

A simple and robust CE-Marked diagnostic assay for complete mutation analysis in the coding regions of the *MEFV* gene by Next-Generation Sequencing. Mutations of the *MEFV* gene affect normal functioning of the pyrin protein, leading to the inflammatory disorder Familial Mediterranean Fever (FMF). FMF MASTR Dx can be used to complete the diagnosis and treatment selection FMF patients, as well as for carrier screening purposes in families at risk.



Application

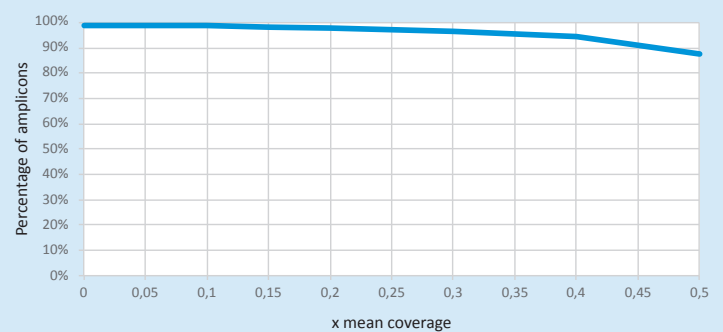
- For the identification of mutations (SNVs and indels) in the Mediterranean Fever gene (*MEFV*).
- For complete mutational spectrum analysis in one assay
- Ready-to-use kit with simple-to-follow protocols

Assay characteristics

Gene analyzed	<i>MEFV</i>
Genomic region analyzed	4.2 kb All exons \pm 30 bp flanking region
Number of amplicons	23
Amplicon length	250-400 bp
Number of plexes	2
DNA amount required	20 ng per multiplex reaction
Validated (CE-IVD) with	MiSeq System, Illumina

FMF MASTR Dx on MiSeq; germline DNA

	DNA extracted from blood
On target read pair counts	94.6 % [87.6 % - 97.6 %]
Uniformity of amplification (0.2x mean coverage)	99.4 %



Graph representing the read pair counts for all 23 FMF MASTR Dx amplicons, showing their uniform representation. To allow comparison between samples, the read counts were normalized.

Performance Characteristics

Performance parameter	DNA Extracted from blood	
	Observed [95 % CI] Using JSI SeqNext	Observed [95 % CI] Using Sophia DDM
Sensitivity	100 % [\geq 99.459 %]	100 % [\geq 99.459 %]
Specificity	100 % [\geq 99.999 %]	100 % [\geq 99.999 %]
Accuracy	100 % [\geq 99.999 %]	100 % [\geq 99.999 %]

Advised maximum number of samples per run

Sequencing System	Illumina MiSeq		
	Reagent Kit		
Flow cell	Nano v2 2 x 251 cycles	v2 2 x 251 cycles	v3 2 x 276 cycles
SNV variant calling Minimal coverage per allele: 50	116	1753	3215



Cat. No.	Product Name	Reactions
MR-2071.024	FMF MASTR Dx	24

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

For In Vitro Diagnostic Use.

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