
I seek
to improve
cystic fibrosis
testing.
See more of what matters with the
MiSeqDx™ Cystic Fibrosis System.
illumina®

Cystic fibrosis (CF) is one of the most common autosomal recessive disorders affecting about 70,000 children and adults worldwide*. Our best defense against this disease is early diagnosis.

Next-generation sequencing (NGS) provides visibility into the whole cystic fibrosis transmembrane conductance regulator (*CFTR*) gene for increased clarity in molecular CF testing. NGS data can help couples understand their reproductive risks and family planning options. These results can also enable earlier detection in affected individuals and selection of optimized therapies. Ultimately, the information can lead to a better quality of life.

Introducing the MiSeqDx Cystic Fibrosis System.

The first and only FDA-cleared NGS system for *in vitro* diagnostic use provides a complete solution for accurate, comprehensive molecular CF testing.

Get a more complete answer the first time

- Accurate data— ≥ 99.99% overall agreement with Sanger sequencing and PCR reference methods at reported positions
- Confident results—sample first pass rate of ≥ 99.7%†
- Established Illumina sequencing by synthesis (SBS) technology‡

See more of the data that matters

- Largest commercially available IVD panel—the MiSeqDx Cystic Fibrosis 139-Variant Assay detects 139 *CFTR* variants§
- Full-gene sequence—the MiSeqDx Cystic Fibrosis Clinical Sequencing Assay accurately sequences protein coding regions and intron/exon boundaries of the *CFTR* gene

Improved CF testing.

The MiSeqDx Cystic Fibrosis System performs the most comprehensive molecular testing to meet all your CF testing needs.

Gain peace of mind.

MiSeqDx Cystic Fibrosis 139-Variant Assay

As the population increases in diversity, so does the need for CF tests that target variants seen in a wider variety of ethnic groups. The MiSeqDx Cystic Fibrosis 139-Variant Assay enables screening of 139 *CFTR* variants, representing a comprehensive set of clinically validated variants in the CFTR2 database[§]. By offering the broadest panel of validated *CFTR* variants, the assay reduces residual risk, providing confidence that you are getting the right answer the first time.

Get the complete genetic story with a single test.

MiSeqDx Cystic Fibrosis Clinical Sequencing Assay

The MiSeqDx Cystic Fibrosis Clinical Sequencing Assay sequences all protein coding regions and intron/exon boundaries, including challenging regions, within the *CFTR* gene. Unlike the tedious Sanger sequencing process, which requires 27 separate PCR steps, the MiSeqDx assay uses a single sequencing test with one PCR step to identify all gene variants, possibly isolating causative variants. This streamlined method drastically shortens the time and expense of current diagnostic odysseys. Ultimately, affected individuals can receive optimal treatment as early as possible.

Easy-to-use, reliable system.

The MiSeqDx Cystic Fibrosis System provides a complete molecular CF testing solution. The easy-to-use workflow with Illumina User Management Software yields high positive and negative predictive values, resulting in an efficient, cost-effective CF testing workflow (Figure 1).

Figure 1: An Overview of the MiSeqDx Cystic Fibrosis System Workflow

Prepare
- Start with genomic DNA
- Create sample sheets
- Generate indexed libraries

Sequence
- Start the MiSeqDx instrument
- Add the library to the pre-aliquoted, ready-to-use MiSeqDx reagent cartridge

Analyze
- See results in the MiSeq Reporter Software
- Interpret results

Single platform. Complete solution.

The MiSeqDx Cystic Fibrosis System runs both the 139-variant and clinical sequencing assays, providing a complete molecular CF testing workflow on a single instrument. This reduces the amount of iterative testing required to produce a comprehensive patient report, eliminates the need to train technicians on the use and maintenance of multiple systems, and consolidates reagent ordering, use, and storage, increasing operational efficiency.

Easy data interpretation.

Results from the MiSeqDx system are presented in an easy-to-read report that can be easily interpreted by a board-certified molecular geneticist or equivalent. All results include:

- Variant type
- HGVS nomenclature
- dbSNP ID
- Call rates

The MiSeqDx Cystic Fibrosis 139-Variant Assay report indicates what variants, out of a possible 139, were present in each tested sample.

The MiSeqDx Cystic Fibrosis Clinical Sequencing Assay report includes base-by-base analysis of variations found within the protein coding regions, intron/exon boundaries, and deep intronic regions.

Seek confidence in your results.

The MiSeqDx Cystic Fibrosis System is the first IVD NGS system for molecular CF testing. With total accuracy and reproducibility rates ≥ 99.7% and a sample first pass rate of ≥ 99.7%, the MiSeqDx system delivers best-in-class performance.

Learn more at **www.illumina.com/cysticfibrosis**

The MiSeqDx Cystic Fibrosis System.

Components

- MiSeqDx Instrument
- MiSeqDx Cystic Fibrosis 139-Variant Assay
- MiSeqDx Cystic Fibrosis Clinical Sequencing Assay

MiSeqDx System Performance Parameters

- Total overall accuracy: ≥ 99.99%†
- Total overall reproducibility: ≥ 99.70%†

MiSeqDx Cystic Fibrosis 139-Variant Assay

- Detects 139 *CFTR* variants as defined by the CFTR2 database§
- Number of samples per assay run: 8–48

MiSeqDx Cystic Fibrosis Clinical Sequencing Assay

- Sequence all protein coding regions and intron exon boundaries
- Detects two large deletions: Exon 2,3 and Exon 22,23
- Detects two deep intronic mutations: 1811+1.6Kb A>G, 3849+10Kb C>T
- Full detection of homopolymer variants (i.e., 2184delA deletion)
- Number of samples per assay run: 8

Ordering Information

Product	Quantity	Catalog No.
MiSeqDx Instrument	1 instrument	DX-410-1001
MiSeqDx Cystic Fibrosis 139-Variant Assay	20 runs, up to 960 samples	DX-102-1003
	2 runs, up to 96 samples	DX-102-1004
MiSeqDx Cystic Fibrosis Clinical Sequencing Assay	6 runs, 48 samples	DX-102-1001

* Cystic Fibrosis Foundation www.cff.org/AboutCF/Faqs/ 26 March 2013.
† Number of samples passing QC metrics on the first attempt in clinical studies; assay specific. Data on file.
‡ Wide adoption in the clinical research community as demonstrated by the number of publications in the PubMed database.
§ Variants defined by the CFTR2 database (http://www.cftr2.org).

For *In Vitro* Diagnostic Use. Contact an Illumina representative for regional availability.

Pub. No. 670-2013-004 Current as of 25 August 2015
Document # 1000000006360 v00

illumina®