

# FMF

StripA<sup>ssay</sup>

## INTENDED USE

The ViennaLab FMF StripA<sup>ssay</sup> provides materials for the isolation of DNA from human whole blood, the in vitro amplification of MEFV gene sequences, and the subsequent detection of twelve mutations by reverse-hybridization.

## INTRODUCTION

Familial Mediterranean fever (FMF) is an autosomal-recessive inherited inflammatory disorder. It is characterized by recurrent, short (usually lasting 2-4 days), self-limiting bouts of fever, accompanied by pain in the abdomen, chest or joints, and sometimes associated with erysipelas-like erythema. The most severe complication is progressive amyloidosis, ultimately leading to renal failure. FMF predominantly affects Turks, Arabs, Armenians and Sephardic Jews, with carrier rates as high as 1:5, but has been observed in lower frequencies throughout the Mediterranean area.

The responsible disease gene, designated MEFV, has been mapped to chromosome 16p13, comprises 10 exons and encodes a protein termed marenostriin or pyrin. While a few founder mutations (e.g. M680I, M694V) are observed in the majority of cases, other MEFV mutations are rare, and the various combinations seem to define the severity of the disease phenotype and the risk to develop renal amyloidosis.

Owing to the rather nonspecific clinical symptoms, molecular genetic analysis significantly improves early and correct diagnosis of FMF, and allows to commence lifelong prophylactic treatment of affected individuals with colchicine.

## PRINCIPLES OF THE ASSAY

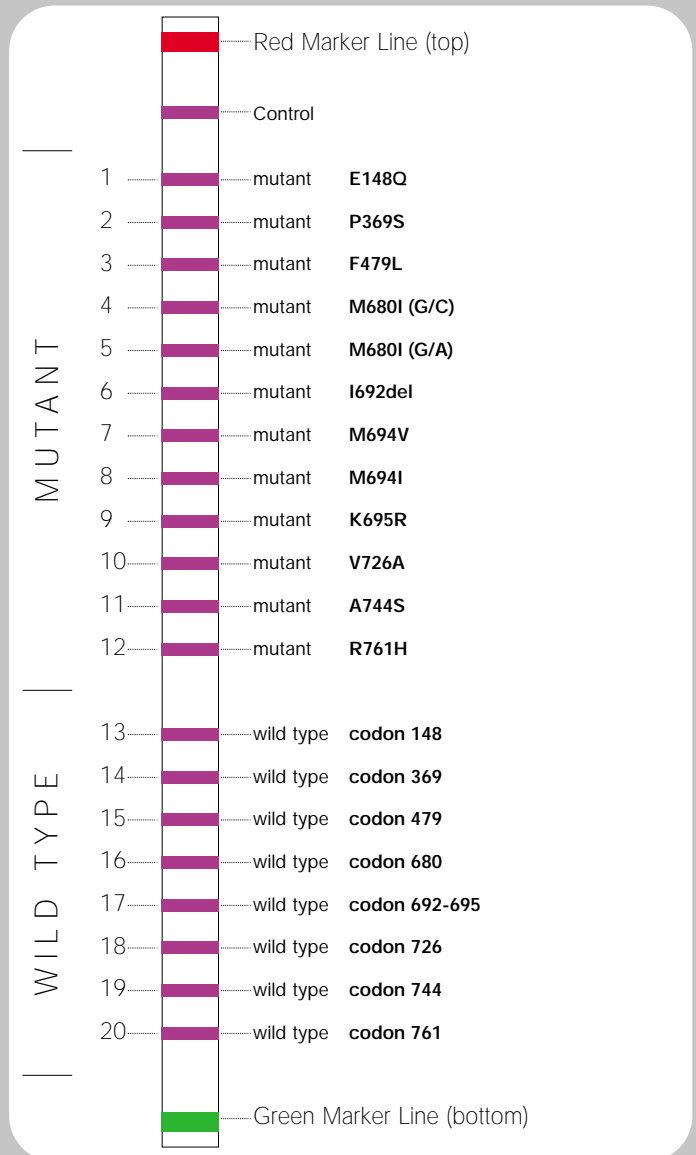
The FMF StripA<sup>ssay</sup> is based on the reverse-hybridization principle, and includes three successive steps: DNA is isolated from anticoagulated blood by a rapid and convenient procedure. Then, MEFV gene sequences are simultaneously in vitro amplified and biotin-labelled in a single («multiplex») amplification reaction. Finally, the amplification products are selectively hybridized to a test strip, which contains oligonucleotide probes (wild type- and mutant-specific) immobilized as parallel lines. Bound biotinylated sequences are detected using streptavidin-alkaline phosphatase and color substrates.

The assay covers the following 12 MEFV mutations: E148Q, P369S, F479L, M680I (G/C), M680I (G/A), I692del, M694V, M694I, K695R, V726A, A744S, R761H.

References: The French FMF Consortium (1997), Nature Genetics 17, 25-31.

Bernot, A., da Silva, C., Petit, J.-L., et al. (1998), Hum. Mol. Genet. 7, 1317-1325.

Teletar, M., Grody, W.W. (2000), Mol. Genet. Metab. 71, 256-260.



# FMMF<sup>StripAssay</sup>

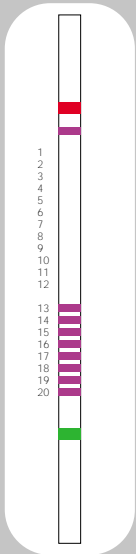
## TEST RESULTS:

For each polymorphic position, one of three possible staining patterns may be obtained:

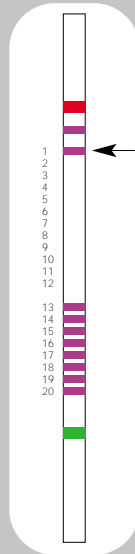
1. wild type probe only: *normal genotype*
2. wild type and mutant probe: *heterozygous genotype*  
(«carrier» individual)
3. mutant probe only: *homozygous mutant genotype*  
(«affected» individual)

## EXAMPLES:

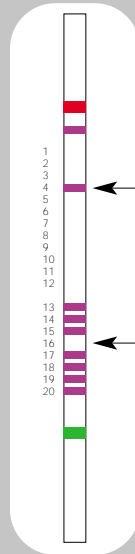
(A.) normal



(B.) E148Q heterozygous



(C.) M680I (G/C) homozygous



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