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A Family Case with Familial Mediterranean Fever

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Introduction

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

Keywords

Familial Mediterranean Fever, serositis, genetic testing

Purpose

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.

Table 1.

Feature	Father	Sibling 1	Sibling 2	Sibling 3	Sibling 4
Recurrent abdominal pain	+	+	+	+	+
Gender	male	male	male	female	female
Arthritis	+	+	-	+	-
Age of onset	45	27	23	9	12
Pleuritic chest pain	+	+	+	+	-
Erythema nodosum	-	+	-	-	-
Genetic variant	FMF-V726A carrier; FMF-E148Q homozygote	FMF-V726A heterozygote; FMF-E148Q heterozygote	FMF-V726A heterozygote; FMF-E148Q heterozygote	FMF-V726A heterozygote; FMF-E148Q heterozygote	FMF-V726A heterozygote; FMF-E148Q heterozygote

Conclusions

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