# Sequencing of MEFV gene in heterozygous Familial Mediterranean Fever patients

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# Abstract

Familial Mediterranean Fever (FMF) is an autosomal recessive auto-inflammatory disease of genetic origin. Screening of MEFV gene mutations determines the definitive diagnosis of FMF. About 30% of patients with clinical FMF possess only one demonstrable mutation and often have typical disease manifestations and respond well to colchicine, the standard treatment for FMF. However failure to find mutations in the MEFV gene does not rule out the disease. The Aim of this study was to perform direct sequencing of the MEFV gene in patients with clinical manifestations having only one hit of mutation by screening tests, searching for another mutation. The study included twenty patients fulfilling the criteria of FMF diagnosis. History taking and laboratory work up was done; this includes complete blood count, Erythrocyte sedimentation rate, C-reactive protein & serum Amyloid A protein assay. Screening for mutations was done by FMF strip assay that screen for the most common mutations by reversed hybridization technique. Sanger Sequencing was performed for the whole MEFV exons and exon-intron boundaries for all FMF patients. The study revealed the absence of other pathogenic variants in heterozygous patients & also in patients with no detectable mutations by

screening method. It was concluded that patients with a single mutation or even without any mutation could be manifesting with the typical attacks of the disease and with complete remission after reasonable dose of colchicine. Sanger sequencing may not reveal a second hit in FMF patients in our population and the screening method which is easier to perform and cheaper may be satisfactory to determine the mutated allele and to manage the patients.

**Keywords:**

Periodic Fever Syndromes – FMF – MEFV – Sequencing – pyrin