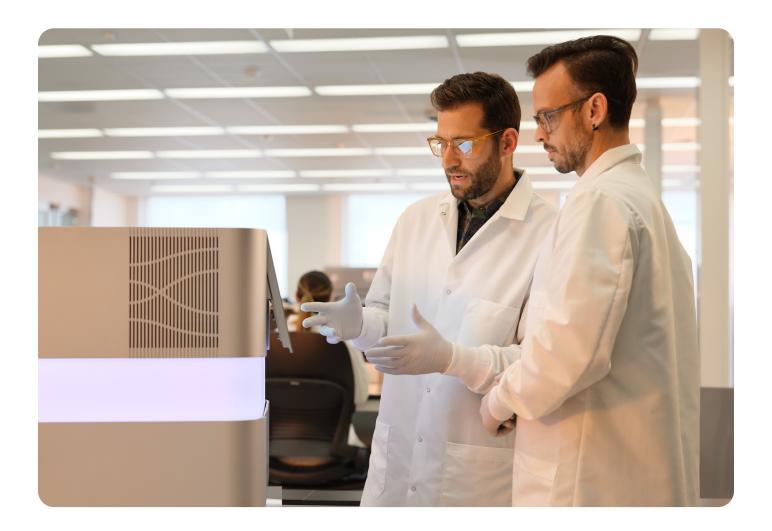
Cole Ferguson, MD, PhD



Illumina as a trusted partner

As scientists explore the molecular mechanisms underlying human health and disease, Illumina stands as a trusted partner, offering a comprehensive suite of solutions to power genomic research. With a diverse library preparation portfolio, high-quality data outputs, and intuitive analysis applications, Illumina workflow solutions enhance NGS capabilities with operational simplicity.

As the global leader in NGS technology, Illumina has installed over 25,000 instruments worldwide and its technology is referenced in more than 400,000 peer-reviewed publications—five times more than all other NGS providers combined.²⁷ Leveraging decades of expertise, Illumina continues to innovate and expand NGS applications, ensuring researchers benefit from reliable technology and precise data to accelerate scientific discoveries.



Summary

NGS offers a comprehensive, high-resolution view of biological systems to expand the discovery power of genomic scientists. With NGS, researchers can:

- Identify variants across thousands of target regions (down to single base resolution) in a single experiment
- Reveal a broader landscape of molecular entities, enabling the discovery of novel drug targets, signaling networks, and markers of disease
- Use an unbiased approach to provide untapped insights into biological phenomena, pathways, and systems

Benchtop sequencing systems are bringing the speed, power, and versatility of NGS to more laboratories across the globe. Low-throughput benchtop sequencing systems enable fast, culture-free microbial analysis and affordable, targeted DNA and RNA sequencing, and library QC. Mid-throughput benchtop systems provide access to data-rich methods, like exome or large-panel sequencing, single-cell analysis, and spatial biology. Illumina benchtop systems provide scientists with the precision, affordability, and accessibility to make their research ambitions a reality.

The beauty of NGS is that it provides a very big picture, and then we can find specific things that are surprising that we want to go deep into. It really opens the door to much wider possibilities, and then it helps us guide the next steps of research

Kristen Jepsen, PhD

Director, IGM Genomics Center University of California San Dlego



Learn more

Benchtop sequencing systems MiSeq i100 Series NextSeg 1000 and NextSeg 2000 Systems

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