



Figure 2: Three-Step MiSeqDx Assay Workflow. Three basic steps for running an assay on the MiSeqDx instrument. Detailed results reports are only available with target-specific assays, such as the MiSeqDx Cystic Fibrosis 139-Variant Assay and the MiSeqDx Cystic Fibrosis Clinical Sequencing Assay.

NGS on the MiSeqDx platform 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. To learn more about SBS chemistry, visit www.illumina.com.

Integrated System Software

The MiSeqDx platform is the first IVD NGS instrument to offer a fully integrated sequencing ecosystem. Instrument software is accessed through a user-friendly touch screen interface. The Illumina Worklist Manager (IWM) software guides you through creation and setup of your sample sheet. MiSeq Operating Software (MOS) guides you through loading the sequencing cartridge (provided pre-loaded with reagents) and provides an overview of quality statistics for monitoring run progress. During the run, MOS also controls the flow cell stage, fluidics system, flow cell temperatures, and captures images of clusters on the flow cell. Upon completion of base calling, the MiSeq Reporter software analyzes assay data and generates the results report.[†]

User Management Software

To ensure proper system use, the MiSeqDx instrument is equipped with Illumina User Management Software and MiSeq Operating Software. Illumina User Management Software enables laboratories to control and trace

system access, ensuring that only authorized personnel are running tests.

Available Kits and Assays

Two IVD assays and one IVD kit are currently available for use on the MiSeqDx instrument:[†]

- The **MiSeqDx Universal Kit** is an amplicon-based sequencing solution that enables users to develop assays targeting the genetic variants most important to them. Users design their own oligonucleotide probes specific to their regions of interest and use the validated reagents in the Universal Kit to prepare libraries and sequence.
- The **MiSeqDx Cystic Fibrosis 139-Variant Assay** detects 139 clinically relevant and functionally verified variants as defined by the CFTR2 database¹.
- The **MiSeqDx Cystic Fibrosis Clinical Sequencing Assay** detects mutations within the protein coding regions and intron/exon boundaries of the cystic fibrosis transmembrane conductance regulator (CFTR) gene.

For added functionality, use Illumina library preparation assays designed for use on the MiSeq® system on the MiSeqDx instrument when run in research mode.

Learn More

Learn more about the MiSeqDx instrument at www.illumina.com/miseqdx.

Reference

1. www.cftr2.org

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

MiSeqDx Instrument Specifications

Instrument Specifications	
Instrument Configuration	
RFID tracking for consumables	
MiSeq Operating Software	
MiSeq Reporter Software	
Instrument Control Computer (Internal)	
Base Unit: Intel Core i7-2710QE 2.10 GHz CPU	
Memory: 16 GB RAM	
Hard Drive: 750 GB	
Operating System: Windows 7 embedded standard	
Light Emitting Diode (LED)	
530 nm, 660 nm	
Dimensions	
WxDxH: 68.6 cm x 56.5 cm x 52.3 cm (27.0 in x 22.2 in x 20.6 in)	
Weight: 54.5 kg (120 lbs)	
Crated Weight: 90.9 kg (200 lbs)	
Power Requirements	
100–240V AC @ 50/60Hz, 10A, 400 W	
Radio Frequency Identifier (RFID)	
Frequency: 13.56 MHz	
Power: 100 mW	
Throughput	
8–48 samples/run, depending upon assay	
Performance Parameters	
Read Length: 2 x 150 bp	
Output: > 1 Gb	
Total overall accuracy: ≥ 99.66% ^a	
Total overall reproducibility: ≥ 99.70% ^a	
Bases with Q30 > 75%	
^a Cystic fibrosis assay-dependent.	

Ordering Information

Product	Catalog No.
MiSeqDx Instrument	DX-410-1001
MiSeqDx Universal Kit	DX-103-1001
MiSeqDx Cystic Fibrosis 139-Variant Assay (20 runs, up to 960 tests)	DX-102-1003
MiSeqDx Cystic Fibrosis 139-Variant Assay (2 runs, up to 96 tests)	DX-102-1004
MiSeqDx Cystic Fibrosis Clinical Sequencing Assay (6 runs, up to 48 samples)	DX-102-1001

Intended Use

The Illumina MiSeqDx is a sequencing instrument that measures fluorescence signals of labeled nucleotides through the use of instrument-specific reagents and flow cells (MiSeqDx Universal Kit 1.0), imaging hardware, and data analysis software. The MiSeqDx platform is intended for targeted sequencing of human genomic DNA from peripheral whole blood samples. The MiSeqDx platform is not intended for whole-genome or *de novo* sequencing. The MiSeqDx Instrument is intended to be used with Illumina-supplied IVD assays and reagent kits.

AAAGAATGATAACAGTAAACACACTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTC AACGTACCGTAAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGCTACCGTGCAACGACGAAAAGAATGATAACAGTAAACACACTTCTGTTAAAC
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AAGATTACTTGATCCACTGATTC AACGTACCGTAAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGCTACCGTGCAACGACGAAAAGAATGATAAC
AACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGCTGTTAAACCTTAAGATTACTTGATCCACTGATTC AACGTACCGTAAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGCTACCGTGCAACGACGAAAAGAATGATAAC

