Familial mediterranean fever

GENERAL INFORMATION

Description:

Defects in MEFV are the cause of familial mediterranean fever. Familial mediterranean fever is an autosomal recessive inherited disorder characterized by recurrent episodic fever, serosal inflammation and pain in the abdomen, chest or joints. It is frequently complicated by amyloidosis, which leads to renal failure. Familial mediterranean fever primarily affects ethnic groups living around the mediterranean basin north-african jews, armenians, arabs and turks. Defects in MEFV are associated with reduced pyrin expression.

Alternative names:

- FMF
- Mediterranean Fever, Familial; Recurrent polyserositis, Periodic disease

Classification:

- Periodic fever syndromes

Inheritance:

Autosomal recessive

OMIM:

- *608107 Familial mediterranean fever; MEFV

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Hyperimmunoglobulinemia D with periodic fever syndrome
- IDR factfile for Tumor necrosis factor receptor-associated periodic syndrome

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Familial Mediterranean fever is divided into types 1 and 2. Patients with FMF type 1 have short episodes of inflammation and serositis including fever, peritonitis, synovitis, pleuritis, and rarely, pericarditis and meningitis. Amyloidoisis is the most severe complication, can lead to renal failure. Patients with FMF type 2 have amyloidosis, as the first clinical manifestation of disease in an asymptomatic individual.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Mediterranean fever, familial, ORPHANET
- Mediterranean fever, familial, eMedicine

Genetic:

- IDdiagnostics
- Familial Mediterranean Fever, GeneTest

Therapeutic options:

- Effective treatment with colchicine. Interferon-alpha for patients with colchicine-resistant disease.
- Mediterranean fever, familial, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: MEFV
Alias(es): Pyrin, Marenostrin

Localization:

Reference sequences:

DNA: AJ003147 (EMBL) , **cDNA:** AF018080 (EMBL) , **Protein:** O15553 (SWISSPROT) Other Sequences

Chromosomal Location:

16p13

Maps:

MEFV (Map View)

Other gene-based resources:

Ensembl: ENSG00000103313, GENATLAS: MEFV, GeneCard: MEFV, UniGene: 173730, Entrez Gene: 4210, euGenes: 4210, GDB: 125263, HomoloGene: 32441

PROTEIN INFORMATION

Description:

Protein function:

Probably controls the inflammatory response in myelomonocytic cells at the level of the cytoskeleton organization.

Subcellular location:

Cytoplasmic, associated with microtubules and with the filamentous actin of perinuclear filaments and peripheral lamellar ruffles (isoform 1); nuclear (isoform 2).

Developmental stage:

First detected in bone marrow promyelocytes. Expression increases throughout myelocyte differentiation and peaks in the mature myelomonocytic cells.

Induction:

In monocytes, by treatment with colchicine and IFN-alpha, and by the proinflammatory cytokines IFN-gamma, TNF-alpha and 1ps. Repressed in monocytes by the antiinflammatory cytokines IL-10, TGF-beta and IL-4. In neutrophils, colchicine, TNF-alpha, lps, IL-10, INF-alpha and IL-4 has no effect on expression. INF-gamma increases expression in neutrophils.

Tissue specificity:

Expressed in peripheral blood leucocytes, particularly in mature granulocytes and to a lesser extent in monocytes but not in lymphocytes. Detected in spleen, lung and muscle, probably as a result of leucocyte infiltration in these tissues. Not expressed in thymus, prostate, testis, ovary, small intestine, colon, heart, brain, placenta, liver, kidney, pancreas. Expression detected in several myeloid leukemic, colon cancer, and prostate cancer cell lines.

Similarity:

Contains 1 b box-type zinc finger.

Domains:

DAPIN domain: 1-92

Other features:

Zinc finger region B box-type: 370-412

Other related resourses:

InterPro: IPR001870; B302, InterPro: IPR003879; Butyrophylin, InterPro: IPR004020; PAAD_DAPIN, InterPro: IPR003877; SPRY_receptor, InterPro: IPR000315; Znf_Bbox, Pfam: PF02758; PAAD_DAPIN, Pfam: PF00622; SPRY, Pfam: PF00643; zf-B_box, PRINTS: PR01406; BBOXZNFINGER, PRINTS: PR01407; BUTYPHLNCDUF, PROSITE: PS50824; DAPIN, PROSITE: PS50119; ZF_BBOX

Expression pattern for human:

Tissue	Exp. (%)	Clones
pooled	73.43	1:5774
blood	26.57	3:47876

Animal models:

Mouse:

MGD: ; 4

OTHER RESOURCES

Societies:

General:

- IPOPI, International Patient Organization for Primary Immunodeficiencies
- The Jeffrey Modell Foundation
- National Center for Biotechnology Information
- Immune Deficiency Foundation
- European Society for Immunodeficiencies

Disease specific:

- FMF community

Other information sources:

- Immunodeficiencies