Gene test information

FAMILIAL MEDITERRANEAN FEVER (MEFV GENE TEST)

- **Background**

  Familial Mediterranean fever (FMF), also called recurrent polyserositis, is characterized by brief recurrent episodes of peritonitis, pleuritis, and arthritis, usually with accompanying fever. FMF occurs within families and is much more common in individuals of Mediterranean descent than in persons of any other ethnicity. Nonsense or missense mutations in the MEFV gene appear to cause the disease in most cases.

  The diagnosis is clinically made on the basis of the history of typical attacks, especially in patients from the ethnic groups in which FMF is more highly prevalent. An acute phase response is present during attacks, with high C-reactive protein levels, an elevated white blood cell count and other markers of inflammation.

  Additionally, diagnosis of FMF can be confirmed by a genetic test that detects mutations in the MEFV gene. Sequencing of exons 2, 3, 5, and 10 of this gene are required to detect disease-associated mutations.

References:

Yepiskoposyan L, Harutyunyan A. Population genetics of familial Mediterranean fever: a review. Eur J Hum Genet. 2007;15:911-916.

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