

# Empowering genomics-based diagnostics

Illumina Dx platforms and solutions



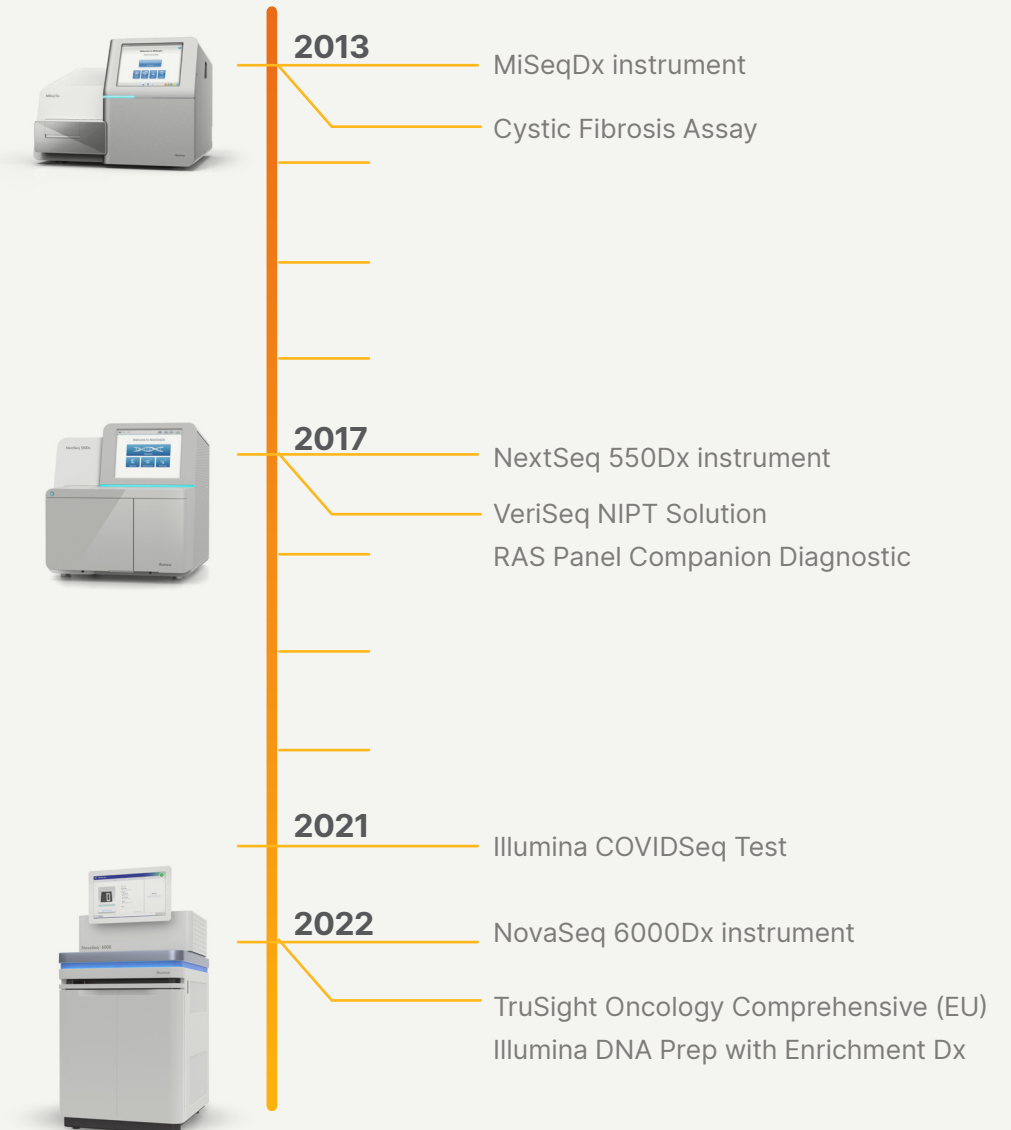
# A decade of innovation

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 healthcare 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 continue 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 healthcare.





NovaSeq™ 6000Dx

illumina



# Regulated NGS instruments for every clinical lab

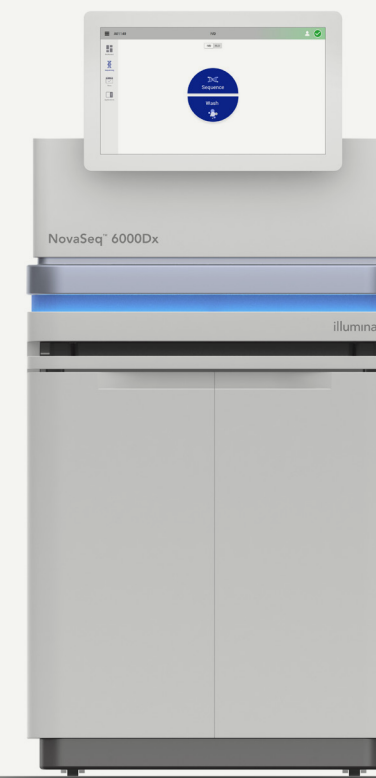
With easy-to-follow workflows and integrated software, the MiSeqDx,<sup>1</sup> NextSeq 550Dx,<sup>2</sup> and NovaSeq 6000Dx<sup>3</sup> instruments deliver accurate, reliable screening and diagnostic testing. All are based on our proven NGS technology and sequencing by synthesis (SBS) chemistry (Figure 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](https://illumina.com/miseqdx)



[illumina.com/nextseq550dx](https://illumina.com/nextseq550dx)



[illumina.com/novaseq6000dx](https://illumina.com/novaseq6000dx)

Figure 1: The Illumina IVD system portfolio

Table 1: Illumina IVD instrument specification

	Sequencing reagent	Read length	Clusters passing filter (per flow cell)	Maximum output	Run time	Data quality (Q30)
MiSeqDx	MiSeqDx Reagent Kit v3	2 × 300 bp	25 million	15 Gb	< 56 hrs	≥ 80%
NextSeq 550Dx	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	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%

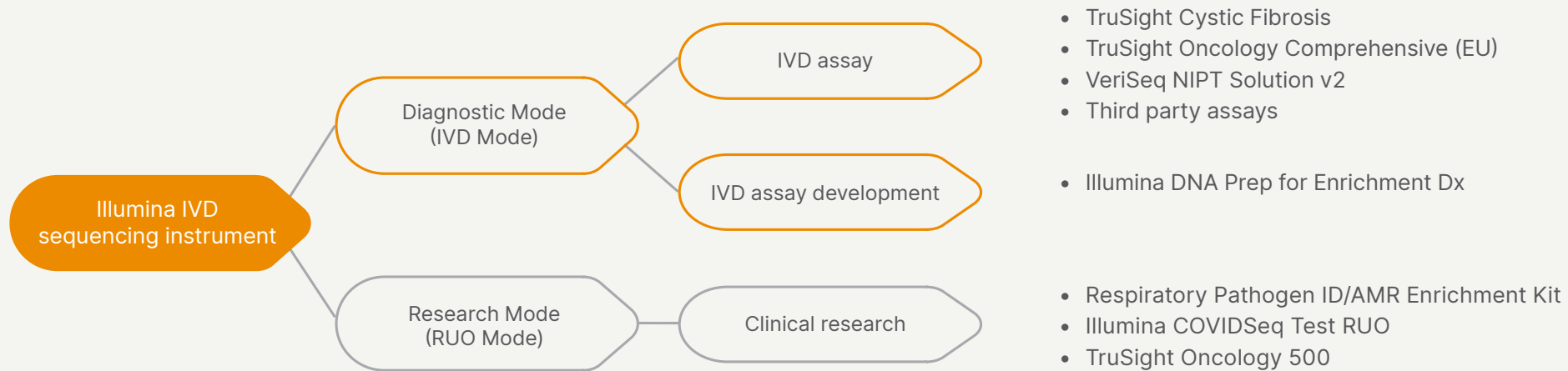
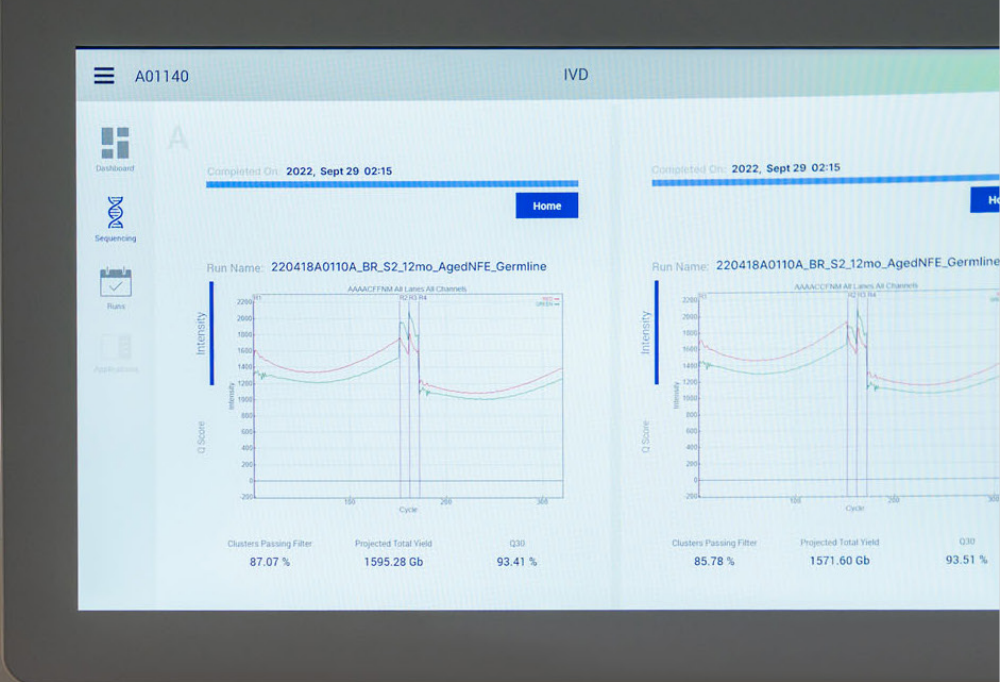


Figure 2: Illumina IVD solution flexibility



NovaSeq™ 6000Dx

# 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).

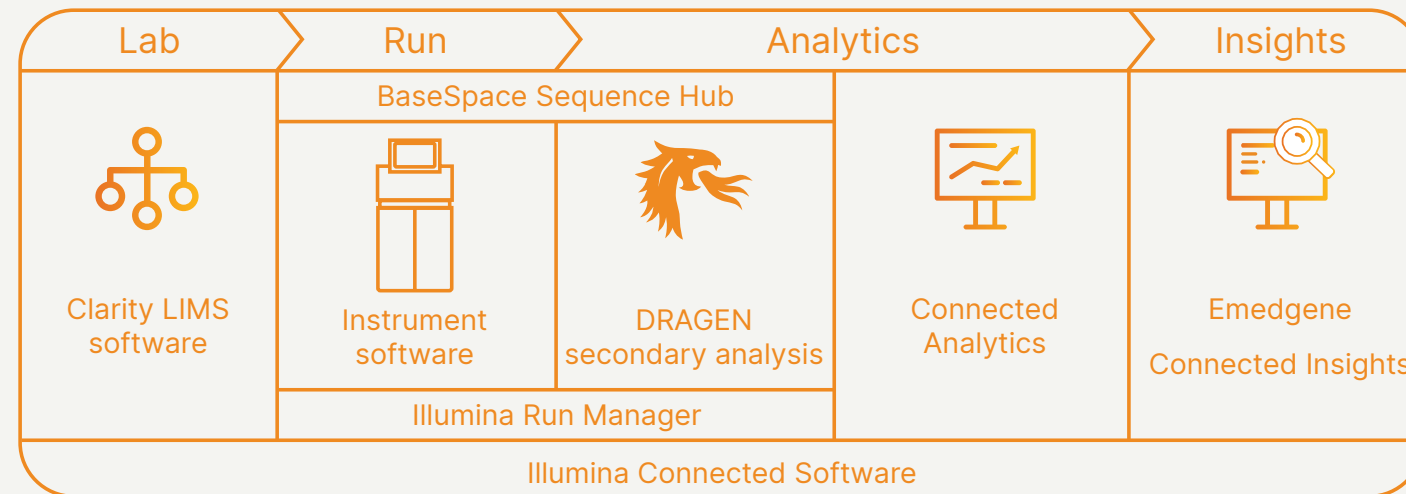


Figure 3: Illumina Connected Software

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
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	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 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.







## 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 (in development).

### Tertiary analysis

#### Emedgene (RUO)

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

# 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
- Infectious disease
- Assay development
- Oncology

\*Assay in development

†EUA approved in certain countries

## Illumina sequencing platform

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

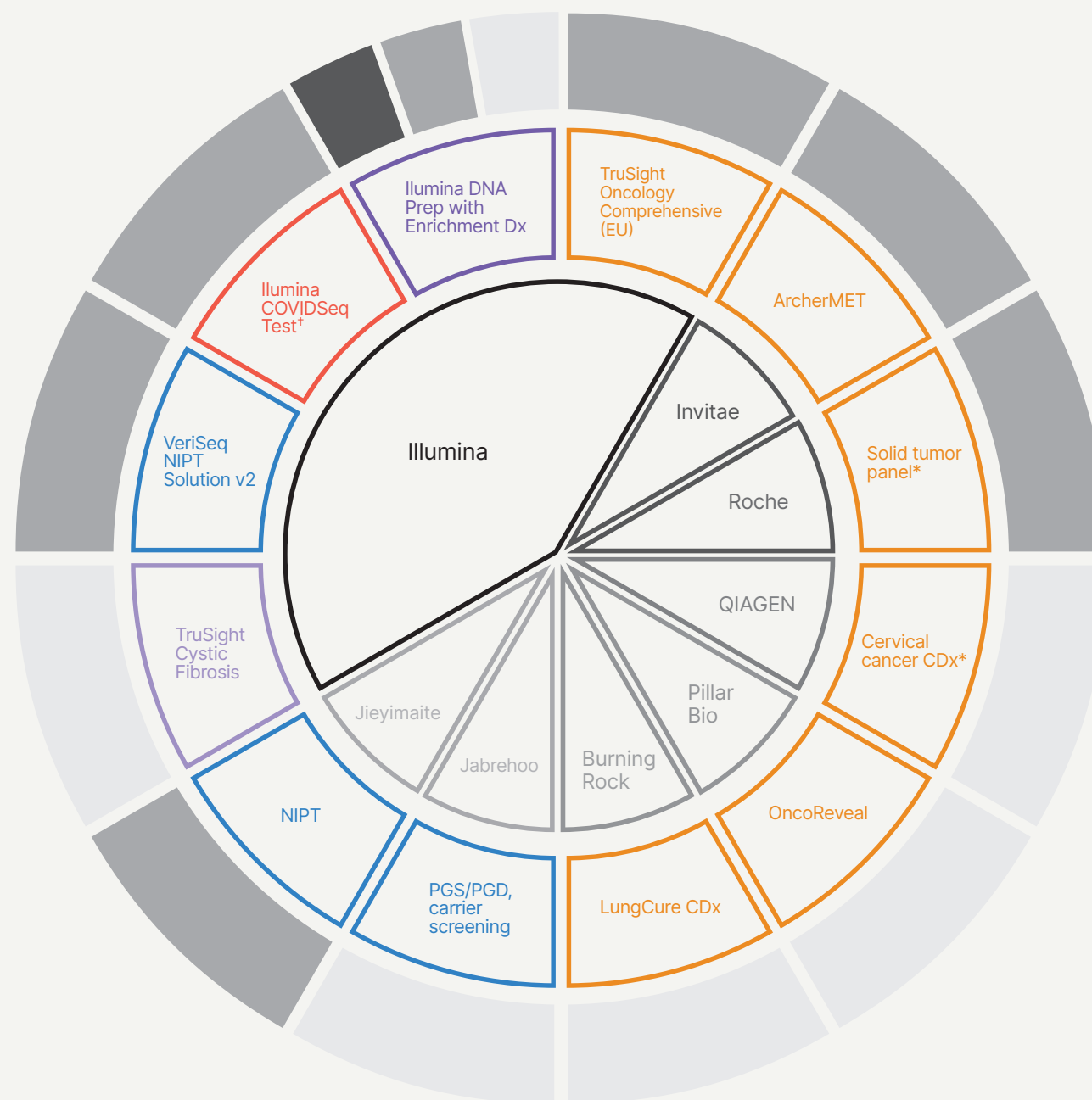


Figure 4: Menu of Illumina and partner IVD assays



Table 3: Supported IVD assays at a glance

Disease area or assay development	Assay	Manufacturer	MiSeqDx	NextSeq 550Dx	NovaSeq 6000Dx	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
Infectious disease	Illumina COVIDSeq Test	Illumina		✓		US (EUA)
Oncology	TruSight Oncology Comprehensive	Illumina		✓		CE
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	Geneseq	✓			China
Oncology	LungCure CDx	Burning Rock	✓			China
Oncology	BRCA1 and BRCA 2	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 of this document and may be subject to change.

# Turn data into answers

## GENETIC DISEASE TESTING

### TruSight™ Cystic Fibrosis

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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.<sup>4</sup>

## NIPT

### VeriSeq™ NIPT Solution v2

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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  $\geq 7$  Mb for all autosomes. VeriSeq NIPT Assay Software v2 automatically performs data analysis and generates a report that provides qualitative results.<sup>5</sup>

## ONCOLOGY TESTING

### TruSight Oncology Comprehensive (EU)

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The first CE-marked IVD, kitted solution for comprehensive genomic profiling (CGP) of patient samples. It enables detection of DNA and RNA variants and biomarker signatures, such as tumor molecular burden (TMB) and microsatellite instability (MSI). TruSight Oncology Comprehensive (EU) analysis module performs base calling and QC, variant calling, and interpretation.<sup>6</sup>

## INFECTIOUS DISEASE TESTING

### Illumina COVIDSeq™ Test (EUA)\*

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This amplicon-based, NGS test detects SARS-CoV-2 in patients suspected of COVID-19. The kit includes steps for viral RNA extraction, library prep, and sequencing. The DRAGEN COVIDSeq Test Pipeline is used for data analysis.<sup>7</sup>

## ASSAY DEVELOPMENT

### Illumina DNA Prep with Enrichment Dx

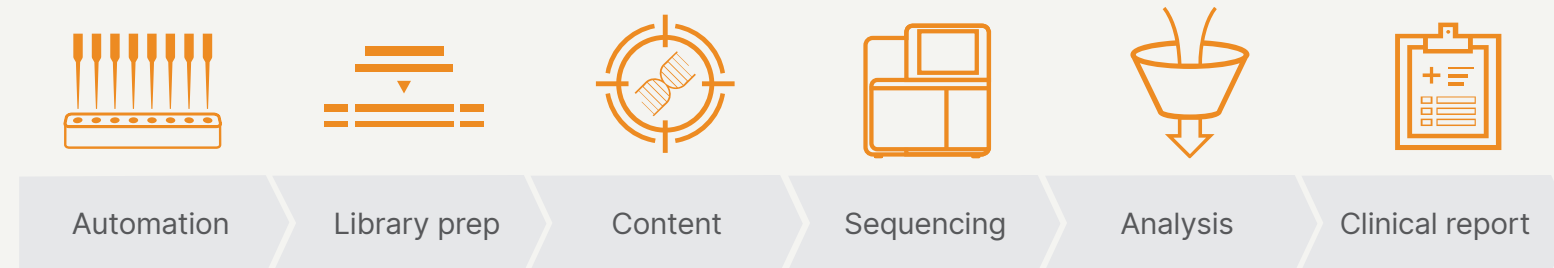
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A library preparation and enrichment solution that is compliant with EU IVDR 2017/746. As part of an NGS workflow on the MiSeqDx, NextSeq 550Dx, and NovaSeq 6000Dx instruments, it enables clinical labs to add targeted sequencing enrichment panels to 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.<sup>8</sup>

\*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-CoV2 only—not for other viruses or pathogens. This test is only authorized for the duration of the emergency declaration for COVID-19.

# IVD test options

When planning IVD assays, there are three approaches to consider



1 End to end workflow



More traditional IVD system, eg, VeriSeq NIPT Solution v2.

2 Platform (in development)



Develop and validate your own assay.

3 Built from components



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 clinical 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	NextSeq 550Dx	NovaSeq 6000Dx
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 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.<sup>10</sup>*

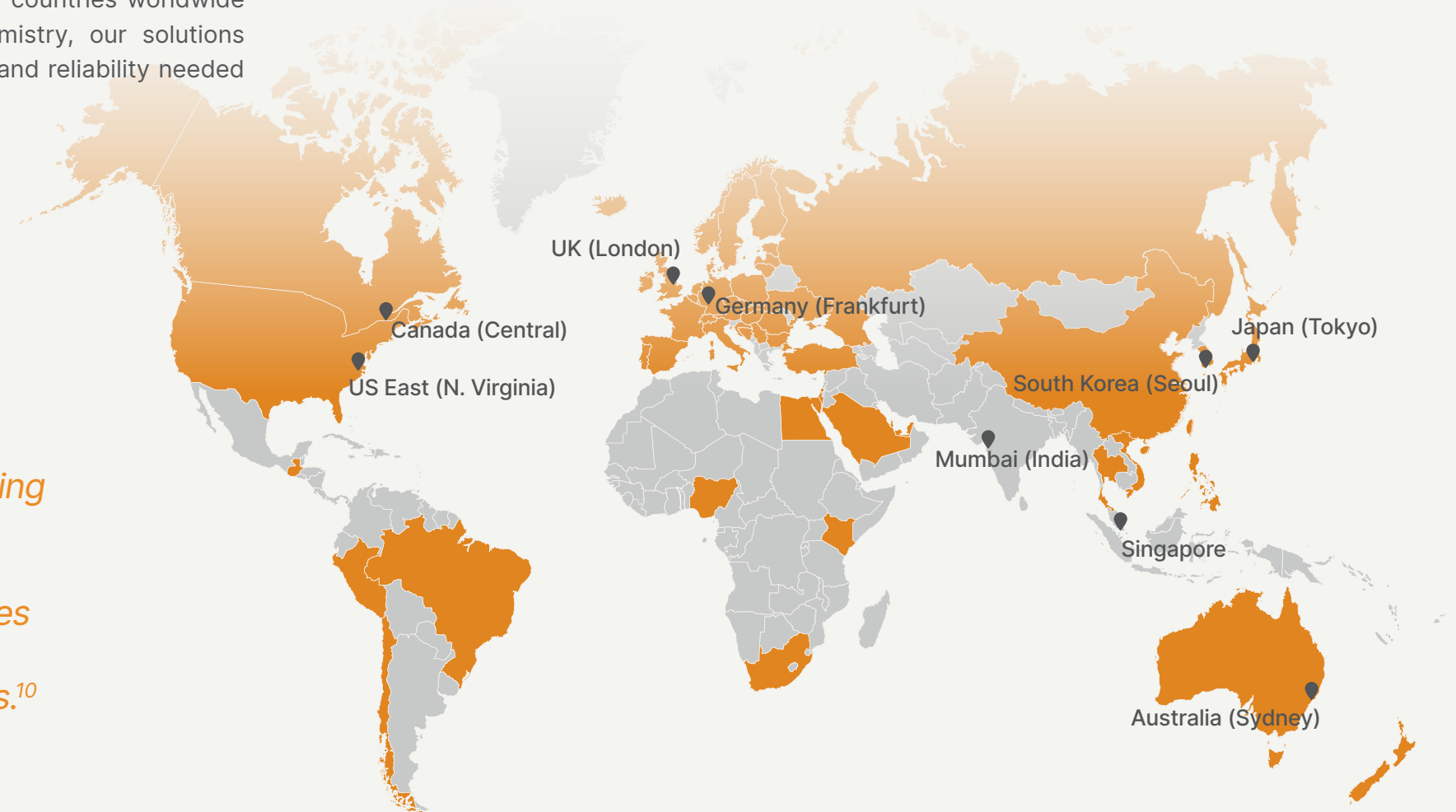


Figure 5: Global distribution of Illumina IVD platforms and solutions and data centers—Illumina diagnostics solutions are in place in countries around the globe (shaded in orange). Deployment of regional data centers via Amazon Web Services (grey) enables data to be stored regionally in accordance with 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](https://www.illumina.com/systems/ivd-instruments.html)



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

Visit us at [illumina.com](https://www.illumina.com).

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



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## 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](https://www.illumina.com/services/instrument-services-training/training.html).

## References

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