SPECIFICATION SHEET

MiSeq™ Dx Instrument

First FDA-regulated, CE-marked IVD NGS instrument for *in vitro* diagnostic use

- Simple instrument operation with an intuitive touch screen interface and an automated workflow
- Exceptional data quality and reliability demonstrated through extensive system verification
- Wide menu of molecular diagnostic assays designed for clinical laboratory environments
- Open platform for custom IVD assay development and option to run other assays in Research Mode

illumina®

For *In Vitro* Diagnostic Use. Not available in all regions and countries and not available for purchase in the Federal Republic of Germany.

Introduction

The MiSeqDx Instrument is the first Food and Drug Administration (FDA)–regulated and Conformité Européene (CE) marked *in vitro* diagnostic (IVD) platform for next-generation sequencing (NGS) (Figure 1). Designed specifically for the clinical laboratory environment, the MiSeqDx Instrument offers a small footprint (0.3 square meters), an easy-to-use workflow, and data output tailored to the diverse needs of clinical labs. Additionally, the on-instrument integrated software enables run setup, sample tracking, user management, audit trails, and results interpretation.* Taking advantage of proven Illumina sequencing by synthesis (SBS) chemistry, the MiSeqDx Instrument enables accurate, reliable screening and diagnostic testing.

Figure 1: MiSeqDx Instrument—The FDA-regulated, CE-marked IVD MiSeqDx Instrument offers a simple workflow, user-friendly software interface, and enhanced user security.

* Detailed results reports are available for target-specific assays, such as the TruSight Cystic Fibrosis 139-Variant Assay and TruSight Cystic Fibrosis Clinical Sequencing Assay.

The NGS advantage

Compared to capillary electrophoresis–based Sanger sequencing, NGS can detect a broader range of DNA variants, including low-frequency variants and adjacent phased variants, with a faster time to result and fewer hands-on steps.[1,2] Illumina SBS chemistry employs natural competition among all four labeled nucleotides, which reduces incorporation bias and allows more robust sequencing of repetitive regions and homopolymers compared to other sequencing systems.[3] Comprehensive results are delivered quickly, eliminating the need for time-consuming reflex testing.

Simple, three-step workflow

Assays run on the MiSeqDx Instrument follow a simple, three-step process (Figure 2) that starts with genomic DNA (gDNA) extracted from human peripheral whole blood specimens or formalin-fixed, paraffin-embedded (FFPE) tissues. DNA samples are prepared for sequencing through the addition of primers, generating indexed libraries for simultaneous capture and amplification of hundreds of targeted regions in multiple samples.

Prepared libraries are sequenced with a ready-to-use, prefilled MiSeqDx reagent cartridge. Simply thaw the cartridge, load the library, insert into the MiSeqDx Instrument, and start sequencing with the push of a button.

NGS on the MiSeqDx Instrument uses Illumina SBS chemistry, in which massively parallel sequencing of millions of DNA fragments occurs by a proprietary reversible terminator–based method. Single bases are detected as they are incorporated into growing DNA strands. Base calls are made directly from signal intensity measurements during each cycle.

Learn more about SBS chemistry at illumina.com.

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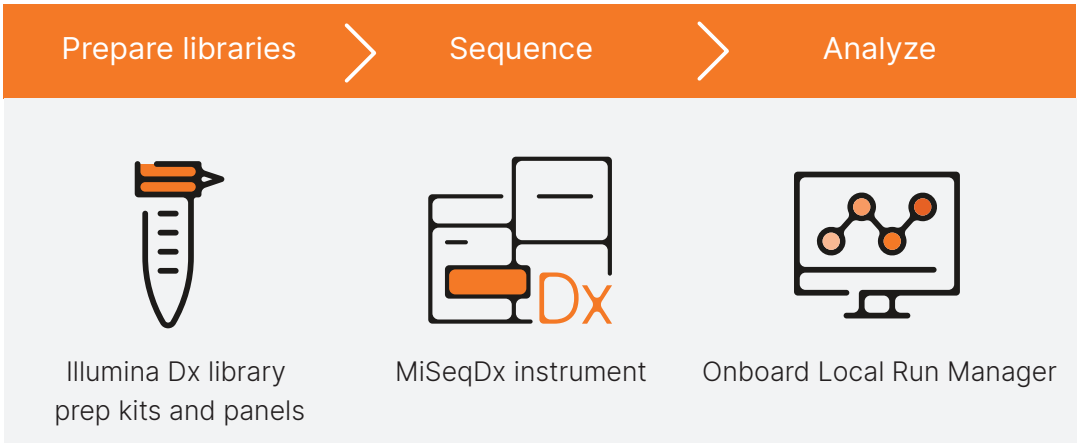

Figure 2: Three-step MiSeqDx assay process—The MiSeqDx Instrument is part of an integrated solution that includes library preparation and data analysis for molecular diagnostic assays. Detailed results reports are only available with target-specific assays, such as the TruSight Cystic Fibrosis 139-Variant Assay and TruSight Cystic Fibrosis Clinical Sequencing Assay.

Integrated system software

The MiSeqDx Instrument offers fully integrated onboard instrument software that can be accessed through a user-friendly touch screen interface. Sequencing runs can be planned and tracked with audit trails using the Local Run Manager software, which supports library tracking and specification of sequencing run parameters. The Local Run Manager software runs on the instrument computer, allowing users to monitor run progress and view analysis results from other computers connected to the same network. After sequencing is completed, Local Run Manager automatically starts data analysis using one of several available analysis modules. Assay-specific analysis modules are available to perform alignment and variant calling of specific variants or across user-defined targeted regions.

User management software

To ensure proper system use, the MiSeqDx Instrument is equipped with an integrated user management system. This enables laboratories to control and trace system access, ensuring that only authorized personnel are running tests.

Available assays and reagents

Multiple *in vitro* diagnostic (IVD) assays and reagents are currently available for use on the MiSeqDx Instrument:†

- **TruSight™ Cystic Fibrosis 139-Variant Assay** detects 139 clinically relevant and functionally verified variants in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene, as defined by the CFTR2 database[4]
- **TruSight Cystic Fibrosis Clinical Sequencing Assay** detects mutations within the protein coding regions and intron/exon boundaries of the *CFTR* gene
- **Illumina DNA Prep with Enrichment Dx** is an enrichment-based sequencing solution that enables clinical labs to add targeted sequencing enrichment panels to their diagnostic applications

For added functionality, use Illumina library preparation assays designed for use on the MiSeq System on the MiSeqDx Instrument when run in Research Mode.

† Detailed results reports are available for target-specific assays, such as the TruSight Cystic Fibrosis 139-Variant Assay and TruSight Cystic Fibrosis Clinical Sequencing Assay.

MiSeqDx instrument specifications

Instrument configuration
RFID tracking for consumables MiSeq operating software Local Run Manager software
Instrument control computer (internal)
Base Unit: Intel Core i7-7700 2.9 GHz CPU Memory: 2 × 8 GB DDR4 SO-DIMM Hard drive: None Solid state drives: 2 × 1 TB SATA Operating system: Windows 10
Light emitting diode (LED)
520 nm, 660 nm
Dimensions
W × D × H: 68.6 cm × 56.5 cm × 52.3 cm (27.0 in × 22.2 in × 20.6 in) Weight: 54.5 kg (120 lb) Crated weight: 90.9 kg (200 lb)
Power requirements
90–264 V AC @ 50/60 Hz, 10 A, 400 W
Radio frequency identifier (RFID)
Frequency: 13.56 MHz Power: 100 mW
Throughput
1–96 samples/run, depending upon assay
Performance parameters
Maximum read length: Up to 2 × 300 bp (Refer to package insert for assay-dependent specifications) Output (2 × 150 bp run): ≥ 5 Gb Reads passing filters: ≥ 15 million Q30 score (at read length of 2 × 150 bp): ≥ 80% Accuracy, germline:[a] >99.9% OPA[b] to reference data Accuracy, somatic:[a] 100% OPA to reference data Reproducibility, germline:[c] 99.88% OPA to reference data Reproducibility, somatic:[d] 99.6% expected call for mutant samples

a. Results based on a representative TruSeq Amplicon-based assay designed to query various genes covering 12,588 bases across 23 different chromosomes using 150 amplicons.
b. OPA: overall percent agreement.
c. Results based on Cystic Fibrosis (CF-139) Assay.
d. Results based on a representative two-gene TruSeq Amplicon-based assay.

Ordering information

Product	Catalog no.
MiSeqDx Instrument	DX-410-1001
MiSeqDx Reagent Kit v3	20037124
Illumina DNA Prep with Enrichment Dx with UD Indexes Set A (16 samples)	20051354
Illumina DNA Prep with Enrichment Dx with UD Indexes Set A (96 samples)	20051352
Illumina DNA Prep with Enrichment Dx with UD Indexes Set B (16 samples)	20051355
Illumina DNA Prep with Enrichment Dx with UD Indexes Set B (96 samples)	20051353

Learn more

MiSeqDx Instrument

Molecular diagnostics

MiSeqDx applications

Illumina DNA Prep with Enrichment Dx

References

1. Shokralla S, Porter TM, Gibson JF, et al. Massively parallel multiplex DNA sequencing for specimen identification using an Illumina MiSeq platform. *Sci Rep*. 2015;5:9687. Published 2015 Apr 17. doi:10.1038/srep09687
2. Precone V, Monaco VD, Esposito MV, et al. Cracking the Code of Human Diseases Using Next-Generation Sequencing: Applications, Challenges, and Perspectives. *Biomed Res Int*. 2015;161648. doi:10.1155/2015/161648
3. Bentley DR, Balasubramanian S, Swerdlow HP, et al. Accurate Whole Human Genome Sequencing using Reversible Terminator Chemistry. *Nature*. 2008;456(7218):53–59. doi:10.1038/nature07517
4. Clinical and Functional Translation of CFTR (CTFR2). cftr2.org. Accessed February 23, 2021.

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Intended use statements

MiSeqDx Instrument intended use (United States)

The MiSeqDx Instrument is intended for targeted sequencing of DNA libraries from human genomic DNA extracted from peripheral whole blood, formalin-fixed, paraffin-embedded (FFPE) tissue, or embryonic tissue, when used with *in vitro* diagnostic (IVD) assays performed on the instrument. The MiSeqDx Instrument is not intended for whole genome or *de novo* sequencing. The MiSeqDx Instrument is to be used with registered and listed, cleared, or approved IVD reagents and analytical software.

MiSeqDx Instrument intended use (European Union, excluding Germany/other)

The MiSeqDx Instrument is intended for targeted sequencing of DNA libraries from human genomic DNA extracted from peripheral whole blood, formalin-fixed, paraffin-embedded (FFPE) tissue, or embryonic tissue, when used with *in vitro* diagnostic (IVD) assays performed on the instrument. The MiSeqDx Instrument is not intended for whole genome or *de novo* sequencing. The MiSeqDx Instrument is to be used with registered and listed, cleared, or approved IVD reagents and analytical software.

MiSeqDx Reagent Kit v3 intended use

The Illumina MiSeqDx Reagent Kit v3 is a set of reagents and consumables intended for sequencing of sample libraries when used with validated assays. The MiSeqDx Reagent Kit v3 is intended for use with the MiSeqDx Instrument and analytical software.

TruSight Cystic Fibrosis 139-Variant Assay intended use

The TruSight Cystic Fibrosis 139-Variant Assay (formerly known as the Illumina MiSeqDx Cystic Fibrosis 139-Variant Assay) is a qualitative *in vitro* diagnostic system used to simultaneously detect 139 clinically relevant cystic fibrosis disease-causing mutations and variants of the Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR*) gene in genomic DNA isolated from human peripheral whole blood specimens. The variants include those recommended in 2004 by the American College of Medical Genetics (ACMG)[1] and in 2011 by the American College of Obstetricians and Gynecologists (ACOG).[2] The test is intended for carrier screening in adults of reproductive age, in confirmatory diagnostic testing of newborns and children, and as an initial test to aid in the diagnosis of individuals with suspected cystic fibrosis. The results of this test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available laboratory and clinical information.

This test is not indicated for use for newborn screening, fetal diagnostic testing, preimplantation testing, or for standalone diagnostic purposes.

The test is intended to be used on the Illumina MiSeqDx Instrument.

References for TruSight Cystic Fibrosis 139-Variant Assay

1. Watson MS, Cutting GR, Desnick RJ, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. *Genet Med* 2004;6(5):387–391.
2. American College of Obstetricians and Gynecologists Committee on Genetics. ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis. *Obstet Gynecol.* 2011;117(4):1028–1031.

TruSight Cystic Fibrosis Clinical Sequencing Assay intended use

The TruSight Cystic Fibrosis Clinical Sequencing Assay (formerly known as the Illumina MiSeqDx Cystic Fibrosis Clinical Sequencing Assay) is a targeted sequencing *in vitro* diagnostic system that resequences the protein coding regions and intron/exon boundaries of the Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR*) gene in genomic DNA isolated from human peripheral whole blood specimens collected in K_2EDTA. The test detects single nucleotide variants, and small indels within the region sequenced, and additionally reports on two deep intronic mutations and two large deletions. The test is intended to be used on the Illumina MiSeqDx Instrument.

The test is intended to be used as an aid in the diagnosis of individuals with suspected cystic fibrosis (CF). This assay is most appropriate when the patient has an atypical

or non-classic presentation of CF or when other mutation panels have failed to identify both causative mutations. The results of the test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available information including clinical symptoms, other diagnostic tests, and family history.

This test is not indicated for use for stand-alone diagnostic purposes, fetal diagnostic testing, for preimplantation testing, carrier screening, newborn screening, or population screening.

Illumina DNA Prep with Enrichment Dx intended use (United States)

The Illumina DNA Prep with Enrichment Dx Kit is a set of reagents and consumables used to prepare sample libraries from DNA extracted from peripheral whole blood and formalin-fixed, paraffin-embedded (FFPE) tissue. User-supplied probe panels are required for the preparation of libraries targeting specific genomic regions of interest. The generated sample libraries are intended for use on Illumina sequencing systems.

Illumina DNA Prep with Enrichment Dx intended use (European Union/other)

The Illumina DNA Prep with Enrichment Dx Kit is a set of reagents and consumables used to prepare sample libraries from genomic DNA derived from human cells and tissue to develop *in vitro* diagnostic assays. User-supplied probe panels are required for the preparation of libraries targeting specific genomic regions of interest. The generated sample libraries are intended for use on Illumina sequencing systems. The Illumina DNA Prep with Enrichment Dx includes software for sequencing run setup, monitoring, and analysis.

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1.800.809.4566 toll-free (US) | +1.858.202.4566 tel
techsupport@illumina.com | www.illumina.com

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