

Enabling core laboratories to expand access to genomics

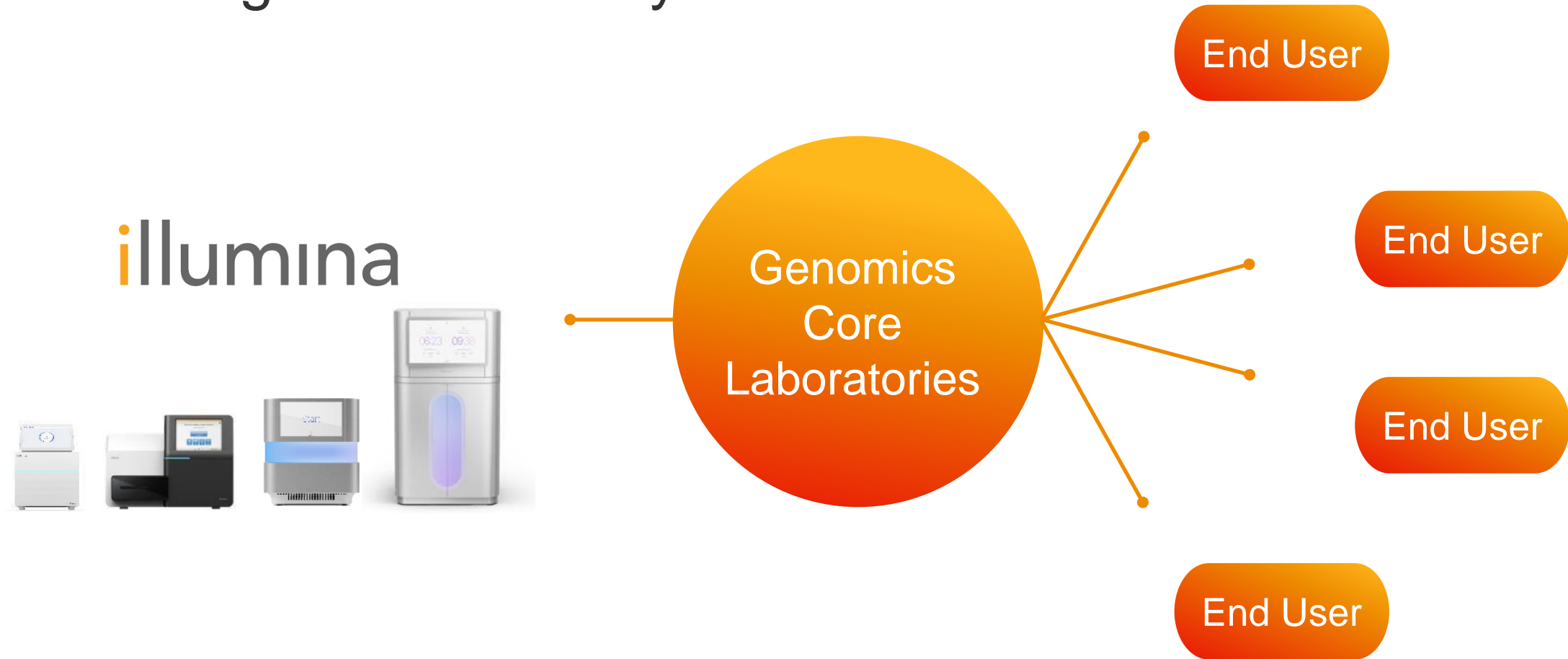
Sam Hester

Staff Product Marketing Manager – Benchtop Systems, Americas

12th October 2023



Core laboratories are an integral partner to vendors within the broader genomics ecosystem



Core labs democratize sequencing by helping researchers access equipment and expertise that would be otherwise inaccessible



Cost and lack of bioinformatics experience are major barriers to entry



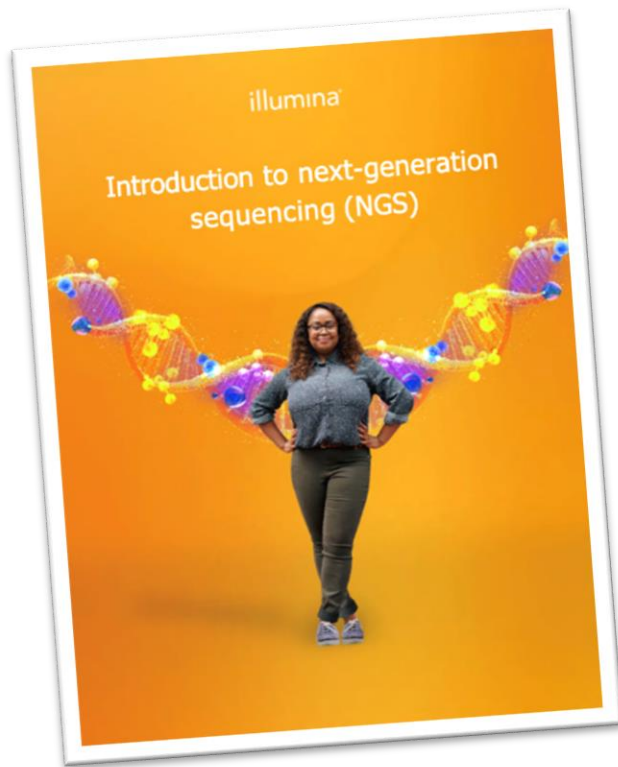
There's an "intimidation barrier" to NGS due to its relative complexity vs traditional methods



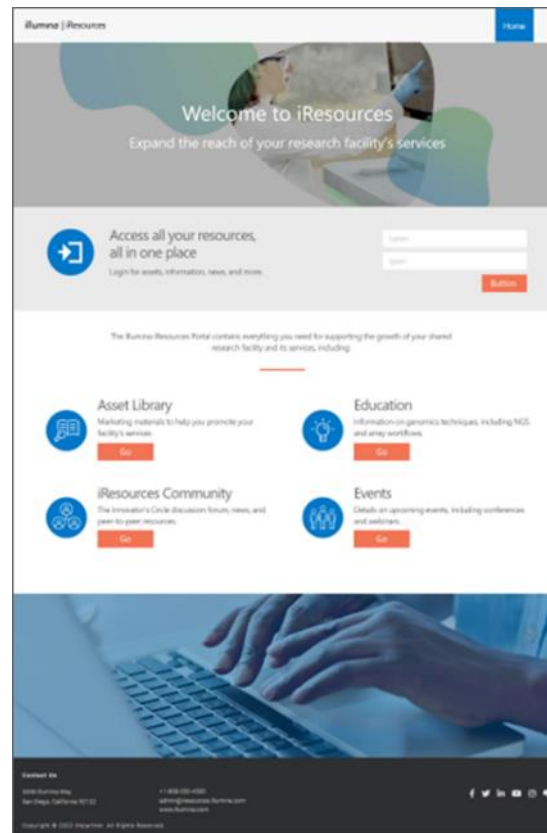
Core labs play a major role in educating the campus community about genomics fundamentals

Illumina is looking to partner with core labs to broaden the reach of genomics to more researchers

Content for researchers that are new to sequencing



Resources for core labs to promote facility services



Technology advancements to improve the core experience



New to Next Generation Sequencing (NGS)

Showing customers the value of collaborating with core
labs and driving new research

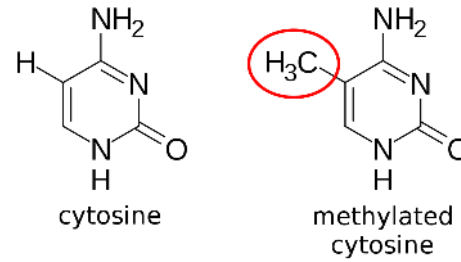


For most research customers, using NGS is a journey that starts small, but grows as they build expertise and confidence



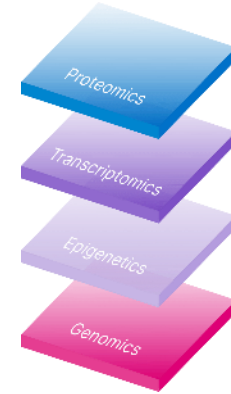
New to NGS

New to NGS refers to the majority of researchers in biology that **do not currently use NGS** for research.



Recent NGS Adopters

Researchers who start using NGS almost invariably do so via **outsourcing** (usually at a local core lab).



Experienced NGS Users

Experienced researchers do not fear complexity and are ready to **reduce inference** from data by **combining omics** or using higher depth approaches.

New to NGS is a set of educational resources that helps researchers learn about NGS and promotes new customer relationships at core labs.

New to NGS focuses on affordable, well-established methods, not products

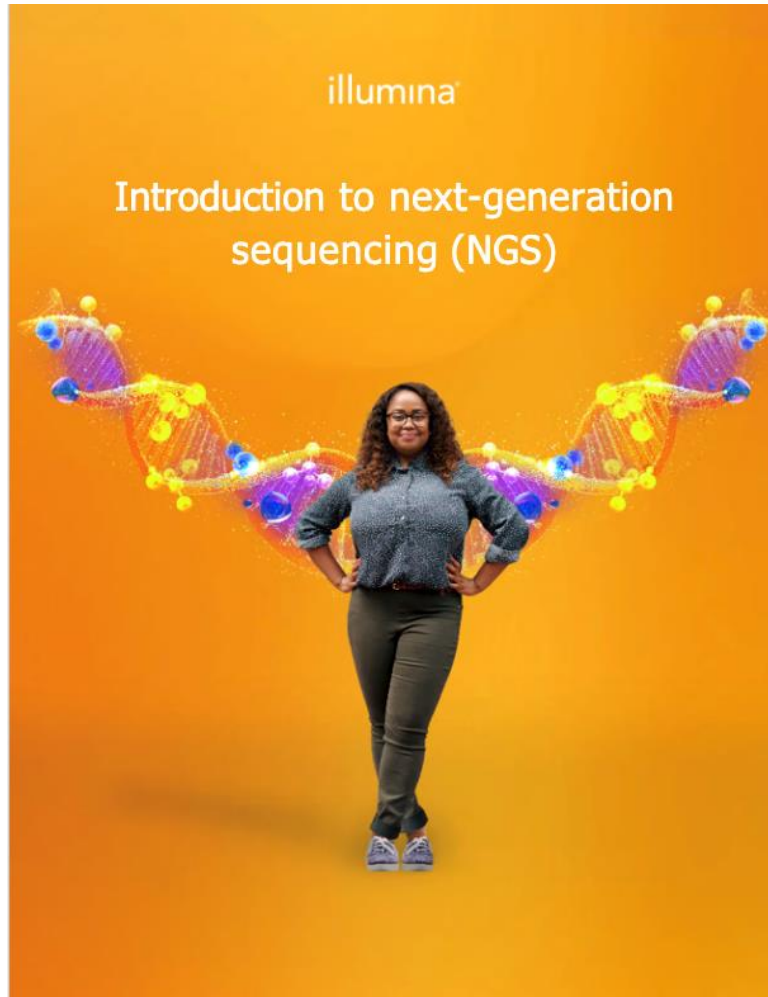
Key Themes:

- You don't need to be an expert to get started, collaborate with your local core lab.
- Sequencing is more accessible and cost-effective than ever before, especially with counting methods
- A modern look at why and when it makes sense to cast a wider net vs targeted approaches.

Key methods for getting started with NGS:

- Bulk RNA Sequencing
- Single Cell RNA Sequencing

New to NGS eBook



Section 2 illumina

How can NGS revolutionize your research?

Benefits of NGS over traditional molecular methods

When analyzing transcripts within a sample, researchers can choose between reading and counting applications, depending on the NGS method used.^{16,18} NGS-based reading methods, which sequence full RNA transcripts, allow researchers to identify rare RNA isoforms, splicing variants, and single-nucleotide variants (SNVs).¹⁷ On the other hand, NGS-based counting methods, which sequence only a part of RNA (or a barcode readout for another analyte), allow researchers to quantify gene expression at a low cost.^{19,20} Finally, NGS methods that combined both reading and counting allow researchers to gain a full view of both sequence identity and abundance of transcripts in a single, high-throughput assay.¹⁹

For variant screening studies with a high number of samples, NGS is the most efficient and cost-effective approach for the sequencing of tens to thousands of genes compared to conventional methods.

Traditionally, qPCR has been the go-to methodology used to count transcripts of interest.¹⁸ Like these qPCR experiments, NGS counting applications (such as 3' RNA-Seq) may not offer full sequence information for a set of expressed genes, but rather, provide just enough information to determine whether their expression levels have changed.²⁰

On the other hand, "counting" refers to the relative quantification of transcripts of interest within a sample, which provides insights into transcript abundances and gene expression changes under differential experimental conditions.^{21,22}

illumina transcriptomics methods offer reading, counting, and combinatorial reading and counting applications, depending on the NGS method used.

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illumina NGS combines the power of traditional reading and counting applications – identify full sequences and quantify expression changes in thousands of transcripts in a single assay with NGS.^{16, 19, 20}

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Section 3 illumina

What does an NGS workflow look like?

NGS at a glance

The overall workflow for an NGS experiment starts with the isolation of genetic material. Once you have your sample, you don't need to be an expert in NGS to get started: send your samples out to a local service provider, such as an academic core lab or a commercial lab.

Many service providers provide end-to-end support, and collaborating early in the experimental design process is the best way to get expert assistance at each step of your NGS workflow, from sample preparation to data analysis.

Collaboration with core labs simplifies the NGS workflow

Workflow

- Your lab: Set up your experiment, Harvest your samples, Follow-up experiments
- Collaborative: Experimental design: partner with a core lab to ensure your experimental design is optimized for NGS, Library preparation, Data interpretation
- Core lab: Sequencing, Bioinformatics

Using a core lab was crucial for my ability to start NGS. Between graduate studies and many other methods that I needed to master, using the core helped me get started without being an expert.

[Core labs] like to get involved with the researcher early on so that we can help direct experimental design, understand their biological question, and then make determinations from there in terms of what is the best method.

illumina RNA Sequencing Workflow Watch now

Dr. Amanda Toney, PhD candidate in Dr. Paula Cohen's lab at Cornell University

Dr. Adrian McNamee, Lead Biologist at the Genomics Innovation Hub

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An accessible introduction to illumina sequencing intended to overcome common intimidation barriers and promote collaboration with core labs

Infographics focus on grad students and postdocs who got started with genomics via core lab collaborations



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➤ RNA-Seq: A good place to start

NGS-based RNA sequencing (RNA-Seq) is a versatile NGS method that examines the expression of thousands of genes in a single assay, making it possible to characterize complex pathways.^{8,9}

"For us, sequencing changed everything. If we wouldn't have been able to use bulk RNA sequencing, we wouldn't have been able to see what's going on at the chromosomal level in our mutants."

Mercedes Carro, PhD, Postdoctoral Associate Cohen Lab, Cornell University



"Working with the core lab gave me confidence. I was less worried about my experiment or quality control issues."

Connor Kean, Graduate Student, Grimson Lab, Cornell University



➤ Get started with the help of NGS professionals

You don't need to be an expert to get started: collaborating with core labs and service providers is the best way to get assistance at each step of the NGS workflow, from designing your experiment to running your samples.

➤ No bioinformatics expertise required

NGS produces large data sets that can be intimidating for new users. Core labs and the latest applications can help you analyze and understand your data, even without bioinformatics experience.

"I think this collaboration really gave me the confidence to know that I can do this type of analysis."

Amanda Touey, Graduate Student, Cohen Lab, Cornell University



New to NGS articles in GEN and The Scientist focus on introduction to common sequencing methods and experiences of real users

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NGS Answers Ambitious Questions

The cost of sequencing continues to decrease. Now is the right time to start using next-generation sequencing to answer your scientific questions and further your research goals

Who has time to wait for a serendipitous discovery to occur? Good science starts with a good question. An unbiased approach to scientific research, like next-generation sequencing (NGS), can answer the questions posed as well as free experimental design from the bounds of prior understanding and preconceived expectations and uncover new findings. NGS provides the power to sequence vast amounts of genetic material at a fraction of the time and cost of traditional methods.¹ As the cost continues to decrease, this technology has become a critical asset to scientists. Now is the right time to leverage this impartial approach to reveal a broader landscape of molecular entities and enable deeper understanding of biological phenomena, pathways, and systems.

Easily Accessible Experts

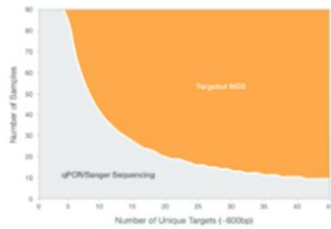
An impactful effect of the genomics era has been the establishment and growth of core labs and service providers. Unlike decades past, a researcher starting a new lab no longer needs to own all of the equipment that is generally needed for a given method. Core labs have all the instruments and knowledge in place to help build expertise and knowledge and often offer training and one-on-one consultations. Many also excel at facilitating advantageous connections between the wide swaths of scientists they service.

Regardless of your level of experience—from novice to expert—a core lab can help with either a portion or the entire process from experimental design to library prep, sequencing, and data analysis.

"The most beneficial thing about a core lab is that it is our job to be on the forefront of our area of expertise, and it is a scientist's job to be on the forefront of their expertise. We can work together to advance science," said Ann Tate, Project Manager, Transcriptional Regulation and Expression (TReX) Facility, Cornell Institute of Biotechnology. "Knowing what your question is as a scientist and just coming to us with that question is often a great first step because we can help you decide what kind of technology will work best for you and how best to use it."

Charlie Johnson, PhD, Director, Genomics & Bioinformatics Services at Texas A&M University, summed it up by saying, "We like to do cool stuff with cool people!"

Core facilities perform a broad range of activities; some are standardized, but many are customized. They constantly tailor their procedures to the data the researcher wants to receive from the experiment. For example, working in a core facility gives Adrian McNairn, PhD,



Cost-effectiveness for targeted resequencing vs. PCR and Sanger Sequencing. The area above the line represents higher cost effectiveness with targeted DNA sequencing compared to Sanger sequencing or qPCR.

Senior Research Associate, Genomics Innovation Hub, Cornell Institute of Biotechnology, an opportunity to both develop methods as well as help teach others how to use them.

"People can be ambitious when it comes to genomics because with the speed of the field, you can dream big and actually be in a position to obtain that goal, especially in collaboration with core facilities to have access to the equipment you would need," McNairn said. "We want to be a space where people can come in, collaborate with us, get the training, have access to reagents, and actually get their projects off the ground and to the point where they have publishable data."

Thousands of Targets—Unique Discoveries

Initially, NGS often intimidates many scientists, from graduate students to postdoctoral fellows to principal investigators. Experimental design, sample collection, and analyzing and interpreting the vast amount of data NGS provides appear insurmountable.

Both Mercedes Carro, PhD, a postdoctoral fellow, and Amanda Touey, a graduate student, study spermatogenesis in the Cohen Lab at Cornell University. Transcriptional regulation and dynamics are critical to understanding spermatogenesis. Part of the puzzle is the differential expression of Argonaute proteins that bind to small noncoding RNAs to regulate gene expression.

Neither came from a sequencing background nor had used NGS.

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Next-Generation Sequencing: A World without Limits

Centralized core facilities and commercial service providers specializing in NGS provide expertise and training for researchers new to the method.

From bulk and single cell methods to spatial and multiomic methods, advancements in sequencing and -omics research are accelerating at an exciting pace. Next-generation sequencing (NGS) provides researchers from various scientific fields the ability to cast a wide net and explore complex pathways and biological processes. With NGS, scientists can take an untargeted approach, generating data from many genetic loci at once.

Such open-ended discovery allows researchers to see details that may be overlooked in targeted approaches that assay one gene at a time. Despite this benefit, the technical aspects of library preparation and data analysis can be daunting for researchers new to NGS. Previously, high sequencing costs, lack of expertise, and enormous data volumes were seen as barriers to entry.^{1,2} Over time, these barriers have diminished thanks to decreased costs and optimized workflows and bioinformatics pipelines.^{3,4} In addition, numerous academic NGS core facilities and commercial service providers have been implemented globally to support researchers on their NGS journeys. Such access to centralized equipment and expertise makes NGS more approachable to researchers new to the method.

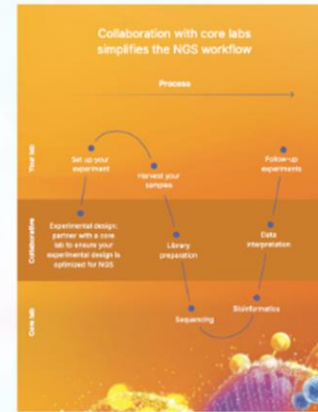
Support for Scientists Diving into NGS

Instead of entering the NGS world alone, scientists can take their experiments to a core facility and receive expert guidance. "Our core motto is reducing barriers to genomic technologies. That means that whatever point you are in your knowledge of genomics, we want to be there to help you," said Ann Tate, project manager for the Transcriptional Regulation and Gene Expression Facility (TReX) in the Biotechnology Resource Center (BRC) at Cornell University.

According to Tate, prior sequencing experience is not important when initiating an NGS project with a core facility. Some researchers may simply hand off their samples and wait for results, while others choose to take a hands-on approach, collaborating with core members from the onset of their project.

"We want to be a makerspace where people can come in, collaborate with us, get the training, have access to reagents, and actually get their projects off the ground and to the point where they have publishable data," said Adrian McNairn, the lead biologist for the Genomics Innovation Hub in the BRC. From new graduate students lacking wet bench experience to tenured professors trying bioinformatics for the first time, researchers can work side-by-side with NGS core members who use their expertise to tailor technologies to specific research questions, train scientists in sequencing best practices, and make data analysis more accessible.

Core facilities and commercial service providers democratize NGS so that all scientists can access this powerful technology. Working with a core that has equipment such as sequencers, incu-



bators, and fragment analyzers provides access in a way that is no longer cost prohibitive. "An example of a cool project that I did with someone who now has access... is iridescent butterfly wings," said Tate. "We were sequencing butterfly wing cells. Five years ago, that would have been cost prohibitive to them." Additionally, samples from lesser-studied species may require changes to existing reagents, workflows, or data analyses, which core members help customize thanks to their deep-seeded NGS knowledge. "We have seen everything from shark eyeballs, to pond scum, to broccoli and cauliflower," Tate stated.

A hands-on, expert approach is also key when processing samples, building libraries, and analyzing data. As McNairn works with researchers, he evaluates the quality of their samples every step of the way. "Since we've worked with them through the whole process, we can go back and troubleshoot the areas that we know maybe went wrong," said McNairn. In addition to support at the facility, core members facilitate connections between researchers

- Explore stories of NGS becoming a crucial tool for early career researchers
- How a network of genomics core labs with Illumina technology can help researchers gain expertise and confidence in NGS approaches
- Focus heavily on actual user interviews so that the case for NGS is made from their real world perspective

New to NGS Poster in The Scientist

Take your research higher with next-generation sequencing.

We're ready when you are. Let's get you started!

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To learn more about how to get started with NGS, download our new eBook.

References

1. Wang Z, Gerstein M, Snyder M. Genomic profiles of transcription initiation and promoter-proximal pausing. *Nature* 2008;455:1079-83.
2. Wang Z, Gerstein M, Snyder M. The three-dimensional structure of the human genome. *Nature* 2009;461:43-52.
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NGS is a powerful tool for answering your research questions, and it's easier than you think to get started.

1. There is definitely no reason to be intimidated...there's so many different ways to find support, especially within the core lab.
Amenda Torrey, PhD Candidate, Cornell University

2. You don't have to be an expert to work with the core lab to understand every step of the sequencing part itself, because the core will help you with that.
Levin Yi, PhD, Postdoctoral Research Associate, Grinnell Lab, Cornell University

3. Your core lab are your ally. They're the ones who are going to help you make your project a success.
Farshad Hassani, MD, PhD, Staffbase Neurosurgeon and Scientist, University of Toronto

Researchers can partner with an accessible network of core labs for comprehensive NGS support.

4. We want to be a research partner to support whatever it is that the scientists need to do for their research.
Teresa Pugh, PhD, Director of Genomics, Senior Principal Investigator, Ontario Institute for Cancer Research

5. The relationship you build with your core lab is critical to the science that you produce. Let us work with you to make sure you have the data that you need in time for that grant submission.
Morgan Tashiro, Director, Genomics Sequencing Informatics, Ontario Institute for Cancer Research

6. The most cost-effective way to get started with sequencing is to work with your core.
Adrian McNamee, PhD, Lead Biologist, Genomics Innovation Hub, Cornell University

Compared to targeted molecular approaches, NGS greatly increases your discovery power.

Unbiased discovery with NGS can:

- Fully characterize complex molecular pathways that would otherwise remain unexplored
- Drive your field of expertise forward
- Find the answers to your toughest research questions

There's never been a better time to start using NGS.

- 100s of expert core labs and service providers across North America
- 96% increase in sequencing reads since 2013
- 87% increase in submissions using NGS methods since 2013

Why and when to consider Illumina NGS

When studying >20 targets, NGS is...	More cost-effective****	More sensitive***	More efficient**
DNA NGS vs. Sanger Sequencing	6,000x less expensive to sequence the human genome	Up to 15% lower threshold for detecting mutant alleles	Up to 1,000x more genes sequenced in a single assay
RNA NGS vs. qPCR	> 150x less expensive to profile the human transcriptome**	More low-frequency variants detected	> 1,000 target regions profiled in a single assay
Protein NGS vs. Flow cytometry	up to 530x more proteins analyzed in a single experiment**	32% higher detection rate of minimal residual disease (MRD) using NGS compared to next-generation flow cytometry (NGF)	0 known biomarkers required compared to traditional antibody-based protein assays*

Collaboration with core labs simplifies the NGS workflow

Process

Your lab: Set up your experiment, Harvest your samples, Follow-up experiments

Collaborative Core Lab: Experimental design (partner with a core lab to ensure your experimental design is optimized for NGS), Library preparation, Sequencing, Bioinformatics, Data interpretation

iResources, a new online portal exclusive to core lab facilities



iResources is an online destination that allows core lab members access to Illumina-provided content in one place

Explore & Engage

Laboratory management can be complex. Finding support doesn't have to be. Access all the resources you need for promoting your shared research facility's services, all in one place.



Asset Library

Marketing materials to help you promote your facility's services.

GO



Education

Information on genomics techniques, including NGS and array workflows.

GO



iResources Community

The Innovator's Circle discussion forum, news, and peer-to-peer resources.

GO



Events

Details on upcoming events, including conferences and webinars.

GO

Asset Library provides educational content and marketing materials for core labs to promote their services and expertise

Asset Library

Collections Playbooks

All

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Search for a specific asset...

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SELECTED COLLECTION: All

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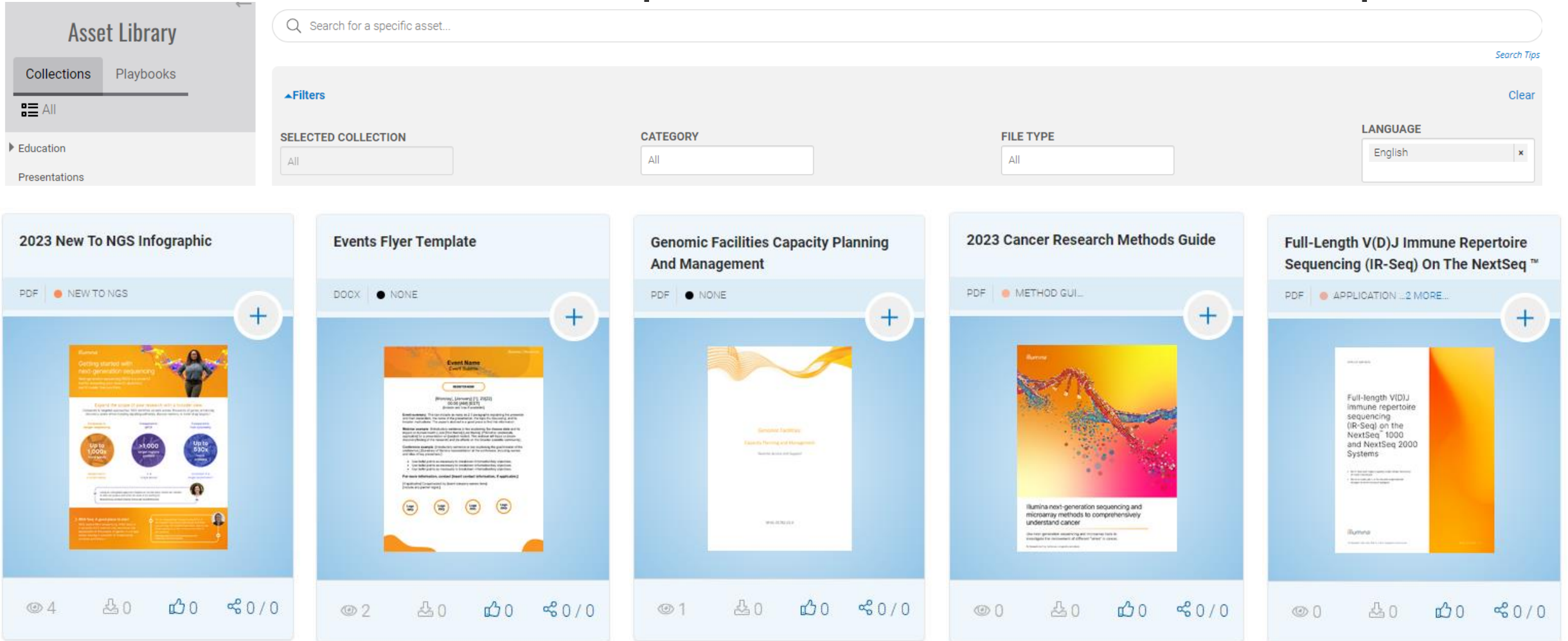
2023 New To NGS Infographic
PDF • NEW TO NGS

Events Flyer Template
DOCX • NONE

Genomic Facilities Capacity Planning And Management
PDF • NONE

2023 Cancer Research Methods Guide
PDF • METHOD GUI...

Full-Length V(D)J Immune Repertoire Sequencing (IR-Seq) On The NextSeq™
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Education section specifically for new core lab team members

Learn how Illumina technology works and what it can do for you

From library prep to data analysis, Illumina offers tools and support throughout the entire NGS and array workflows. Learn more about each part of the process.

Methods



Learn about key methods like WTS, Single-cell RNA-Seq, WES, and more.

[Learn More](#)

Arrays



Resources about Illumina's bead-based array technology.

[Learn More](#)

Library Prep



Discover ways to optimize workflow with fast and simple library prep.

[Learn More](#)

Sequencing



Information about the diverse types of sequencing.

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QC & Troubleshooting



Discover methods for QC and troubleshooting to ensure confidence and accuracy in your sequencing.

[Learn More](#)

Analysis



Learn about the myriad of options for analyzing your samples with the latest software and informatics tools.

[Learn More](#)

iResources Community has the latest news in genomics and sequencing and links to the ABRF Community Site



Sequencing News

Read the latest news related to genomics, including NGS, arrays, and more.

GO



Peer-to-Peer Resources

Share methods and learn how others are using NGS and array technologies in their shared research facilities.

GO

Welcome to the Core Community

Collaborate with peers to share strategic advice, solve challenges and develop new approaches.

[Click here for a tutorial on how to use the Community](#)

Links both for Illumina events and webinars and ABRF conferences (like WACD!)

Stay informed of upcoming events

Browse upcoming conferences and webinars to connect with the community and stay up to date.



Core Community

Welcome to the Core Community

ABRF 2024 Annual Meeting
Save the Dates....

The Value of ABRF Membership

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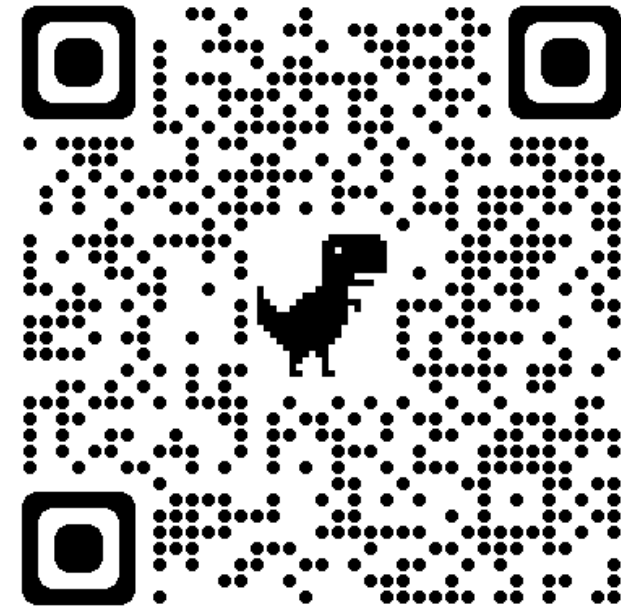
iResources Core Lab Portal

We're just getting started together!

Join now to access our new source of core lab content.

Give us feedback! What else would you like to see?

iresources@illumina.com



Technology updates and innovation roadmap

The NovaSeq™ X Series provides more power to unlock the genome



Powerful

XLEAP-SBS™ Chemistry
2x speed improvement
2.5x throughput improvement¹

>20,000 Genomes per year

Sustainable

Only high-throughput instrument with ambient ship reagents

61% Reduction of climate change impact²

90% Reduction in waste

Cost-Effective

Only high-throughput instrument with integrated analysis

\$200 Genome with analysis³

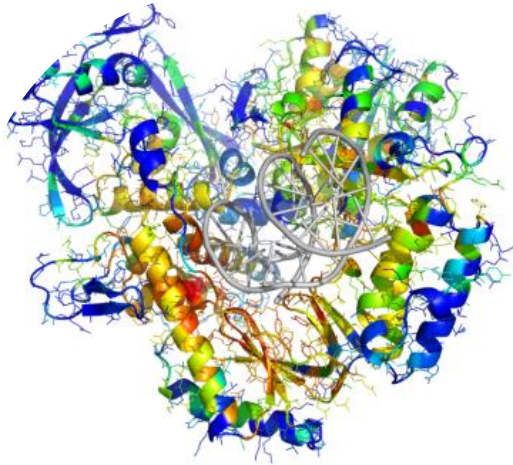
The NovaSeq X Series has **>40** new patents pending, and took **5** years and **1,500** scientists, engineers, developers, and designers to create

¹ Compared to NovaSeq 6000.

² Preliminary results of streamlined LCA conducted by external party regarding climate impact per Gb of genetic code of NovaSeq X 10B. 300 cycle kit compared to NovaSeq 6000 S4 300 cycle kit per Gb of genetic code.

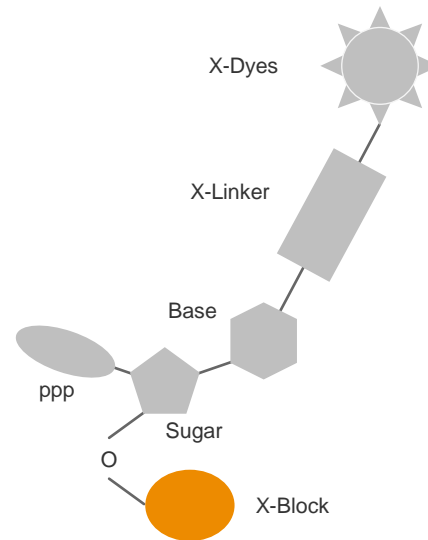
³ High-quality whole genome sequencing at scale with alignment, variant calling and lossless compression. \$200 genome will be enabled by the 25B flow cell on NovaSeq X Series in Q4 of 2023.

Higher performance with XLEAP-SBS chemistry on NovaSeq X series and NextSeq™ 1000/2000



Novel Polymerase

Faster incorporation, higher fidelity



X-Block, X-Linker, X-Dyes

Most resistance to heat, ~50x more stable in solution, ~500x more stable lyophilized, faster block cleave

Enables ambient shipment*



Faster cycle times



Greater accuracy



More sustainable**

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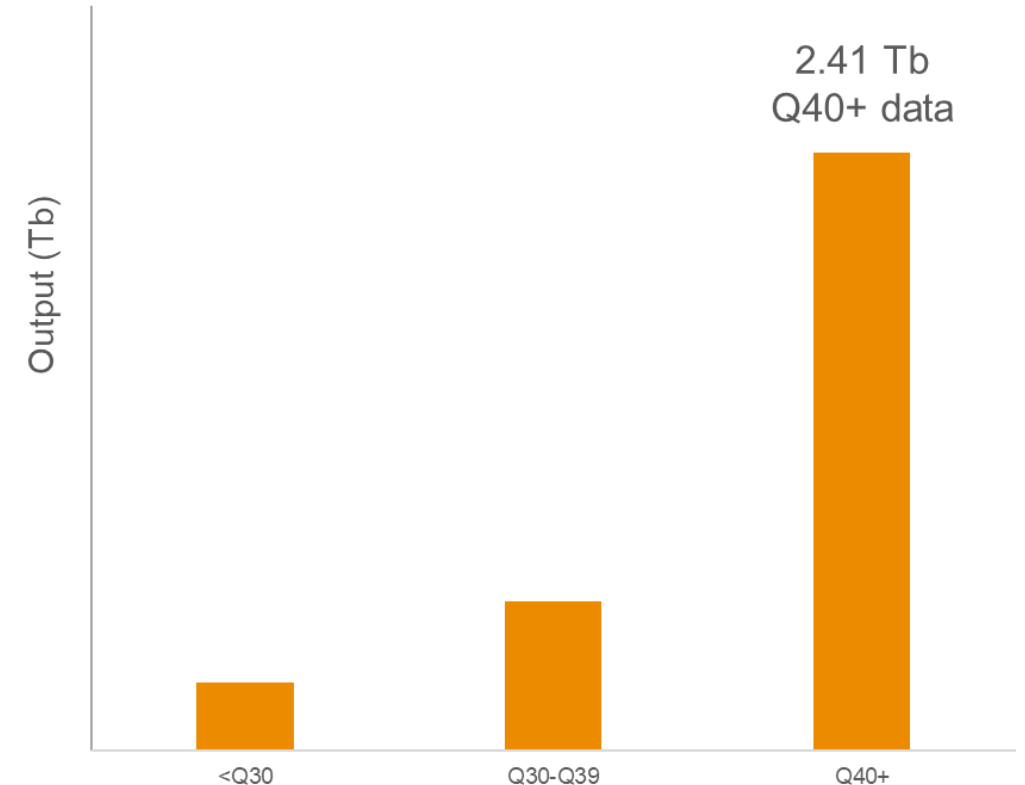
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*Ambient shipment currently on NovaSeq X series only

**as compared to NovaSeq 6000

Illumina continues to demonstrate industry-leading performance – NovaSeq X data exceeds specifications

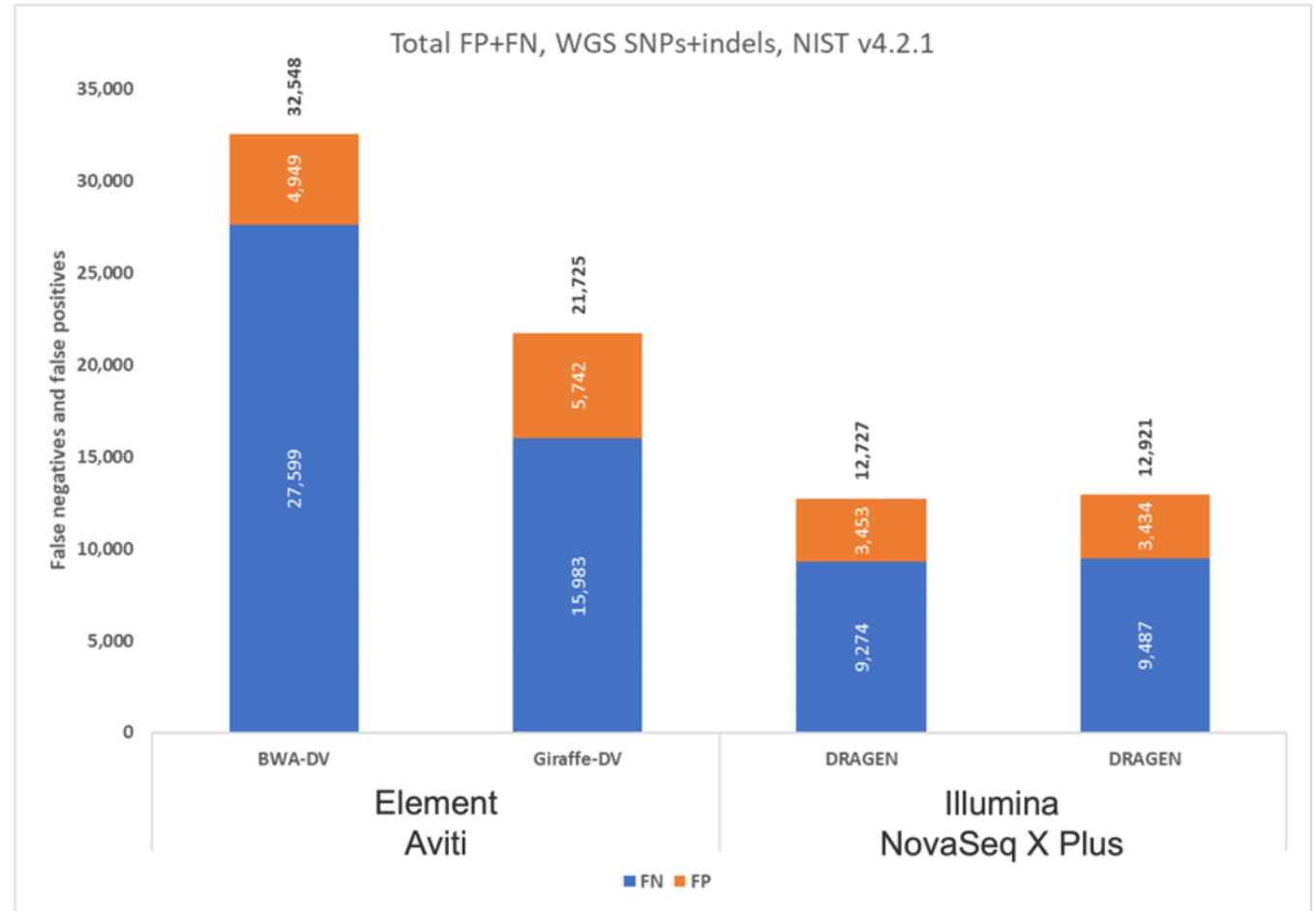
- NovaSeq X 10B specifications:
 - Data by Q-score: **85% \geq Q30**
 - Yield: **3 Tb**
- Unbinned Q-scoring shows most bases \geq Q40*
 - **2.41 Tb** of called bases measured \geq Q40
 - Human PCR-free WGS 2x150 bp run



*Data generated with NovaSeq X 10B flow cell while measuring un-binned Q-scores via R&D configuration of RTA basecalling software

NovaSeq X + DRAGEN™ – more accurate variant calling

- Reported quality claims must align with assay accuracy to hold value
- Market participants claim higher %>Q40 output yet final accuracy results do not reflect an advantage
- Example: >30x human whole genome PCR-free (HG002 / NA24385)



<https://go.elementbiosciences.com/access-seq-datasets-060622>

Shrinking the Environmental Impact of High-Throughput Sequencing



61% Reduction of climate change impact¹



90% Reduction in waste



90% Reduction in weight

The NovaSeq X Series is **the only** high-throughput instrument with ambient ship reagents

¹ Preliminary results of streamlined Life Cycle Assessment (LCA) conducted by external party regarding climate impact (per Gb of genetic code of NovaSeq X 10B 300 cycle kit compared to NovaSeq 6000 S4 300 cycle kit for the US market, and 41% reduction for UK market (savings are reduced for UK due to replacement of some air freight with truck transport)

More multiomics than ever before with 25B and better batching with 1.5B on the NovaSeq X series in Q4 2023



1.5B Flow Cell

≥1.6B clusters

~165-500 Gb

100c, 200c, 300c

≥85-90% Q30

~13-21 hrs



25B Flow Cell

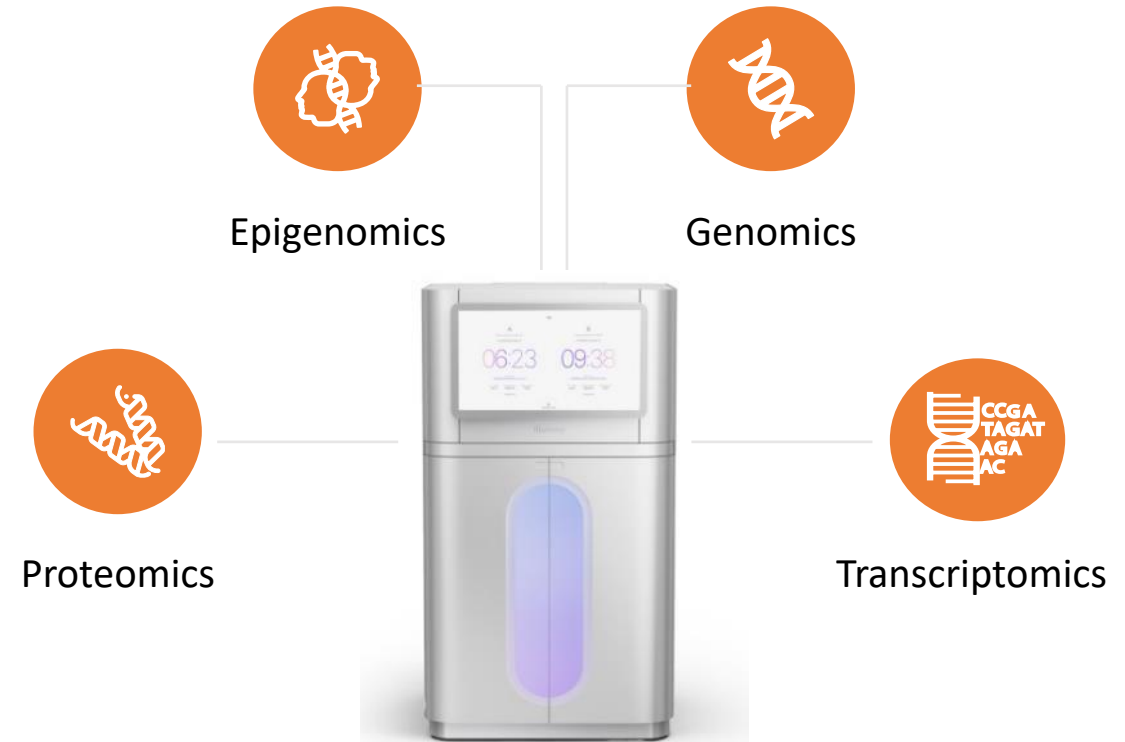
≥26B clusters

~8 Tb

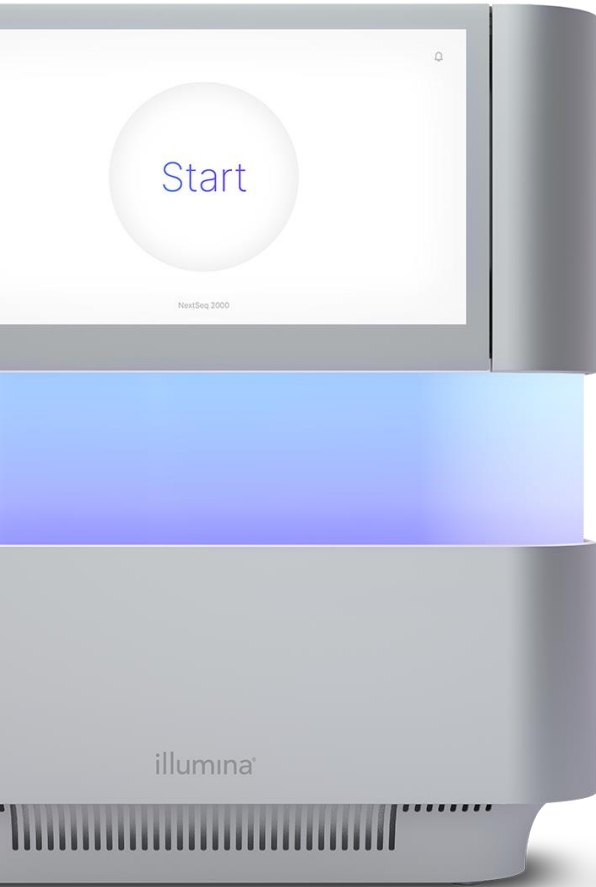
300c

≥85% Q30

~48 hrs



The NextSeq 1000/2000 has revolutionized benchtop sequencing



Flexible and Scalable

11 kit configurations with read lengths from **<50bp to 2x300 bp**; supports wide range of applications, from microbes and panels to large genomes



Illumina Quality

A field-tested solution for providing reliable and consistent results, with thousands of publications and **1800+ system placements**



Streamlined Workflow

On-board denature & dilute, cluster generation simplifies run set up, **onboard DRAGEN** delivers simple, accurate and integrated analysis, no wash maintenance needed








Continuous Innovation

Coming in H1 2024, **XLEAP-SBS Chemistry** will unlock the next level of scale, cost efficiencies, and capabilities

Longer reads capabilities on NextSeq 1000/2000 enables customers to expand applications and scale more efficiently

P1 & P2 600c kits



	READ NUMBER	100M & 300M
	OUTPUT	60Gb & 180Gb
	RUN TIME	34 hr & 44 hr
	QUALITY	≥ 80% of bases higher than Q30
	APPLICATIONS	Metagenomics, immune repertoire, sWGS

NextSeq 1000/2000 with XLEAP-SBS™ Chemistry will unlock the next level of scale, cost efficiency and capabilities

Coming Q1 2024



New P4 Flow Cell*
500Gb, 1.7B Reads**



XLEAP SBS

Coming Q2 2024



Existing P1/P2/P3 Kits
Improved run time and performance

Pricing improvements for all new XLEAP-SBS kits

XLEAP-SBS compatibility will be available with a FREE software upgrade upon release

*Available only on the NextSeq 2000. **Preliminary spec target only. 500Gb for P4 300c kit.

P4 reagents powered by XLEAP-SBS chemistry on NextSeq 2000

A variety of read lengths for application flexibility at scale

P4 available to order Q1 2024
~1.7B Clusters

50 cycles

~83 Gb
12-13 hrs

100 cycles

~167 Gb
19-20 hrs

200 cycles

~333 Gb
31-32 hrs

300 cycles

~500 Gb
43-44 hrs



NextSeq 1000/2000's flexibility enables customers to expand applications and scale efficiently

	P1	P2	P3	P4 Coming Q1 2024	Legend
De novo assembly (sWGS)	✓	✓			2x50 or below
Targeted metagenomics	✓	✓			2x100
Shotgun metagenomics, metatranscriptomics	✓	✓			2x150
Immune Repertoire Profiling	✓	✓			2x300
Targeted Panels (amplicon, enrichment)	✓ ✓	✓ ✓			
RNA-Seq & Gene Expression	✓	✓	✓	✓	
Single Cell & Spatial Profiling	✓	✓	✓	✓	
Exome & Large Panels		✓ ✓	✓ ✓	✓ ✓	
WGS & Liquid biopsy			✓	✓	

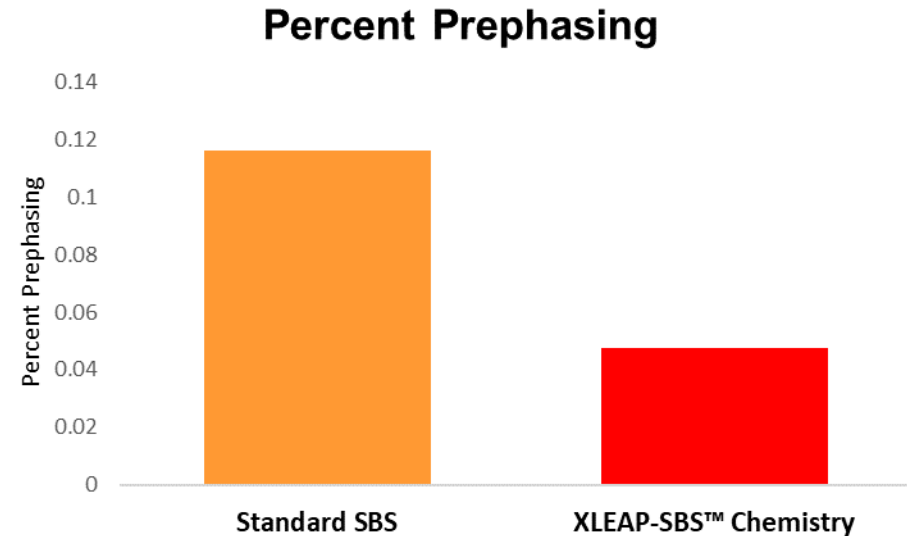
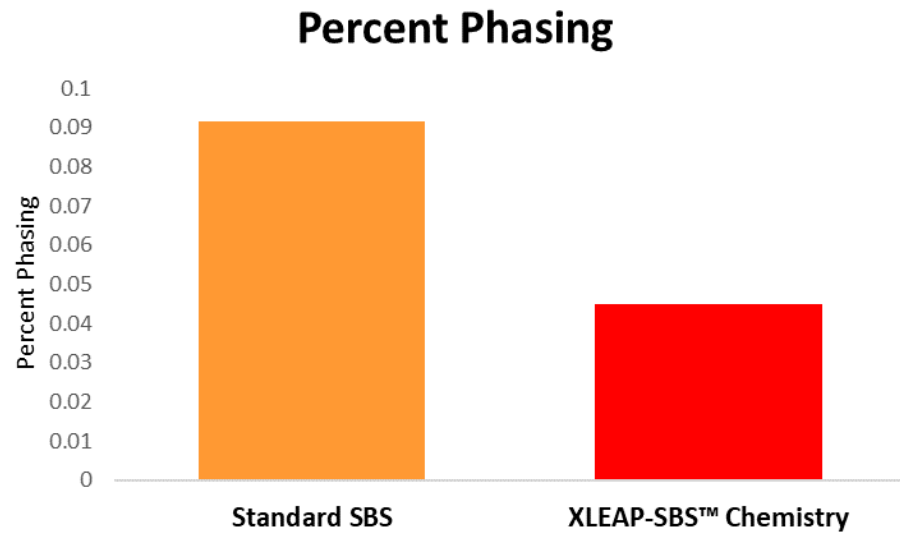
Enabling read length dependent applications

Improved batching size for scalability

Faster TAT and increased batching flexibility

P4 will maximize batching and scale for applications such as exome, single cell, and spatial sequencing on a benchtop system

XLEAP-SBS drives improved quality and performance on NextSeq 1000/2000

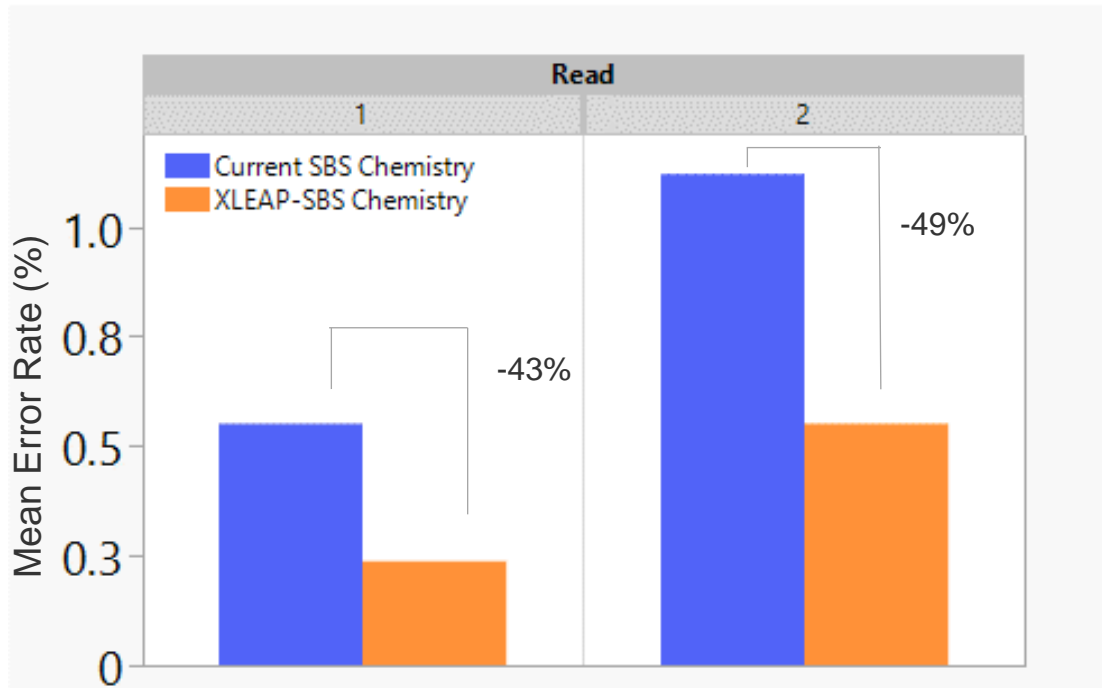


Greater than **50% reduction in phasing and prephasing** relative to standard Illumina SBS chemistry on NextSeq 1000/2000

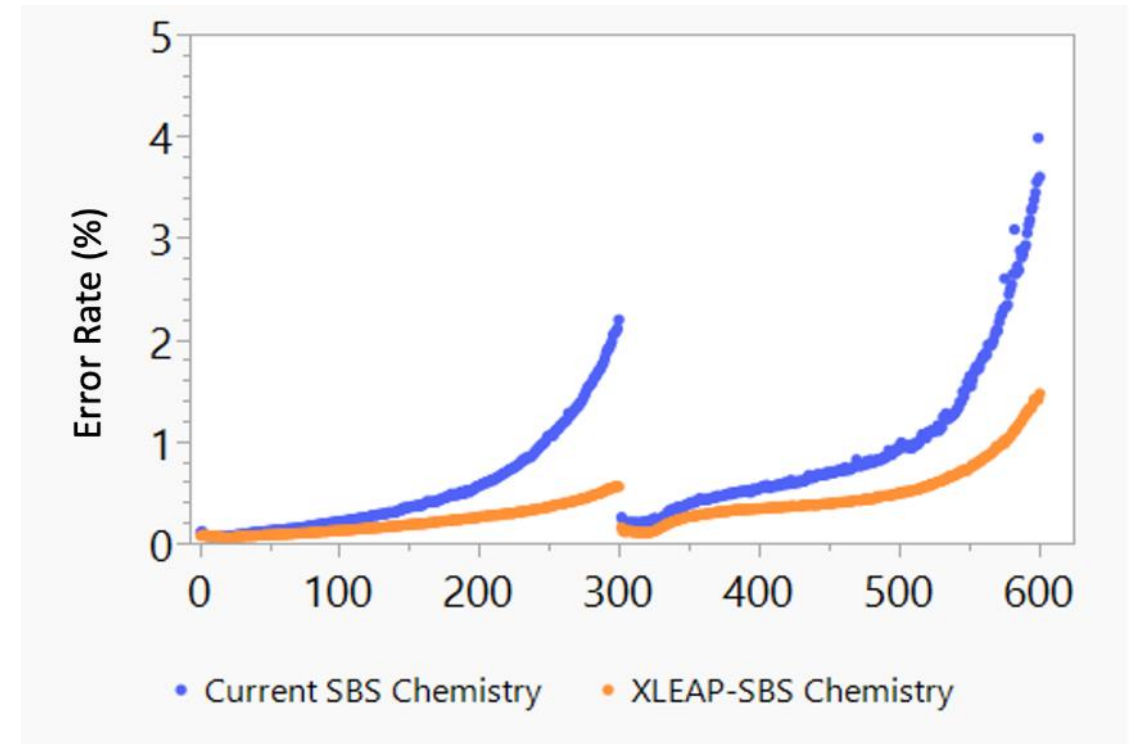
Data collected on R&D NextSeq 1000 / 2000 system.

XLEAP-SBS 600 cycle kits improves data quality on NextSeq 1000/2000

XLEAP-SBS error rates are significantly lower than current SBS Chemistry error rates

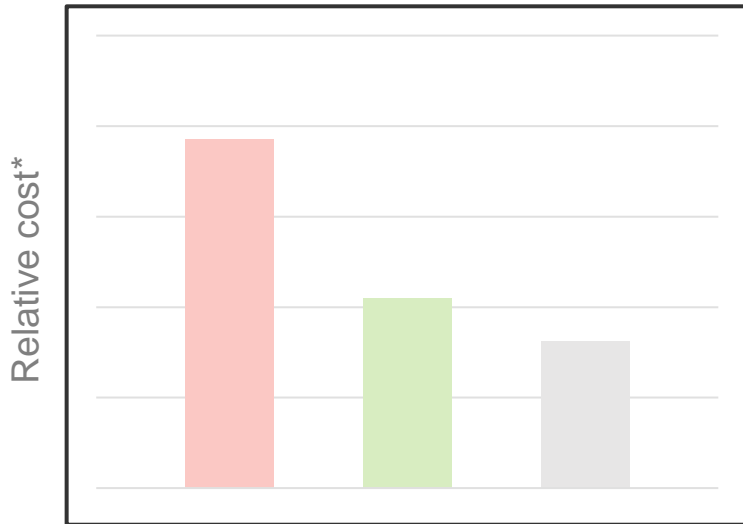


Testing demonstrates high quality data, especially at the end of the read length

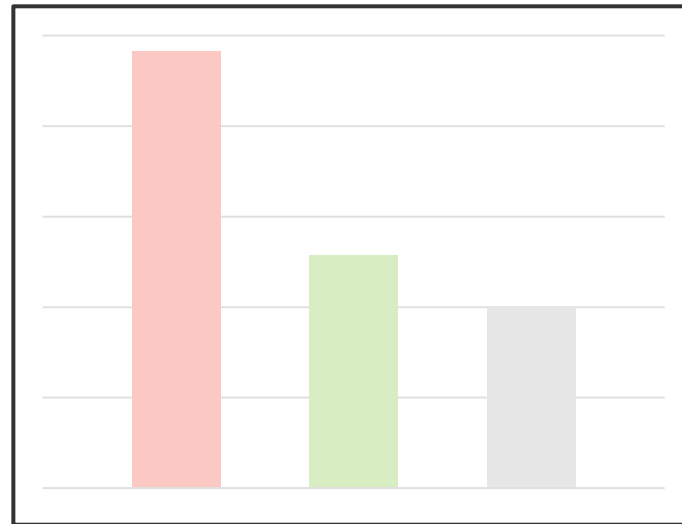


XLEAP-SBS enables improved sequencing costs for data intensive methods with the P4 flow cell

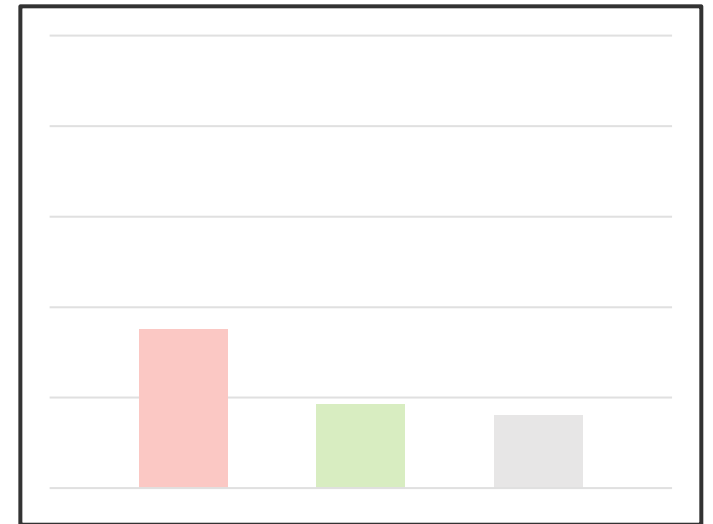
Whole Exome Sequencing



Transcriptomics



Single Cell RNA

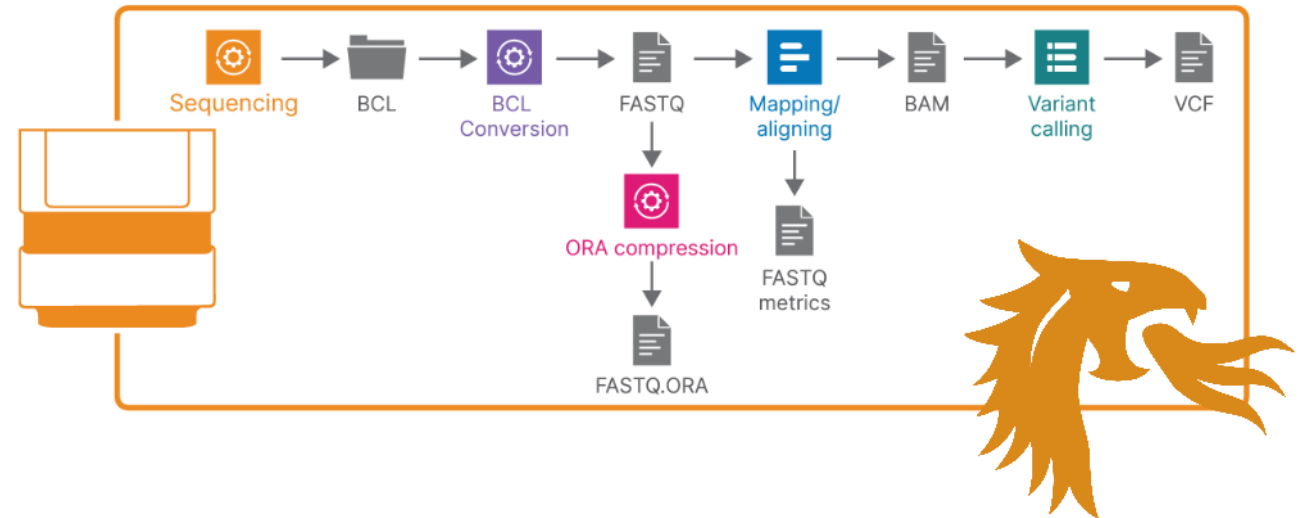


■ P3 Current SBS ■ P4 XLEAP-SBS ■ NovaSeq X 1.5B

NextSeq 1000/2000 is the only benchtop sequencer with integrated, onboard analysis—at no extra cost

- Provides secondary analysis up to **16x faster** than open-source methods
- **Saves up to 80% on storage costs** with DRAGEN ORA file storage
- Achieves **~4x lossless compression** of FASTQ files
- Enables push button analysis, integrated with the instrument, at **no additional cost**

Save >\$100K over 5 years*



*Assuming data is analyzed with DRAGEN onboard and stored with Illumina cloud platforms (ICA or BaseSpace). Savings will vary.

DRAGEN v4.2 coming to NextSeq 1000/2000 in Q1 2024 – more accurate and comprehensive than ever before



Accuracy

- Enhanced Multigenome (graph) reference and Machine Learning (ML) improve small variant calling accuracy
- Improved CNV and Structural Variant calling accuracy
- New targeted callers for higher genotyping accuracy - HBA, LPA and RH, CYP21A2
- Accurate genotyping for 5 more pharmacogenes: BCHE, ABCG2, NAT2, F5, UGT2B17

Comprehensiveness

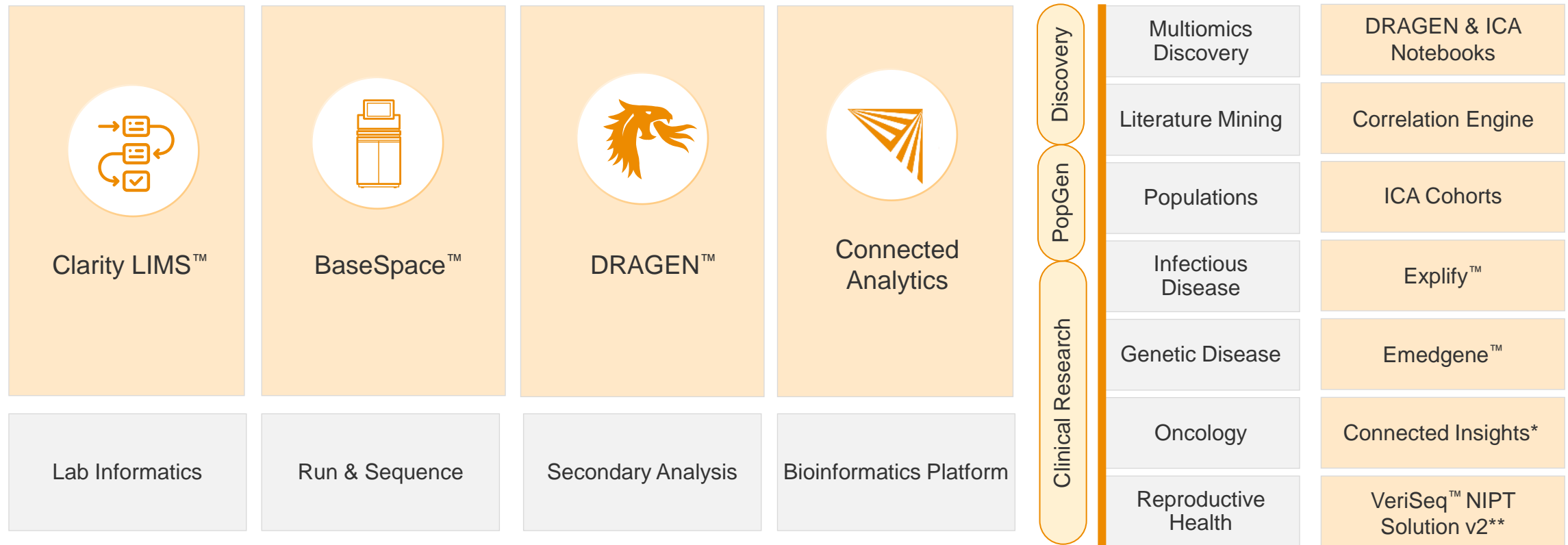
- Support for Telomere-to-Telomere CHM13 v2.0 reference
- DRAGEN Amplicon with CNV and SV support
- DRAGEN Germline with high sensitivity mode
- Sex chromosome low allele frequency variant support
- Imputation for haploid species and sex chromosomes
- Integrated RPIP/UPIP secondary analysis pipelines (Beta)

Efficiency

- Increase ORA compression speed up to 30% when mapping/align step is enabled
- Runtime improvements for joint genotyping pipelines compared to previous release

Connected Software enables both discovery and development

Illumina Connected Software



* Not available in all countries. Illumina Connected Insights supports user-defined tertiary analysis through API calls to third-party knowledge sources.

** For In Vitro Diagnostic Use – Not available in all countries

Thank you

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