Empowering genomics-based diagnostics

Illumina Dx platforms and solutions

illumina®

A decade of innovation

At Illumina, we are committed to improving human health by unlocking the power of the genome. For more than a decade, we've brought our unparalleled expertise as a genomics industry leader to revolutionize health care through next-generation sequencing (NGS)-based diagnostics.

Illumina has led the way since 2013, with the launch of the MiSeq™ Dx Instrument, the first FDA*-regulated, CE-marked NGS instrument for *in vitro* diagnostic (IVD) use. We continued to innovate, building regulated NGS platforms that include the NextSeq™ 550Dx and NovaSeq™ 6000Dx Instruments, and developing novel IVD assays across areas of interest.

Illumina genomic IVD solutions empower clinical laboratories to find the answers they're looking for to improve patient outcomes. Our wide range of products, from novel NGS and microarray assays to robust instruments and accurate data analysis solutions, are built to withstand the rigor and precision of the IVD world.

Together, Illumina diagnostic solutions are ushering in a new era of genomics-based health care.

2013

MiSeqDx Instrument

Cystic Fibrosis Assay

2017

NextSeq 550Dx Instrument

VeriSeq NIPT Solution

RAS Panel Companion Diagnostic

2021

Illumina COVIDSeq Test

2022

NovaSeq 6000Dx Instrument

TruSight Oncology Comprehensive

Illumina DNA Prep with Enrichment Dx

* FDA, Food and Drug Administration


NovaSeq™ 6000Dx
illumina

Regulated NGS instruments for every clinical lab

With easy-to-follow workflows and integrated software, the MiSeqDx,[1] NextSeq 550Dx,[2] and NovaSeq 6000Dx[3] Instruments deliver accurate, reliable screening and diagnostic testing. All are based on proven Illumina NGS technology and sequencing by synthesis (SBS) chemistry (Figure 1, Table 1). Using these Instruments, clinical labs can develop, validate, and perform their own NGS tests, run Illumina or third-party assays, and pursue various clinical research applications in Research (RUO) Mode (Figure 2).

illumina.com/miseqdx

illumina.com/nextseq550dx

illumina.com/novaseq6000dx

Figure 1: The Illumina IVD system portfolio

For *In Vitro* Diagnostic Use. Not available in all regions and countries.

Table 1: Illumina IVD instrument specification

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

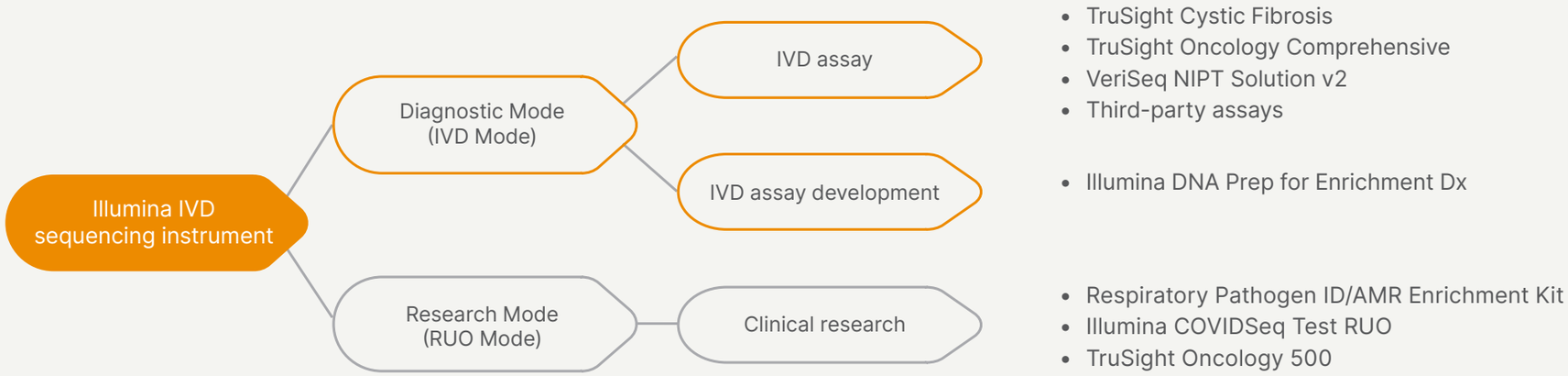

Figure 2: Illumina IVD solution flexibility

 For *In Vitro* Diagnostic Use. Not available in all regions and countries.

Plan. Monitor. Analyze.

Illumina offers a suite of informatics software that can be accessed via streamlined user interfaces to create and monitor sequencing runs, analyze data, and view results (Figure 3, Table 2).

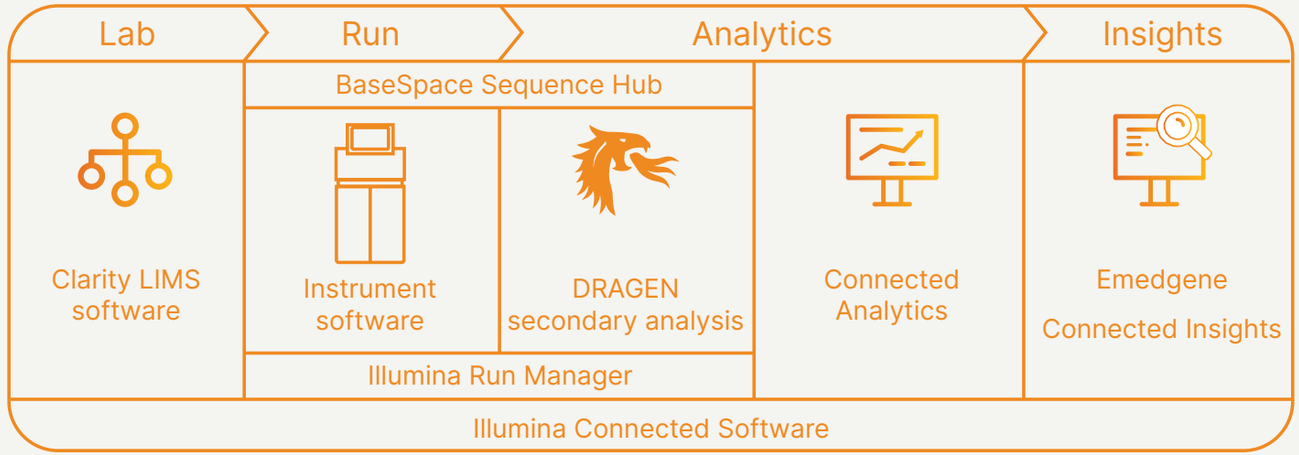

Figure 3: Illumina Connected Software

Table 2: Illumina informatics options for Dx instruments

	Instrument mode	MiSeqDx Instrument	NextSeq 550Dx Instrument	NovaSeq 6000Dx Instrument
Informatics software		Local Run Manager	Local Run Manager Illumina Run Manager DRAGEN Server	Illumina Run Manager DRAGEN Server
Compatible with DRAGEN Server	IVD	-	✓	✓
Connected with BaseSpace Sequence Hub	RUO	-	-	✓
Connected with Illumina Connected Analytics	RUO	-	-	✓
Compatible with Clarity LIMS software	RUO	✓	✓	✓
Compatible with Emedgene software	RUO	-	✓	✓
Compatible with Connected Insights	RUO	✓	✓	✓

IVD mode–specific software

Local Run Manager

Local Run Manager software is an integrated solution designed to create sequencing runs, monitor run status, analyze sequencing data, and view results. The software integrates with instrument control software and includes various analysis modules for different assays.

Illumina Run Manager

Illumina Run Manager allows users to pair sequencing instruments with a DRAGEN™ server and enables automatic launch of secondary analysis. The software provides parallel workflows for sequencing and analysis to serve labs with quick turnaround time. Various app-based DRAGEN secondary analysis workflows are available for Illumina assays.

Lab optimization

Clarity LIMS™ software

Clarity LIMS software is an innovative laboratory information management system (LIMS) that enables labs using Illumina sequencing systems to run samples faster, track them easily, and achieve or maintain regulatory compliance efficiently. It is easy to use, implement, and configure.

For *In Vitro* Diagnostic Use. Not available in all regions and countries.

Research capabilities

Large-scale data management and analysis (RUO)

BaseSpace Sequence Hub

BaseSpace Sequence Hub is a research use only (RUO) genomics cloud-computing platform designed to provide simplified data management and analytical sequencing tools in a user-friendly format. It provides flexibility and convenience with an array of tools, enabling users to set up and monitor runs, access and analyze data stored in the cloud with a curated set of analysis apps, and securely share data with collaborators.

Illumina Connected Analytics

Illumina Connected Analytics is a secure genomics data platform to operationalize informatics and drive 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.

Secondary analysis (RUO/IVD)

DRAGEN Server

DRAGEN secondary analysis provides accurate, efficient, and comprehensive genomic data analysis solutions to address common challenges, including lengthy compute times and massive volumes of data. When paired with an IVD sequencing instrument through Illumina Run Manager, a DRAGEN Server enables clinical labs to analyze NGS data accurately and efficiently, using various applications designed for specific IVD assays.

Tertiary analysis (RUO)

Emedgene™ software

Emedgene software is an explainable artificial intelligence (XAI)–powered genomic analysis platform enabling high throughput interpretation. Delivering dramatic reductions in analysis time, Emedgene software provides a highly configurable, streamlined platform for generating evidence-backed research insights.

Connected Insights

Illumina Connected Insights is a cloud-based platform enabling tertiary analysis through API*–based calls. The platform allows labs to streamline interpretation and reporting and scale up their NGS operations by providing user-defined, automatable workflows and powerful knowledge sources for filtering for a diverse range of research application areas.

* API, application programming interface

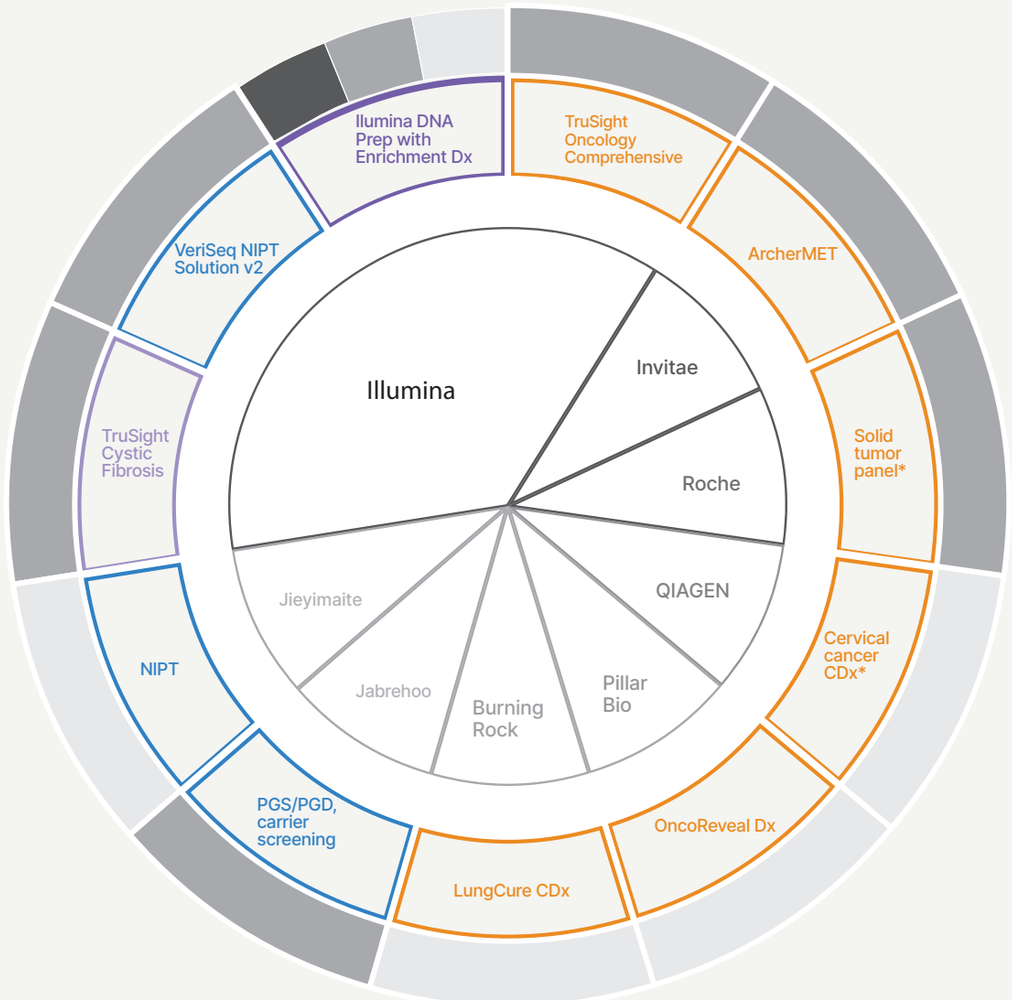

Figure 4: Menu of Illumina and partner IVD assays

IVD innovation. Expanded.

Illumina offers NGS-based IVD assays and kits enabling clinical laboratories to apply the power of genomics to noninvasive prenatal testing (NIPT), rare genetic disease testing, oncology testing, and more (Figure 4, Table 3). We are working with more and more partners to develop clinical assays to address the world's greatest health challenges (Figure 4).

Disease area or assay development

- Rare genetic disease
- Reproductive health
- Assay development
- Oncology

Illumina sequencing platform

- MiSeqDx instrument
- NextSeq 550Dx instrument
- NovaSeq 6000Dx instrument

*Assay in development.

†EUA approved in certain countries.



Table 3: Supported IVD assays at a glance

Disease area or assay development	Assay	Manufacturer	MiSeqDx Instrument	NextSeq 550Dx Instrument	NovaSeq 6000Dx Instrument	Regulatory approval
Genetic disease	TruSight Cystic Fibrosis	Illumina	✓			US, CE
Genetic disease	PrismGuide IRD Panel System	Sysmex	✓			Japan
Reproductive health	VeriSeq NIPT Solution v2	Illumina		✓		CE
Oncology	TruSight Oncology Comprehensive	Illumina		✓		US, CE, Japan
Assay development	Illumina DNA Prep with Enrichment Dx	Illumina	✓	✓	✓	US, CE
Oncology	oncoReveal Dx	Pillar	✓			US, CE, China
Oncology	ArcherMET	Invitae	✓			Japan
Oncology	OncoGuide NCC Oncopanel System	Sysmex		✓		Japan
Reproductive health	VeriSeq PGS	Jabrehoo	✓			China
Oncology	Essencare	Geneseeq	✓			China
Oncology	LungCure CDx	Burning Rock	✓			China
Oncology	*BRCA1* and *BRCA2*	AmoyDx	✓			China, CE
Oncology	*EGFR*/*KRAS*/*BRAF*/*PIK3CA*/*ALK*/*ROS1*	Geneis	✓			China
Oncology	*KRAS*/*NRAS*/*BRAF*/*PIK3CA*	Genecast	✓			China
Oncology	Onco Core	3DMed	✓	✓		China

Supported assays in this table are current as of the publication (June 2025) of this document and may be subject to change.

Turn data into answers

GENETIC DISEASE TESTING

TruSight™ Cystic Fibrosis

A fully integrated molecular testing solution for cystic fibrosis on the MiSeqDx Instrument. The CF 139-Variant 2.0 and CF Clinical Seq 2.0 Local Run Manager analysis modules enable detection of 139 clinically relevant *CFTR* variants or comprehensive sequence information for all protein coding regions in the *CFTR* gene, respectively.[4]

ASSAY DEVELOPMENT

Illumina DNA Prep with Enrichment Dx

A library preparation and enrichment solution that is compliant with European Union *In Vitro* Diagnostic Regulation 2017/746. As part of an NGS workflow on the MiSeqDx, NextSeq 550Dx, and NovaSeq 6000Dx Instruments, it enables clinical labs to add targeted sequencing enrichment panels to their menu of diagnostic applications. DRAGEN for Illumina DNA Prep with Enrichment Dx App on NovaSeq 6000Dx Instrument performs alignment and variant calling in DNA for somatic and germline mutations.[8]

ONCOLOGY TESTING

TruSight Oncology Comprehensive

TruSight Oncology Comprehensive is a qualitative *in vitro* diagnostic test that uses targeted next-generation sequencing to detect variants in 517 genes using nucleic acids extracted from formalin-fixed, paraffin-embedded (FFPE) tumor tissue samples from cancer patients with solid malignant neoplasms using the Illumina NextSeq 550Dx Instrument. The test can be used to detect single nucleotide variants, multinucleotide variants, insertions, and deletions from DNA, and fusions in 24 genes and splice variants in one gene from RNA. The test also reports a Tumor Mutational Burden (TMB) score.[6]

NIPT

VeriSeq™ NIPT Solution v2

An IVD test that uses PCR-free, whole-genome sequencing to expand prenatal screening to all rare autosomal aneuploidies, sex chromosome aneuploidies, and partial duplications and deletions ≥ 7 Mb for all autosomes. VeriSeq NIPT Assay Software v2 automatically performs data analysis and generates a report that provides qualitative results.[5]

*This test is not available in all countries or regions. COVIDSeq is only available to authorized laboratories under Emergency Use Authorization or similar authorization for the detection of SARS-CoV02 only–not for other viruses or pathogens. This test is only authorized for the duration of the emergency declaration for COVID-19.

For *In Vitro* Diagnostic Use. Not available in all regions and countries.

IVD test options

When planning IVD assays, there are three approaches to consider

1 End-to-end workflow

IVD

More traditional IVD system, eg, VeriSeq NIPT Solution v2

2 Platform (in development)

IVD

Develop and validate your own assay

3 Built from components

IVD | IVD | RUO | IVD | IVD | IVD

Choose individual components to build your own assay

Clinical research in RUO Mode

The MiSeqDx, NextSeq 550Dx, and NovaSeq 6000Dx Instruments feature RUO Mode, enabling labs to pursue various clinical research applications (Table 4).

Table 4: Supported clinical research panels in RUO mode at a glance

Disease area	Assay	MiSeqDx instrument	NextSeq 550Dx instrument	NovaSeq 6000Dx instrument
Oncology	TruSight Tumor 170		✓	
Oncology	TruSight Oncology 500		✓	
Oncology	TruSight Oncology 500 High-Throughput			✓
Oncology	TruSight Hereditary Cancer	✓	✓	
Oncology	TruSight RNA Pan Cancer	✓	✓	
Oncology	TruSight RNA Fusion	✓	✓	
Genetic disease	Illumina DNA Prep with Exome 2.0 Plus Enrichment		✓	✓
Genetic disease	TruSight One/TruSight One Expanded		✓	✓
Infectious disease	Respiratory Pathogen ID/AMR Enrichment Panel	✓	✓	
Infectious disease	Urinary Pathogen ID/AMR Enrichment Kit	✓	✓	
Infectious disease	COVIDSeq Assay	✓		

Supported assays in this table are current as of the publication (June 2025) of this document and may be subject to change.

Empowering clinical sequencing around the globe

Illumina offers trusted solutions, with NGS-based IVD assays and sequencing instruments installed in over 55 countries worldwide (Figure 5). Powered by Illumina SBS chemistry, our solutions deliver high-quality data with the accuracy and reliability needed for diagnostic testing.

Comprehensive genomic profiling using NGS can identify more clinically relevant variants than conventional testing approaches and dramatically shorten the diagnostic odyssey for patients.[10]

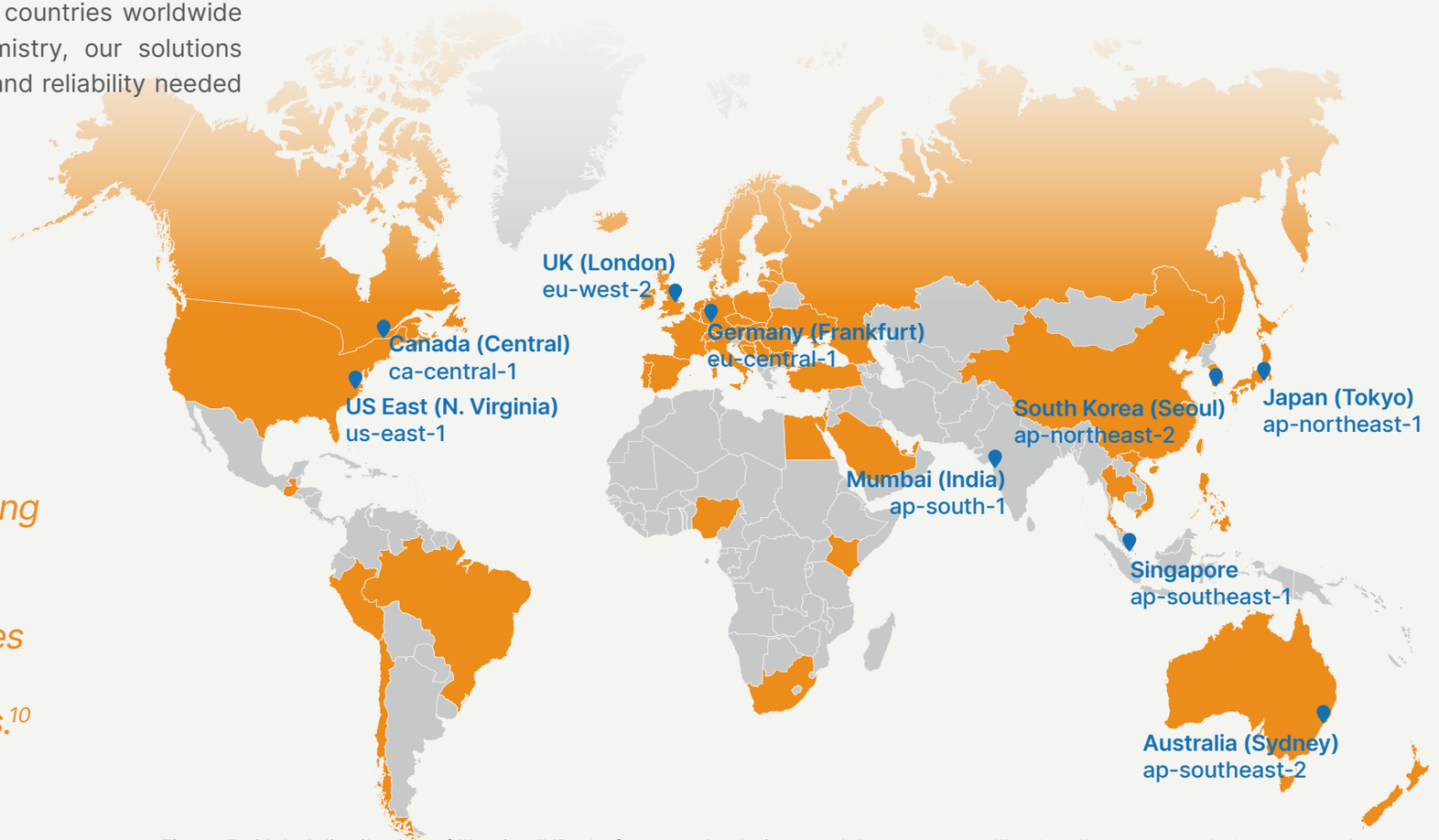

Figure 5: Global distribution of Illumina IVD platforms and solutions and data centers—Illumina diagnostics solutions are in place in countries around the globe (shaded in orange). Deployment of regional data centers via Amazon Web Services (gray) enables data to be stored regionally in accordance with local data protection laws and regulations.

Welcome to a world of support

Illumina service and support begins 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.

Contact your Illumina sales representative to find out more about our diagnostic solutions.
illumina.com/systems/ivd-instruments.html

illumina®

We are always available for questions, insights, and conversation.
Visit us at illumina.com.

1.800.809.4566 toll-free (US) | +1.858.202.4566 tel

For *In Vitro* Diagnostic Use. Not available in all regions and countries.

Illumina qualification services

Meet compliance requirements by taking advantage of Illumina comprehensive qualification services at www.illumina.com/services/instrument-services-training/qualification-services.html.

Illumina training

Get high-quality results on Illumina technology even faster with instructor-led, hands-on courses and web-based training options at www.illumina.com/services/instrument-services-training/training.html.

References

1. Illumina. MiSeqDx Instrument. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/miseqdx-instrument-data-sheet-m-na-00005/miseqdx-instrument-data-sheet-m-na-00005.pdf. Published 2024. Accessed June 18, 2025.
2. Illumina. NextSeq 550Dx Instrument. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/nextseq-550-dx-spec-sheet-m-gl-00035/nextseq-550-dx-instrument-spec-sheet-m-gl-00035.pdf. Published 2025. Accessed June 18, 2025.
3. Illumina. NovaSeq 6000Dx Instrument. illumina.com/systems/ivd-instruments/novaseq-6000dx.html. Accessed June 18, 2025.
4. Illumina. TruSight Cystic Fibrosis. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/trusight-cystic-fibrosis-data-sheet-m-gl-00145/trusight-cystic-fibrosis-data-sheet-m-gl-00145.pdf. Published 2022. Accessed June 18, 2025.
5. Illumina. VeriSeq NIPT Solution v2. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/veriseq-nipt-solution-v2-m-apj-00036/veriseq-nipt-solution-v2-m-apj-00036.pdf. Published 2024. Accessed June 18, 2025.
6. Illumina. Illumina COVIDSeq Test Instructions for Use. support.illumina.com/content/dam/illumina-support/documents/documentation/chemistry_documentation/Illumina-COVIDSeq-Test/illumina-covidseq-test-instructions-for-use-1000000128490-03.pdf. Published January 2022. Accessed June 18, 2025.
7. Illumina. Illumina DNA Prep with Enrichment Dx. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/illumina-dna-prep-enrichment-dx-data-sheet-m-gl-00743/illumina-dna-prep-enrichment-dx-data-sheet-m-gl-00743.pdf. Published 2024. Accessed June 18, 2025.
8. Illumina. TruSight Oncology Comprehensive. illumina.com/content/dam/illumina/gcs/assembled-assets/marketing-literature/trusight-oncology-comprehensive-eu-data-sheet-m-gl-00020/trusight-oncology-comprehensive-ce-ivd-data-sheet-m-emea-00069.pdf. Published 2024. Accessed June 18, 2025.

M-GL-00769 v3.0