

# Famillial Mediterranean Fever in Centrale and Eastern European Countries with Low Prevalence – Why Shoud We Care?

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**Abstract :** Familial Mediterranean Fever (FMF, MIM249100) is the most common of all hereditary periodic fever syndromes. It is a result of *MEFV* gen mutation („Mediterranean Fever“), usually inheritide in an autosomal recesive pattern. *MEFV* gene codes for the protein pyrin/marenostrin which seems to have an important role in the regulation of IL-1 $\beta$  activation. Disease is characterised by recurrent, self limited, febrile episodes with serositis, synovitis, and occasionally, skin involvement. Attacks are often associated with high levels of acute phase reactants.

FMF usually presents in childhood and primarily affects population in eastern Mediterranean countries where FMF is first in differential diagnosis of patient with reccurent fever. In others, diagnosis is often significantly delayed (7.3 years (range 0.3-76)) mostly due to low awareness of the disease. It is important to recognise as it could be completelly controlled by long-term therapy. Untreated disease carries risk of severe complications, including renal failure from AA amyloidosis. Diagnosis relies on clinical suspicion followed by genetic testing. There are several sets of diagnostic criteria developed for adults and the last one for children (Turkish FMF pediatric criteria). Genetic testing for FMF is not routinely available Eastern and Central European countries.

One clinical aspect is atracting a lot of attention in literature lately: an increased rate of *MEFV* gene mutations among patients with inflammatory disease other then FMF, such as vasculitis (Henoch-Scholein purpura, polyarteritis nodosa) and various childhood rheumatic diseases, suggesting that the same gene (*MEFV*) may be responsible for more than a single disease or syndrome. It is another important reason why should we care about FMF and *MEFV* gene mutations in low prevalence eastern and central european countries.

**Keywords:** Familial Mediterranean Fever, *MEFV* gene, low prevalence FMF countries