FAMILY CASE WITH FAMILIAL MEDITERRANEAN FEVER (FMF)

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Background. Familial Mediterranean fever (FMF) is an auto-inflammatory disease characterized by periodic episodes of fever and recurrent polyserositis. It is caused by a dysfunction of pyrin as a result of various mutations within MEFV gene, some causing very severe cases, while others may have milder symptoms **Objective of the study.** To report the case of a family in which 5 members were confirmed genetically with mutations characteristic for FMF out of which 4 displayed similar symptoms. The 4 members displaying signs are the father and the 3 out of 6 siblings(2 males+ 1 female) **Material and Methods.** Disease history and results of genetic testing of the family members **Results.** The main presenting complaint in all members is the recurrent abdominal pain variably followed by cramps and flatulence, with/without diarrhea which manifest as attacks for 2-3 days a month. Symptomatic disease onset varied from 9 to 45 y.o., 3 members have left knee arthritis, 4 members have pleuritic chest pain, one has erythema nodosum in both shins and one member is asymptomatic. The father tested genetically as follows FMF-V726A carrier; FMF-E148Q homozygote. and all siblings FMF-V726A heterozygote; FMF-E148Q heterozygote. 4 patients manage to control the disease with diet and colchicine, and the asymptomatic one doesn't use colchicine because of breastfeeding period. **Conclusion.** Although traditionally fever is a considered a hallmark of FMF, with the discovery of genetic mutations, we can confirm a greater variety of clinical presentation, not all cases presenting with all classical symptoms. even-though all siblings have the same mutation they have different symptoms.

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