Familial Mediterranean Fever

Genetics

-Gene: MEFV (Pyrin; 16p13)

-AR

Clinical findings/Dysmorphic features

-Type 1: recurrent short episodes of inflammation, serositis and fever; peritonitis, synovitis, pleuritis, pericarditis, meningitis; amyloidosis severe complication: if untreated --> renal failure

-Type 2: amyloidosis as first clinical manifestation in asymptomatic individual

Etiology

-Variant p.Met694Val in more than 90% of affected Jewish of North African origin

Pathogenesis

-Mutations result in increased IL-1 responsiveness --> increased inflammatory attacks

Genetic testing/diagnosis

-Sequencing: 75-90%; no In/Del reported

-Targeted analysis first: Armenian, Turkish, Arab, North African Jewish, Iraqi Jewish, AJ

Others

-Up to 25% of individuals with FMF have only one MEFV pathogenic variant identified

-If only one mutation: diagnosis of FMF can be confirmed by a 6-month trial of colchicine