

## Questioning the diagnosis of familial Mediterranean fever based solely on *MEFV* mutation: a critical review of overlapping features with inflammatory bowel disease

Sirs,

After reading the case report published by Hoshi *et al.* we were intrigued by the clinical picture presented about the overlapping features between familial Mediterranean fever (FMF) and inflammatory bowel disease (IBD) (1). However, we have some concerns about the efficacy of colchicine treatment in this particular patient. We think a more cautious interpretation is warranted in this case.

First, the authors concluded that colchicine treatment resulted in a complete clinical cure. As is known, diseases such as Crohn's disease (CD) and ulcerative colitis (UC) are characterised by periods of exacerbation and remission. Therefore, the patient's symptoms may have resolved spontaneously despite colchicine administration. Furthermore, the report does not discuss whether cytokine profile changes caused by previous drugs, such as infliximab and 5-aminosalicylic acid, or the patient's immune response contributed to the clinical improvement.

And most importantly, FMF is not diagnosed by detecting a mutation in the *MEFV* gene alone (2). Therefore, the presence of a mutation in exon 2 of the *MEFV* gene is

insufficient for diagnosing FMF. Even if the authors claim that they did not diagnose FMF but detected an FMF mutation, this is misleading. The title implies a diagnosis of FMF and leads readers to assume that the patient has FMF. In addition, the contribution of exon 2 mutations to pathogenesis is being questioned more and more every day (3). Clinical findings are essential according to established diagnostic criteria such as the Tel-Hashomer or Eurofever criteria. There have been no exceptions to this so far; extensive case studies are an example (4, 5). The evaluation of the findings in this patient as the first FMF attack is also problematic. Because if there is a second attack, it is considered to be a disease with recurrent attacks. As the studies show, there is still much more to learn about FMF (6, 7). In conclusion, we recommend that the authors respond to these concerns and that future studies be reviewed in this respect.

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