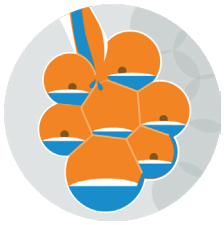
Multiplicom
A part of Agilent Technologies

# CFTR MASTR Dx

**A simple and robust CE-Marked diagnostic assay for complete mutation analysis of the *CFTR* gene by Next-Generation Sequencing. Mutations of the *CFTR* gene affect functioning of the chloride ion channels in epithelial cell membranes, leading to cystic fibrosis and other *CFTR*-related diseases.**



## Application

- For first or second line testing in your CFTR work-up
- For whole blood & dried blood spots derived DNA
- For complete mutational spectrum analysis

validated for dried blood spots

## Assay characteristics

| | |
|---|---|
| Gene analyzed | *CFTR* |
| Genomic region analyzed | 8.7 kb<br>All exons ± 30 bp flanking region, selected introns, part of promoter |
| Number of amplicons | 48<br>including 11 control amplicons |
| Amplicon length | 300-450 bp |
| Number of plexes | 2 |
| DNA amount required | 20 ng per multiplex reaction |
| Validated (CE-IVD) with | MiSeq® System, Illumina |

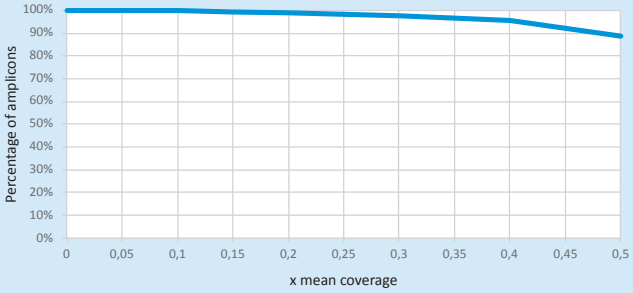
| Mutations included | | |
|---|---|---|
| *All exonic SNVs & small indels*<br>*CNVs** | *~30 bp of intronic flanking sequence* | |
| *HP variants* | *intron 9 (8):* | *c.1210-34TG(m); c.1210-12T(n)* |
| *Selected deep intronic regions:* | *intron 7 (6b):* | *c.1002-1110_1113delTAAG*<br>*c.1002-1111A>C* |
| | *intron 12 (11):* | *c.1679+1634A>G (1811+1.6kbA->G)*<br>*c.1680-877G>T (1811+1643G->T)*<br>*c.1680-883A>G* |
| | *intron 22 (19):* | *c.3718-2477C>T (3849+10kbC->T* |
| *Promoter region* | *up to position c.-108* | |

**For Research Use Only. Not for use in diagnostic procedures*

## Performance Characteristics

### CFTR MASTR Dx on MiSeq, germline DNA

| | DNA extracted from blood | DNA extracted from dried blood spots |
|---|---|---|
| On target read pair counts | *98.92 %*<br>*[92.70 %-99.51 %]* | *99.90 %*<br>*[99.53 %-99.98 %]* |
| Uniformity of amplification (0.2x mean coverage) | *97.83 %* | *100 %* |




*Graph representing the read counts for all 48 CFTR MASTR Dx amplicons, showing their uniform representation. To allow comparison between samples, the read counts were normalized.*

## Performance Characteristics

### CFTR MASTR Dx on MiSeq; germline DNA

| Extracted from blood | | | Extracted from dried blood spots | |
|---|---|---|---|---|
| Performance parameter | Observed [95 % CI] Using JSI SeqNext | Observed [95 % CI] Using Sophia DDM | Observed Using JSI SeqNext | Observed Using Sophia DDM |
| *Sensitivity* | *100 % [ ≥ 99.217 %]* | *100 % [ ≥ 98.202 %]* | *64/64 = 100 %* | *64/64 = 100 %* |
| *Specificity* | *99.990 % [99.975 % –99.994 %]* | *100 % [ ≥ 99.999 %]* | *1438/1438 = 100 %* | *1438/1438 = 100 %* |
| *Accuracy* | *99.990% [99.975 % –99.994 %]* | *100 % [ ≥ 99.999 %]* | | |
| *Repeatability & reproducibility* | *99.997 % [99.988 % –99.999 %]* | *100 % [ ≥ 99.990 %]* | | |

## Workflow

### Advised maximum number of samples per run

| Sequencing System | Illumina MiSeq® | | |
|---|---|---|---|
| | Reagent Kit | | |
| *Flow cell* | *Nano v 2 2 x 251 cycles* | *v2 2 x 251 cycles* | *v3 2 x 276 cycles* |
| *SNV variant calling Minimal coverage per allele: 50* | *52* | *786* | *1441* |
| *SNV and CNV* variant calling Minimal coverage per amplicon: 200* | *26* | *393* | *720* |

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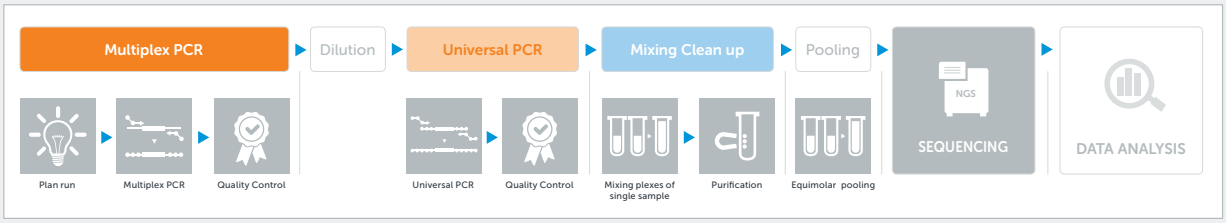


## Publications

**Supporting publications:**

Applicability and Efficiency of NGS in Routine Diagnosis: In-Depth Performance Analysis of a Complete Workflow for CFTR Mutation Analysis. Andrien Pagin et al. (2016)
*PLOS ONE DOI: 10.1371/journal.pone.0149426, 22 Feb 2016**

**This study was done by a third party and was verified by Multiplicom.*

## Order information

| Cat. No. | Product Name | Reactions | PR Number |
|---|---|---|---|
| *MR-2021.024* | *CFTR MASTR Dx* | *24* | *PR7000-1421* |
| *MR-2021.048* | | *48* | |

*MID (Molecular Identifiers) kits are necessary to complete the workflow.*

*5991-8404EN*
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*For In Vitro Diagnostic Use.*

1709 - EU

Multiplicom N.V. [ a ] Galileïlaan 18, B-2845 Niel, Belgium [ w ] www.agilent.com [ e ] mtp.customerservice@agilent.com [ t ] +32 3 289 14 00 [ f ] +32 3 400 23 16