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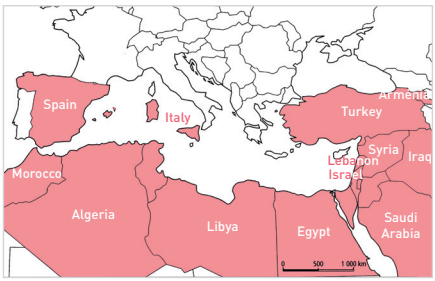
Focus on...

Familial Mediterranean Fever (FMF)

Familial Mediterranean Fever - MEFV gene (MEditerranean FeVer) - molecular study by New Generation Sequencing

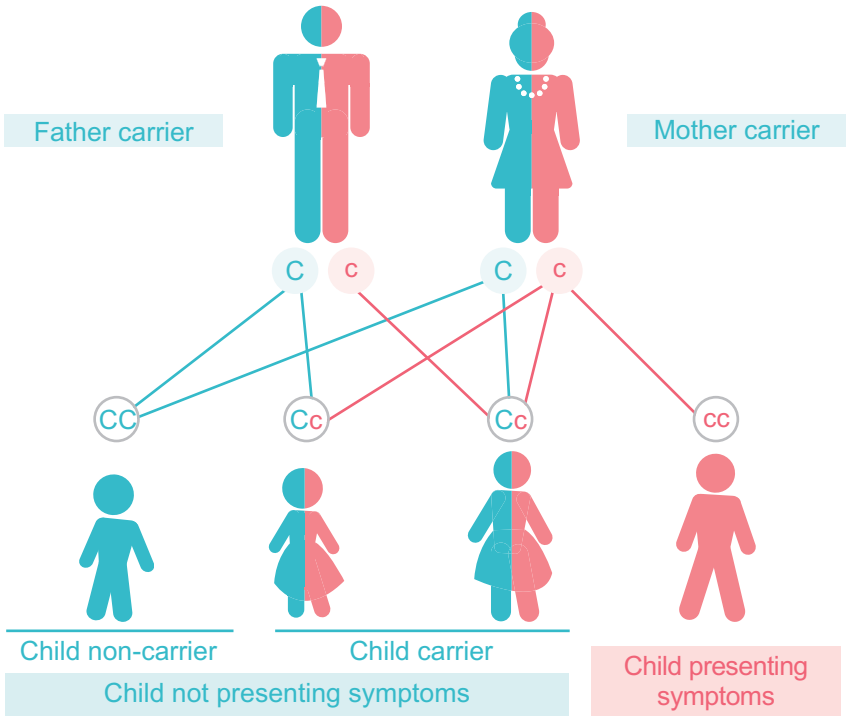
FMF: what is it?

Familial Mediterranean Fever (FMF) is a hereditary autoinflammatory disease characterized by recurrent attacks of fever and serositis resulting in abdominal, chest, joint and muscle pain. FMF is the most common familial relapsing fever. It mainly affects the populations of the Middle East and the Mediterranean basin, particularly Sephardic Jews, Armenians, Arabs and Turks, with a prevalence ranging from 1/150 to 1/1000.



In 90% of cases, the first onset of the disease is before the age of twenty.
The main long-term complication is **AA amyloidosis**, a severe condition with a poor prognosis. Colchicine remains the therapy of choice in the prevention of crises and complications.
It is therefore crucial that the diagnosis of FMF is made, so that this treatment can be initiated.

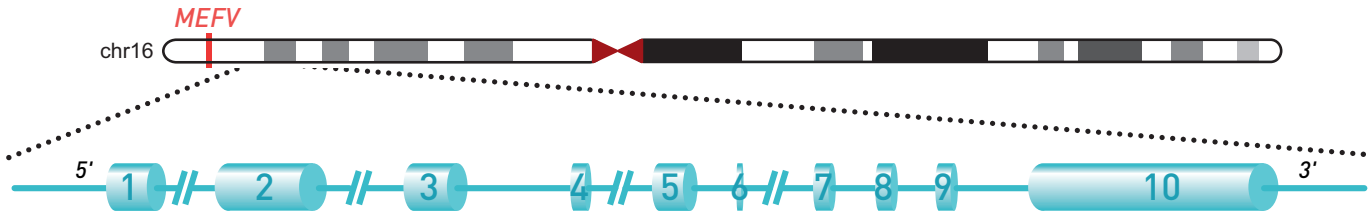
Familial Mediterranean Fever: an autosomal recessive disease



Clinical suspicion of FMF can be confirmed by studying the MEFV gene (MEditerranean FeVer), which consists of 10 exons and is located on the short arm of chromosome 16. At Eurofins Biomnis, we use New Generation Sequencing technology (NGS) to obtain a complete sequence of the 10 exons of this gene.

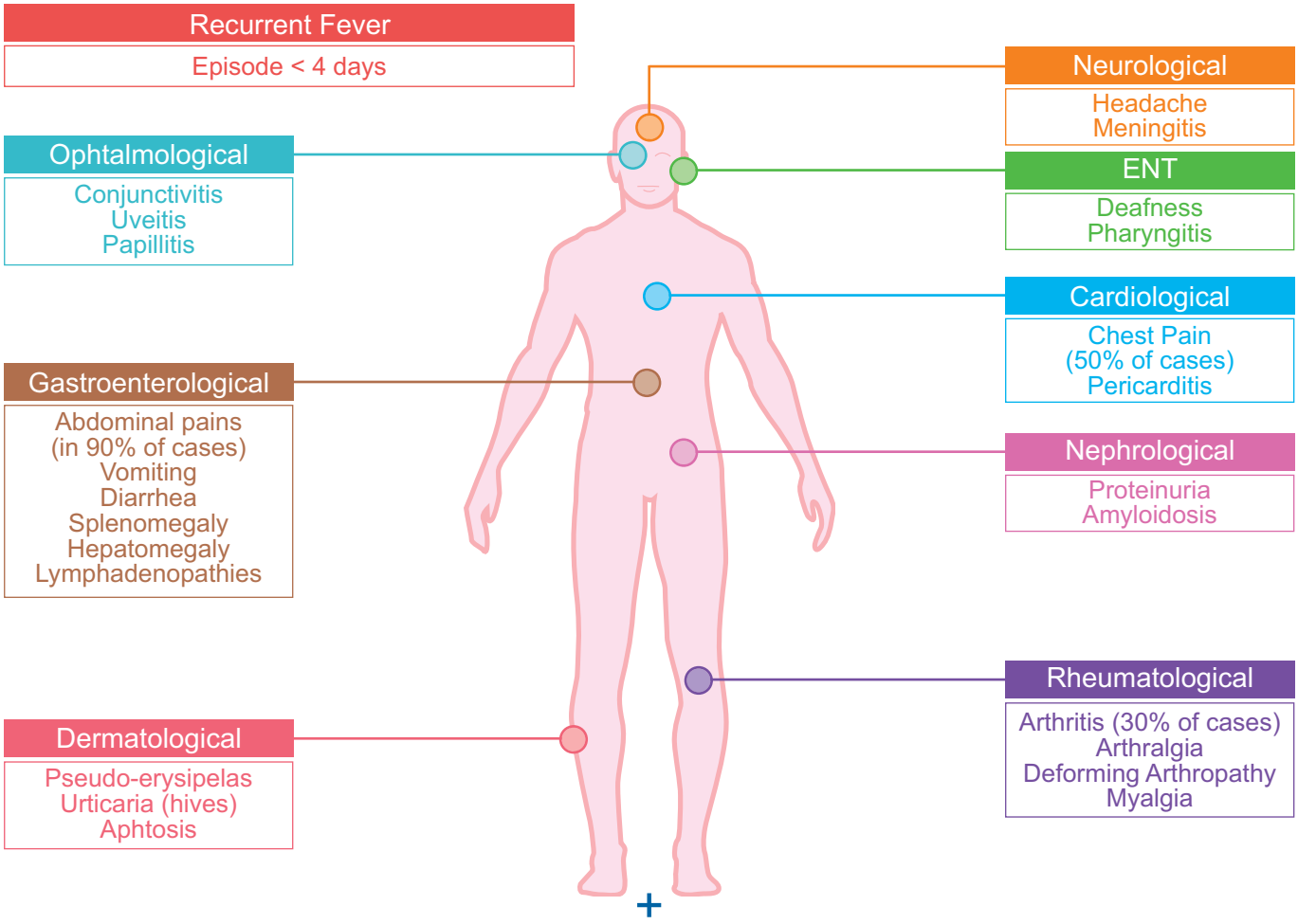
The presence of any pathogenic or probably pathogenic variation or a variation of unknown significance (VSI) is systematically confirmed by a second technique (Sanger sequencing).

Clinical interpretation of the observed genotype is provided in the return report. The testing of the parents may also sometimes be proposed to clarify the genotype-phenotype correlation and for the purposes of genetic counselling.



With Next Generation Sequencing, all 10 exons of the MEFV gene can be analysed

Screening for the MEFV gene: what are the indications?



Because of this wide variety of symptoms, the diagnosis of FMF is a real challenge.

*ESR: Erythrocyte Sedimentation Rate; CRP: C-Reactive Protein; SAA: Serum Amyloid A

References to literature:
Giancane et al, Ann Rheum Dis, 2015: Evidence-based recommendations for genetic diagnosis of familial Mediterranean fever.
Shinar et al, Ann Rheum Dis, 2012: Guidelines for the genetic diagnosis of hereditary recurrent fevers.

Pre-analytic conditions for testing for FMF

- **Eurofins Biomnis analysis code:** FMF
- **Type of sample:** 5mL of whole blood plasma EDTA
- **Temperature:** Ambient (refrigerated if transport > 48h)
- **Turnaround time:** 1 month
- **Technique:** New Generation Sequencing (NGS)

Please note: ESSENTIAL to join the informed consent form and the information document **R36 INTGB – Study of the Familial Mediterranean Fever Gene** with each request.
These documents can be downloaded from our website at **www.eurofins-biomnis.com**

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