Neutrophilic Lobular Panniculitis as an Expression of a Widened Spectrum of Familial Mediterranean Fever

Familial Mediterranean fever (FMF) is considered to be an autosomal recessive disease, though it is controversial.1,2 The marenosmin-encoding fever gene (MEFV) is responsible for FMF. The most frequent mutation is M694V, which represents a genetic risk factor for development of amyloidosis3 and more severe disease. The classic clinical picture consists of generally short recurrent febrile episodes, serositis, and erysipelas-like erythema (ELE). Colchicine is the treatment of choice for prevention of the attacks and AA amyloidosis.4

Discussion | In the multiple articles that describe clinical and genetic features of FMF, 10% to 40% heterozygous mutations were detected.1,5 In fact, there is a series of 94 patients6 carrying a single mutated allele and sharing clinical features with our case: a younger age of onset, longer febrile periods, and a majority of skin eruptions that were not typical ELE. The experts highlight the clinical and therapeutic importance of these single mutations and propose a therapeutic trial with colchicine to support FMF diagnosis.7,8 Finally, some authors warn about an expanded spectrum of FMF with new recurrent clinical manifestations that should be considered in cases with rare mutations and mutations in heterozygosis.7

The genetic explanation for developing symptoms while carrying a single mutated allele lies in several hypotheses9: a dominant inheritance with incomplete penetration under certain environmental backgrounds, oligogenism, difficulties in the detection of rare mutations, and pseudodominance phenomenon.
Lobular neutrophilic panniculitis has been reported in 3 adult patients with FMF. In all cases, there was a long personal history of FMF with serositis and periodic fever. Two of them had typical ELE and were undergoing hemodialysis. However, in our patient, panniculitis was the main clinical manifestation along with periodic fever. We report a case encompassed in the clinical spectrum of FMF. To our knowledge, this is the first observation of lobular panniculitis as the main clinical expression of FMF. Some patients misdiagnosed as having idiopathic infantile febrile panniculitis could be included in the spectrum of FMF and might benefit from a correct diagnosis and treatment preventing a complication as severe as AA amyloidosis.

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