Familial Mediterranean Fever (FMF) Media Backgrounder

Familial Mediterranean Fever (FMF)
A Rare Autoinflammatory Disease

Familial Mediterranean Fever (FMF) is a rare, debilitating autoinflammatory disease that can affect both children and adults. Patients with this genetically inherited disease experience attacks of fever, rash and painful inflammation that affects the joints, abdomen and tissues that protect organs including the heart and lungs. Each attack lasts between one and four days.

FMF is one of six conditions that make up the periodic fever syndromes (PFSs); along with tumor necrosis factor (TNF) receptor-associated periodic syndrome (TRAPS), Cryopyrin-Associated Periodic Syndromes (CAPS), hyperimmunoglobulinemia D and periodic fever syndrome (HIDS), Pyogenic sterile Arthritis, Pyoderma gangrenosum and Acne (PAPA) syndrome, and Blau syndrome, and is the most prevalent of these syndromes. The disease onset of FMF usually occurs under the age of 20 years, with two thirds of patients being diagnosed before the age of 5 years.

FMF is a rare disease, so few data exists on its incidence and prevalence in the general population. An estimate of its prevalence is 100–500 in one million people. Although FMF is a worldwide disease, it is far more prevalent in populations living around the Mediterranean basin, including in Armenian, North African, Arab, Turkish and non-Ashkenazi Jewish people. The prevalence of the disease in these populations can reach as high as 1 in 200 individuals.

The role of inflammation in FMF
FMF is a genetically-inherited condition caused by mutations in a gene called MEFV. This gene encodes a protein called pyrin, which is involved in the regulation of inflammation and immunity. Pyrin acts to regulate the activation of caspase-1, which in turn controls the production of IL-1 beta, a potent proinflammatory cytokine (a type of immune system signalling molecule). Excessive production of IL-1 beta plays a major role in FMF.

Unsuccessfully treated FMF can lead to severe complications
One of the most severe complications of FMF is amyloidosis, which is estimated to occur in 50–60% of untreated patients. This long-term complication involves the production of a protein called serum amyloid A (SAA) during inflammation, and can lead to liver or kidney failure. In some instances, amyloidosis can be fatal.

Diagnosis and treatment challenges
FMF symptoms can appear to mimic many common acquired disorders such as infections, acute appendicitis and arthritis. This can delay diagnosis for many years and subject patients to extensive investigation, including exploratory surgery. Colchicine has been used to treat FMF but in 10–15% of patients, the response to colchicine may be low or absent, meaning that these patients continue to suffer the debilitating symptoms of FMF. These patients also have an increased risk of developing amyloidosis. There is currently no accepted alternative treatment for patients with colchicine-resistant FMF. With limitations in treatment with colchicine and the lack of alternative options, there remains a strong unmet need for therapies that can address the symptoms associated with FMF. The role of IL-1
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beta production in the pathogenesis of FMF is a promising area of research that could yield potential new treatments.

References

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