Familial Mediterranean Fever (FMF)

Presented by
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What is FMF?

- Inherited condition
- Common in people of Armenian, Arabic, Turkish and Jewish descent (Mediterranean region); 1 in 250 to 1 in 1,000 people
- Manifests in painful episodes with fever (usually in childhood)
Symptoms

- Painful inflammation in the chest (lungs), abdomen or joints
- Fever, sometimes rash
- 12-72 hour episodes
- 1st episode in childhood, teenage years
- Untreated, can lead to amyloidosis and renal failure
- Type 2: amyloidosis as first clinical symptom
Classical Diagnosis

- Exclusionary
- History of repeated fever and inflammation
- Risk assessment
- Favorable response to continuous colchicine treatment
- Amyloidosis (for Type 2)
Classical Treatment

- No known cure
- Non steroidal anti-inflammatory drugs
- Colchicine (prevents inflammation and deposition of amyloid)
Molecular Genetics

- Autosomal recessive (Mendelian), although some compound heterozygous and heterozygous will manifest symptoms
- Mutation (specific) of the MEFV gene on chromosome 16, locus 16p13
- Highly penetrant
What Causes the Symptoms?

- MEFV gene codes for pyrin
- Pryin is a protein responsible for regulating inflammation
Gene\n
• Yes
• Classical diagnosis confirmation
• For non-classic or mild cases, additional sequence analysis (inside or outside exon 10)
• Carrier testing (for heterozygous parents)
• Prenatal diagnosis
Genetics Better Therapies?

- Yes, but still in managing the symptoms rather than treating the disease
- Anakinra (IL-1 receptor inhibitor)
- Research into other treatments
- Testing allows for better diagnosis
- Family planning
References

- http://healthyparent.com/FMF/genes.html