Piece together

Illumina solutions for genetic and rare disease

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illumina®

Rare disease is a

The genome holds the solution

There are > 6000 known rare diseases, with more being discovered every year, affecting 2-6% of the global population.[1-3] Individuals suspected of having a rare disease often face a long search for a diagnosis.

Involve multiple tests

×8 Include up to 8 physicians

Can take more than 5 years

Result in 2-3 misdiagnoses

UP TO **80%** OF RARE DISEASES ARE GENETIC

With up to 80% of rare diseases being genetic or having a genetic subtype,[1,2 4-6] it is imperative that we interrogate the genome to find answers and shorten the diagnostic odyssey.

Simplified solutions for complex problems

Illumina offers wide range of NGS solutions, from robust library preparation kits to proven NGS instruments and highly accurate software solutions, that are built to deliver unparalleled insights into heritable genetic disease.

Illumina NGS systems, at the core of every rare disease workflow

Powered by proven NGS technology and sequencing by synthesis (SBS) chemistry, Illumina sequencing systems form the core of an integrated, sample-to-answer workflow. There is an Illumina system to help you answer the toughest questions in rare disease research (Table 1 and Table 2).

TABLE 1
ILLUMINA **MID-** TO **HIGH-**THROUGHPUT SEQUENCING SYSTEMS

About	Simplified flexibility for lower throughputs			Tried-and-true platform for high-performance sequencing			
Flow cell	P1	P2	P3	SP	S1	S2	S4
Flow cells processed per run	1	1	1	1 or 2	1 or 2	1 or 2	1 or 2
Output range	30 Gb	40–120 Gb	60–360 Gb	80–800 Gb	167–1000 Gb	417–2500 Gb	2000–6000 Gb
Run time	10-34 hr	13-44 hr	11-48 hr	13–38 hr	13–25 hr	16–36 hr	< 45 hr
Clusters passing filter per flow cell	100M	400M	1.2B	800M	1.6B	4.1B	10B
Maximum read length	2 × 300 bp	2 × 300 bp	2 × 150 bp	2 × 250 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp

MOST POWERFUL and MOST SUSTAINABLE

NovaSeq X			NovaSeq X Plus		
1.5B	10B	25B	1.5B	10B	25B
1	1	1	1 or 2	1 or 2	1 or 2
165–500 Gb	1000–3000 Gb	8000 Gb	165–1000 Gb	1000–6000 Gb	8000–16,000 Gb
13–21 hr	18–24 hr	~48 hr	13–21 hr	18–24 hr	~48 hr
1.6B	10B	26B	1.6B	10B	26B
2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp	2 × 150 bp

TABLE 2
KEY METHODS BY SYSTEM

Method	NextSeq 1000 and NextSeq 2000	NovaSeq 6000	NovaSeq X Series
WES	●	●	●
Human exomes per run	4-48	40-500	41-750
WGS	--	●	●
Human genomes per run	--	4-48	4-128
Long-read WGS	--	●	●
Human genomes per run	--	4	4-11

WES, whole-exome sequencing; WGS, whole-genome sequencing

NextSeq 1000 and NextSeq 2000 Systems—simplified workflow for various applications

NovaSeq 6000 System—scalable throughput for dynamic volume demands

NovaSeq X Series—breakthrough innovations for groundbreaking discoveries

NextSeq™ 1000 and NextSeq 2000 Systems—simplified workflow for various applications

The NextSeq 1000 and NextSeq 2000 Sequencing Systems offer sequencing power for mid- to high-throughput methods for more moderate sample numbers. They offer innovative design features, DRAGEN™ onboard, an intuitive, simplified workflow, and flexibility of scale for various applications, all in a benchtop system.

NovaSeq™ 6000 System—scalable throughput for dynamic volume demands

The NovaSeq 6000 Sequencing System is a robust, scalable platform that has been adopted by leading hospital, commercial, and academic labs and featured in countless publications. Designed to adapt to your needs, it delivers deep and broad coverage and a flexible sequencing workflow for advanced applications.

NovaSeq X Series—breakthrough innovations for groundbreaking discoveries

The NovaSeq X and NovaSeq X Plus Sequencing Systems provide extraordinary sequencing power to fuel data-intensive methods like WGS, single-cell sequencing, and multiomics. Numerous technical innovations, including XLEAP-SBS™ chemistry and onboard DRAGEN analysis, enable maximum throughput and accuracy to deliver meaningful insights at scale.

Illumina library prep, from targeted panels to the whole

VERSATILE LIBRARY PREP KITS

Illumina offers various library preparation kits, enabling researchers to examine DNA variation from small, targeted regions to the entire genome, including the most challenging sequences.

Library prep kit	TruSight One Sequencing Panels	Illumina DNA with Exome 2.0 Plus Enrichment	Illumina DNA PCR-Free Prep	Illumina Complete Long Read Prep, Human
Method	Targeted enrichment	WES, targeted enrichment	WGS	WGS
Instrument compatibility	NovaSeq 6000, NextSeq 2000 Systems	NovaSeq 6000, NextSeq 2000, NextSeq 1000 Systems	NovaSeq 6000 System	NovaSeq X Series, NovaSeq 6000 System
Hands-on time	~2 hr	~2 hr	~45 min	~6.5 hr
Turnaround time	~6.5 hr	~6.5 hr	~1.5 hr	~8 hr
Input	10-1000 ng	50-1000 ng	25-300 ng	50 ng
Sample types	Genomic DNA (gDNA)	gDNA, blood, saliva	gDNA, blood, saliva	gDNA
Automation available	Yes	Yes	Yes	Automation capable
PCR protocol	Yes	Yes	No	No
Library quant needed?	No	No	No	No
Fragmentation included?	Yes–on bead	Yes–on bead	Yes–on bead	Yes–on bead

WGS

Illumina DNA PCR-Free Prep

Highly accurate WGS without PCR-induced bias to provide highly uniform coverage, even in genomic regions with uneven base composition.

Learn more

Illumina Complete Long Read Prep, Human

Makes accurate and comprehensive WGS easily accessible by enabling both long and short reads on the same instrument, with included DRAGEN on BaseSpace™ Sequence Hub analysis.

Learn more

Targeted enrichment

TruSight™ One Sequencing Panels

Two enrichment panel options target < 6700 disease-associated genes in exonic regions.

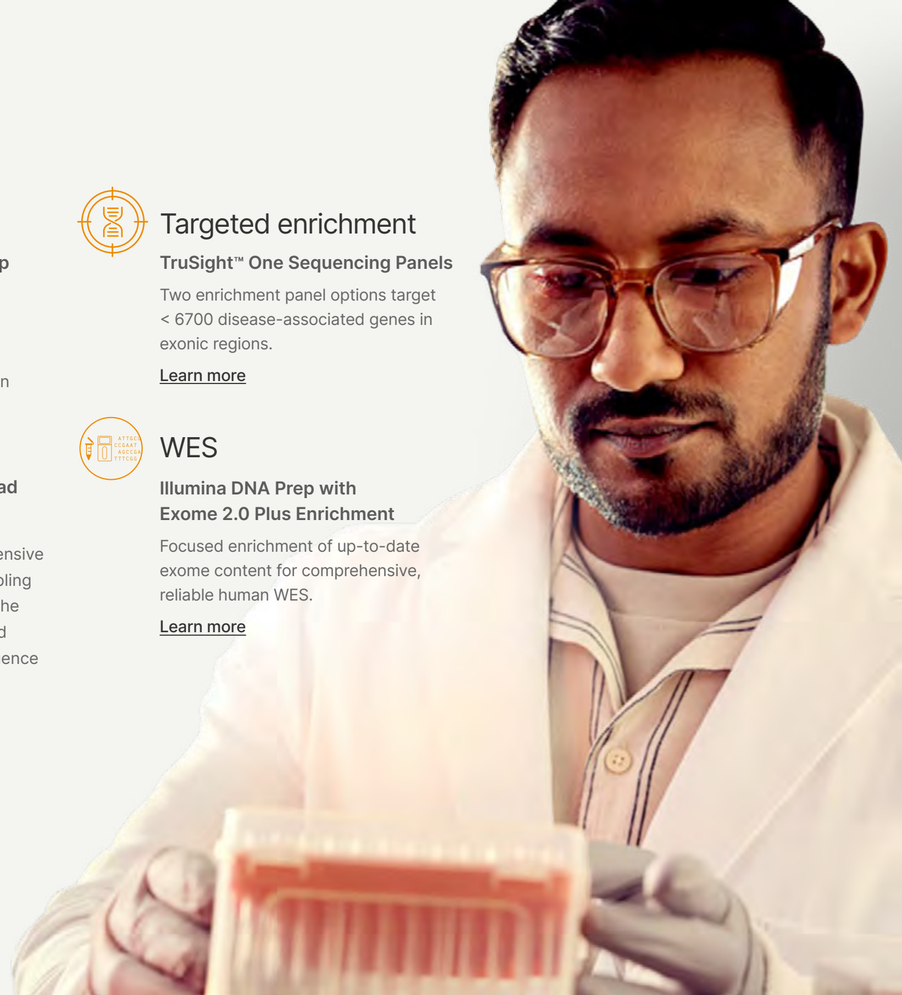
Learn more

WES

Illumina DNA Prep with Exome 2.0 Plus Enrichment

Focused enrichment of up-to-date exome content for comprehensive, reliable human WES.

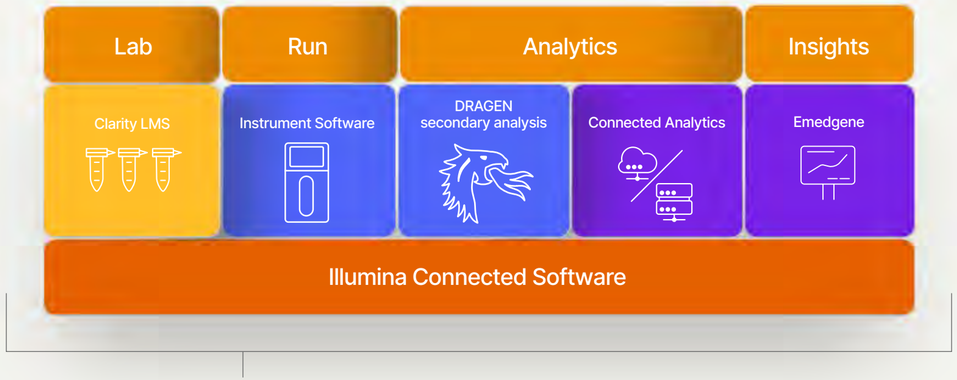
Learn more



Illumina Connected Software—The final piece in finding an answer

OVERVIEW

Illumina offers comprehensive software solutions to help reduce bioinformatics bottlenecks and streamline genomics workflows. Whether a lab is just getting started or in rapid scale mode, Illumina Connected Software unlocks the power of your rare disease data.



Illumina Connected Software

Balancing approachability with customization, Illumina Connected Software offers data analysis, exploration, and management for a variety of bioinformatics expertise levels. From single sample analysis through population-wide studies, Connected Software is seamlessly integrated with Illumina sequencing systems for a highly efficient lab ready to scale.

Simplified lab optimization

Clarity LIMS™ software

Innovative laboratory information management system (LIMS) enabling labs using Illumina sequencing systems to run samples faster, track them easily, and more. It is easy to use, implement, and configure.

Learn more

Accurate secondary analysis

DRAGEN secondary analysis

Highly accurate sequencing by synthesis (SBS) chemistry plus DRAGEN secondary analysis deliver award-winning germline and somatic variant calling.* With onboard DRAGEN analysis available on select instruments, users can gain significant cost savings for accurate, comprehensive, and efficient NGS analysis. DRAGEN secondary analysis pipelines are also available on Illumina cloud platforms and as a server.

Learn more

Explainable AI-powered interpretation

Emedgene™ software

Emedgene software is an explainable artificial intelligence (XAI)–powered platform enabling high-throughput, user-defined interpretation workflows for rare disease research. Delivering a streamlined experience and dramatic reductions in data interpretation time, Emedgene provides a highly configurable and automatable platform for evidence-backed insights.

Learn more

Large-scale data management and analysis

Illumina Connected Analytics

A secure genomics data platform that operationalizes informatics and drives scientific insights. A central component for labs using Illumina sequencing systems, Connected Analytics enables users to build and customize analysis pipelines, execute production workflows at scale, and explore and share data and results.

Learn more

High standards for data privacy

To meet the most stringent security requirements, our software products are built with security and compliance at the core. Data sharing security and governance, audit trails with encryption, and controlled sharing ensure your data are kept safe and secure.

Trusted technology partners

Dedicated to your success, the Illumina Informatics Services team brings a staff of bioinformaticians, data scientists, and designers to help you customize and optimize your analysis workflow and minimize your development burden.

*PrecisionFDA Truth Challenge V2. precision.fda.gov/challenges/10.

INTEGRATED solutions for meaningful insights

Example workflows for genetic disease research:

WES

Focus sequencing efforts on protein-coding regions for variants reported in public databases

Prepare library	Sequence	Analyze	Interpret
			
Illumina DNA Prep with Exome 2.0 Plus Enrichment	NextSeq 2000	DRAGEN Enrichment app	Emedgene

For more information about these products, click the links above.

WGS

Achieve uniform coverage and exceptional variant calling performance

Prepare library	Sequence	Analyze	Interpret
			
Illumina DNA PCR-Free Prep	NovaSeq 6000	DRAGEN Germline app	Emedgene

WGS WITH LONG READS

Call variants in challenging regions, phase haplotypes, and more

Prepare library	Sequence	Analyze	Interpret
			
Illumina Complete Long Read Prep, Human	NovaSeq X	DRAGEN ICLR Analysis in BaseSpace Sequence Hub	Emedgene

For more information about these products, click the links above.

Illumina diagnostic solutions

Illumina genomic in vitro diagnostic (IVD) solutions empower clinical laboratories to find the answers they're looking for to directly impact patient outcomes. Our wide range of products, encompassing the entire NGS workflow, are built to withstand the rigor and precision of the IVD world.

The Illumina IVD instrument portfolio

With easy-to-follow workflows and integrated software, the MiSeq™Dx, NextSeq 550Dx, and NovaSeq 6000Dx instruments deliver accurate, reliable screening and diagnostic testing. They are built with the same proven NGS technology and SBS chemistry as our research use only (RUO) platforms.

MiSeqDx

NextSeq 550Dx

NovaSeq 6000Dx

Sequencing reagent	MiSeqDx Reagent Kit v3	NextSeq 550Dx High-Output Reagent Kit v2.5 (300 cycles)	NextSeq 550Dx High-Output Reagent Kit v2.5 (75 cycles)	NovaSeq 6000Dx S2 Reagent v1.5 Kit (300 cycles)	NovaSeq 6000Dx S4 Reagent v1.5 Kit (300 cycles)
Read length	2 × 300 bp	2 × 150 bp	2 × 75 bp	2 × 150 bp	2 × 150 bp
Clusters passing filter (per flow cell)	25M	400M	400M	4.1B	10B
Maximum output	15 Gb	120 Gb	30 Gb	1 Tb	3 Tb
Run time	< 56 hr	< 35 hr	< 11 hr	≤ 40 hr	≤ 45 hr
Data quality (Q30)	≥ 80%	> 75%	> 80%	> 85%	> 85%

IVD-compliant assays

Illumina offers NGS-based IVD assays and kits enabling clinical labs to apply the power of genomics to genetic disease testing.

TruSight Cystic Fibrosis

A fully integrated molecular testing solution for cystic fibrosis on the MiSeqDx instrument. This assay detects 139 clinically relevant CFTR variants or enables comprehensive sequencing of all protein coding regions of the CFTR gene.

Learn more

Illumina DNA Prep with Enrichment Dx

A library preparation and enrichment solution that is compliant with European Union (EU) IVD Regulation (IVDR) 2017/746. As part of an NGS workflow compatible with any Illumina IVD instrument, it enables clinical labs to add targeted enrichment panels to their menu of diagnostic applications.

Learn more

IVD-specific software

Local Run Manager

Local Run Manager is an integrated user-interface for the MiSeqDx and NextSeq 550Dx Instruments. It is designed to create sequencing runs, monitor run status, analyze data, and view results. The software includes various assay-specific analysis modules.

Learn more

Illumina Run Manager

Integrated with the NovaSeq 6000Dx Instrument, Illumina Run Manager allows users to pair their sequencing instrument with a DRAGEN server to enable automatic launch of secondary analysis pipelines. Various app-based DRAGEN workflows are available for Illumina assays.

DRAGEN for Illumina DNA Prep with Enrichment Dx App

The DRAGEN for Illumina DNA Prep with Enrichment Dx App performs alignment and variant calling in DNA for somatic and germline mutations.

Example IVD-compliant workflows for genetic disease testing

Illumina offers integrated solutions for clinical labs to deliver results that will impact patient outcomes.

Cystic Fibrosis testing

Perform cystic fibrosis screening with comprehensive coverage of known disease-associated variants or the ability to sequence all protein-coding regions and intron/exon boundaries of the CFTR gene.

To learn more, read "Confirming cystic fibrosis diagnosis in newborns using TruSight Cystic Fibrosis"

Flexible support for content

Expand your menu of diagnostic offerings with support for panels that meet the required specifications, including Illumina or third-party fixed and custom panels of varying sizes, such as those designed for WES.

To learn more, read "High-quality variant calling with the NovaSeq 6000Dx instrument"

Summary

The Illumina mission is to improve human health by unlocking the power of the genome. Regardless of the question or scale of the experiment, we offer integrated, comprehensive solutions to empower researchers to make the next great discovery to achieve a greater understanding of rare disease. We are committed to shortening the diagnostic odyssey and developing NGS-based products to usher in a new era of genomics-based healthcare.

Welcome to a world of support

Illumina service and support begin when your Illumina instrument is delivered. Our scientists and engineers are ready to assist with instrument installation and laboratory setup. In addition to onsite support, courses are available to train users on various workflows.

Illumina scientists are available 24 hours a day, five days a week globally to answer questions every step of the way.

Illumina Qualification Services

Meet compliance requirements by taking advantage of Illumina comprehensive qualification services.

Illumina Training

Get high-quality results on Illumina technology even faster with instructor-led, hands-on courses and web-based training options.

Contact Illumina

Contact your Illumina sales representative to find out more about our solutions.

Learn more

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illumina®

No rare disease will go unseen.

We are always available for questions, insights, and conversation.

Visit us at illumina.com

M-GL-01413 v1.0

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