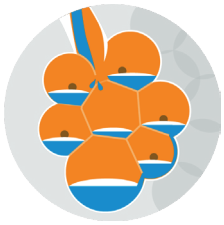
Multiplicom
A part of Agilent Technologies

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 All exons ± 30 bp flanking region, selected introns, part of promoter
Number of amplicons	48 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

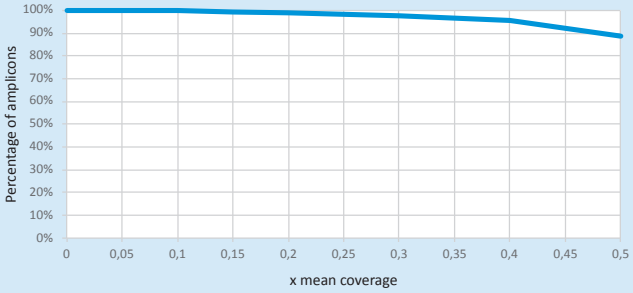
Mutations included		
All exonic SNVs & small indels *CNVs**	*~30 bp of intronic flanking sequence*	
HP variants	*intron 9 (8):*	*c.1210-34TG(m); c.1210-12T(n)*
Selected deep intronic regions:	*intron 7 (6b):*	*c.1002-1110_1113delTAAG* *c.1002-1111A>C*
	intron 12 (11):	*c.1679+1634A>G (1811+1.6kbA->G)* *c.1680-877G>T (1811+1643G->T)* *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 %* *[92.70 %-99.51 %]*	*99.90 %* *[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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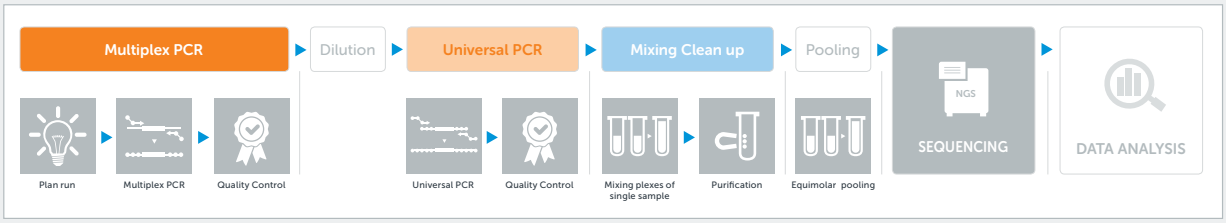

Publications

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

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