

Unusual presentation of familial Mediterranean fever: atypical hyperaemic recurrent skin lesions

Sirs,

Familial Mediterranean fever (FMF) is the most common autoinflammatory disease (1). It is an autosomal recessive disease which primarily affects Jewish, Turkish, Arabic, and Armenian populations (2, 3). It is characterised by recurrent fever accompanied by peritonitis, pleuritis, arthritis, or erysipelas-like erythema. With the aid of research in relevant literature, we discuss a rarely reported case that has atypical cutaneous manifestation of FMF. The originality of this case is polymorphism of skin lesions of FMF.

Here we present a patient who was predominantly presenting with recurrent multifocal hyperemic lesions on scalp, was diagnosed with FMF and discussed skin involvement in FMF with regard to the literature.

A 52-year-old man was admitted with a multifocal hyperaemic scalp lesion that had been present for 6 years. He had had primary hypertension, type 2 diabetes mellitus, for 6 years. Before this admission, he was seen by a physician in the dermatology department whose prior diagnosis was neurodermatitis or dermatitis. For that reason, steroid injection was started for 8 weeks that diminished his complaints, but did not totally eliminate them. He had no fever, no abdominal, chest, or joint pain. Medications taken at home were insulin and amyloidipin. His parents were non-consanguineous. His parents' ethnicity is Armenian. He has hyperaemic multifocal shiny scalp lesions (Fig. 1). There was abdominal distention, with tenderness in the right upper quadrant. Other physical examinations showed no abnormal findings. A skin biopsy taken from the lesion showed Amyloid A (AA) type amyloidosis.

During the process of investigation, tests for anti-nuclear antibodies, rheumatoid factor and anti-cyclic citrullinated peptide were negative. The tuberculin test also was negative. Rectal specimen was positive for amyloidosis. At that time, sequencing of the whole exon of the *MEFV* gene was performed and a homozygote mutation on R202Q Exon 2 in the *MEFV* gene was detected.

The patient was diagnosed with FMF according to the Tel-Hashomer criteria and then confirmed with genetic analysis. Scalp lesions were successfully treated with colchicine 1.5 mg/day without any relapse during the 3-month follow-up.

Fig. 1.



Familial Mediterranean Fever is characterised by recurrent fever attacks associated with polyserositis. Less frequently encountered symptoms are skin lesions such as erysipelas-like erythema, erythema nodosum, and panniculitis (3). In this case, we showed that atypical hyperemic recurrent shiny scalp lesions have been described in rarely cutaneous manifestations of FMF. The reported frequency of this skin presentation may vary according to ethnic group. It has been noted in 20.9% of Turks, 46% of Jews, 3% of Arabs, and 8% of Armenians (1). In our case report, the origin of the patient's parents is Armenian, an ethnic group in which skin presentation is less common than other ethnic groups.

FMF patients with similar genotype may express different disease phenotypes (4). FMF type 1 is characterised by classical symptoms like recurrent fever, synovitis, peritonitis, and pleuritis. Like in our cases, some individuals develop amyloidosis without having recurrent inflammatory episodes (FMF type 2). The symptoms and severity vary among affected individuals. This difference may be due to the effect of the environment or other modifier genes, epigenetics. Previous studies have shown that secondary amyloidosis has been observed less commonly among Armenian FMF patients in the USA (5).

Amyloidosis is the most severe complication of FMF and leads to renal failure. In our cases, secondary causes of amyloidosis were excluded such as autoimmune diseases like rheumatoid arthritis, and chronic

infections like tuberculosis, and cancers like Hodgkin's lymphoma, renal cell carcinoma. In conclusion, the originality of this case is atypical presentation of FMF that AA type amyloidosis on skin lesions were detected without any other symptoms of FMF. Our report emphasises that we should pay attention to repeated atypical skin eruptions for early detection of atypical FMF.

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Competing interests: none declared.

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