Familial Mediterranean Fever (FMF)

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Presented by
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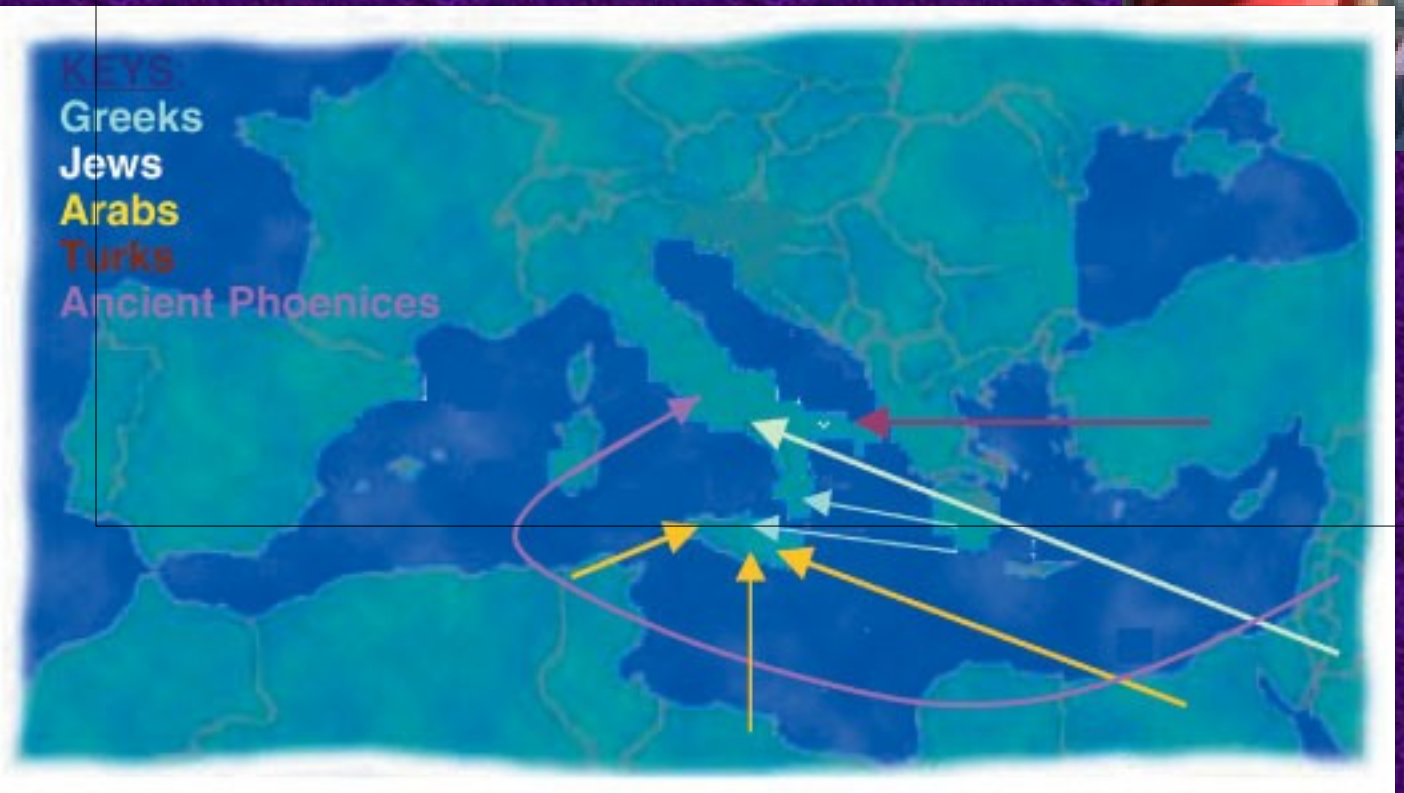
What is FMF?

- 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)

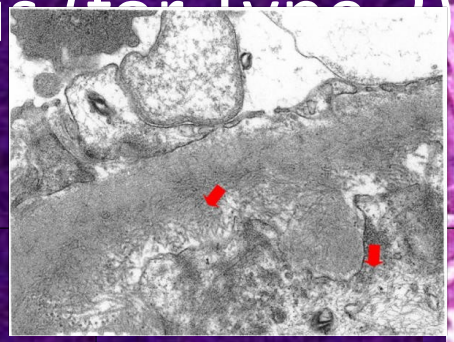


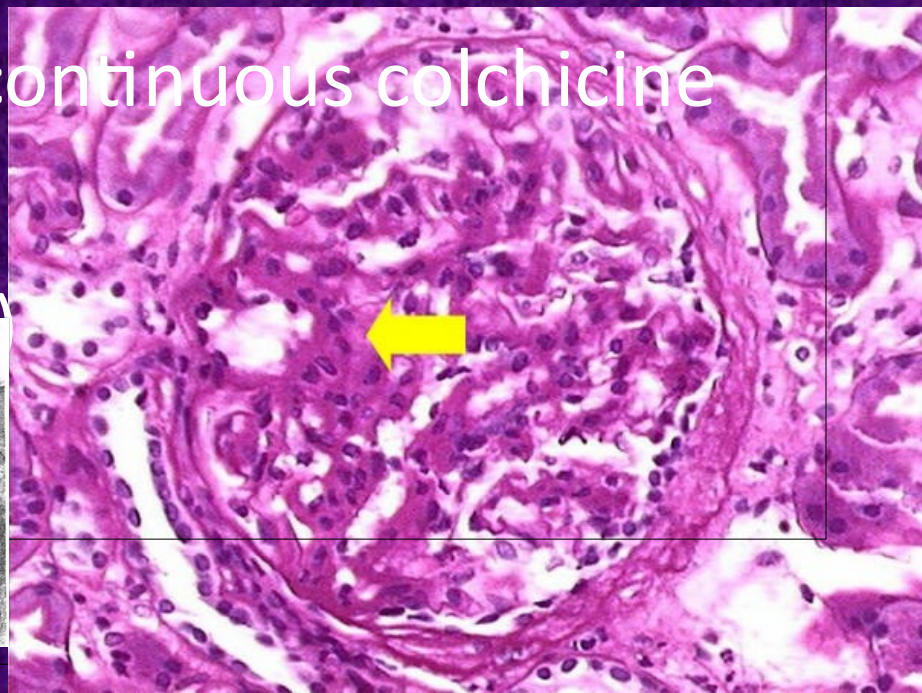
Symptoms

- 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)



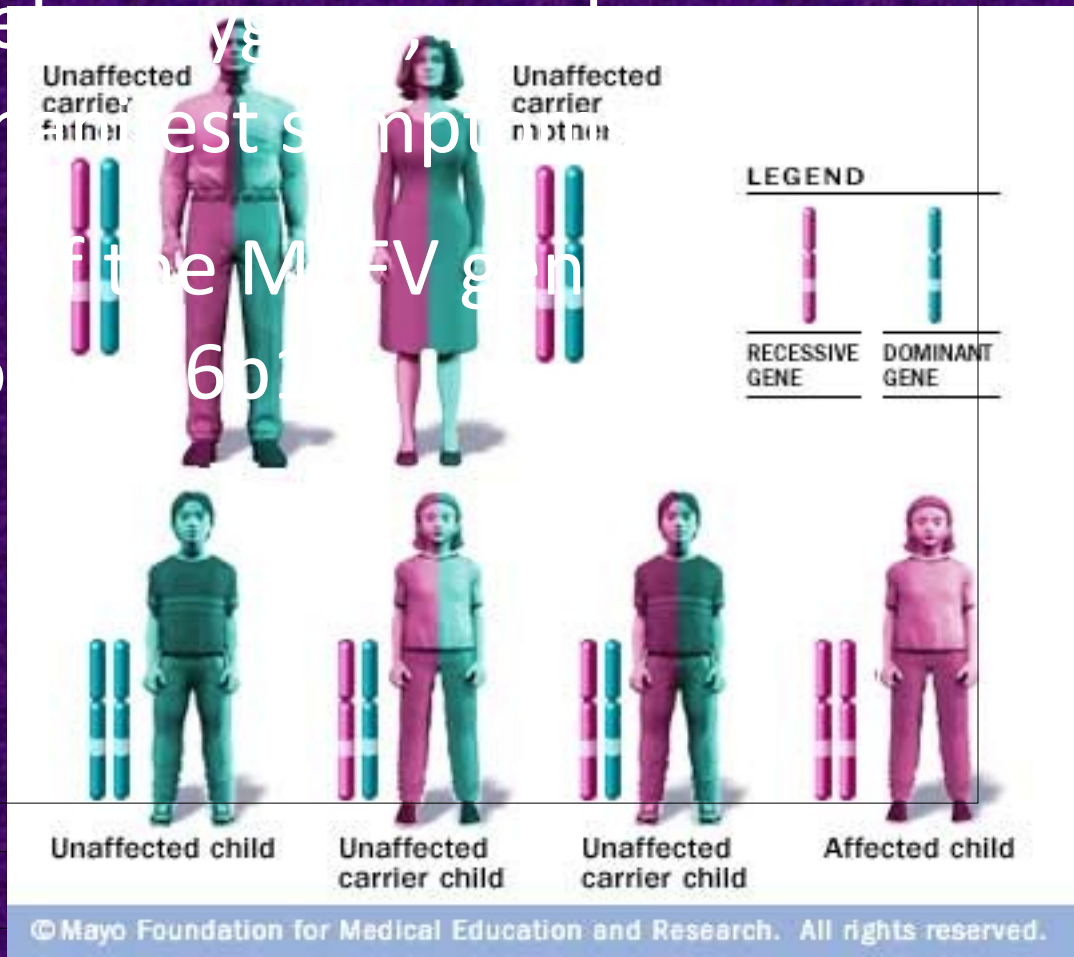


Classical Treatment

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

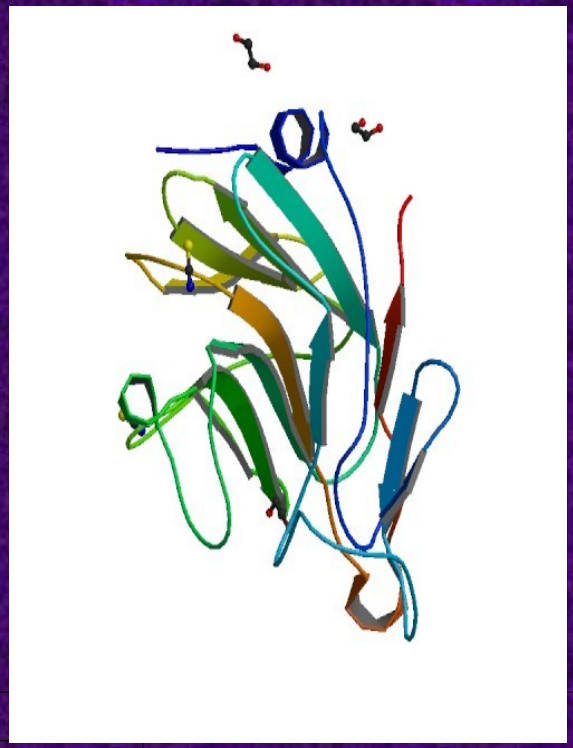
Molecular Genetics

- Autosomal recessive (Mendelian), although some compound he[illegible] heterozygous will m[illegible]est s[illegible]
- Mutation (specific) [illegible] the M[illegible]FV g[illegible] chromosome 16, lo[illegible]6p1[illegible]
- Highly penetrant



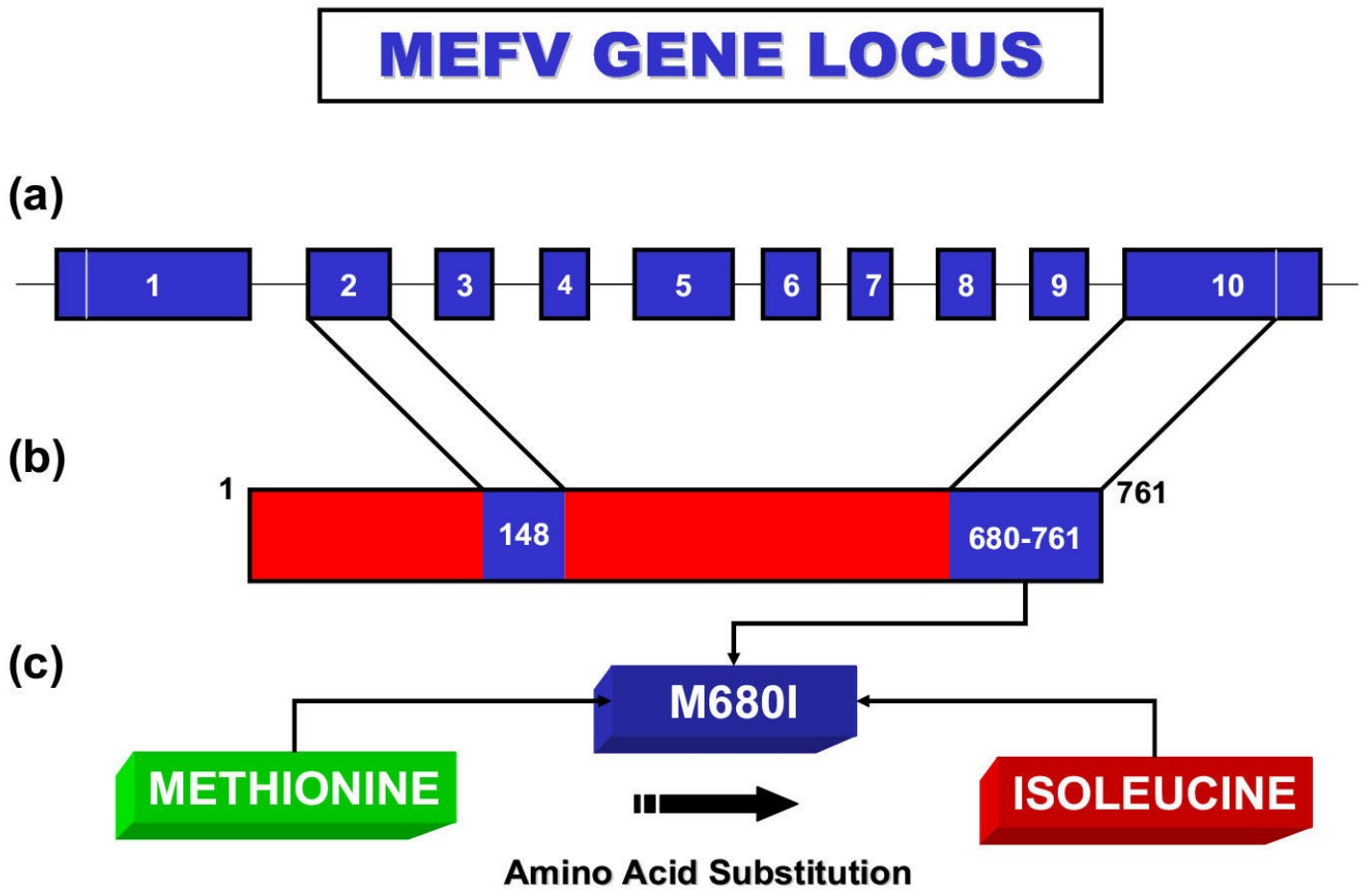
What Causes the Symptoms?

- MEFV gene codes for pyrin
- Pryin is a protein responsible for regulating inflammation



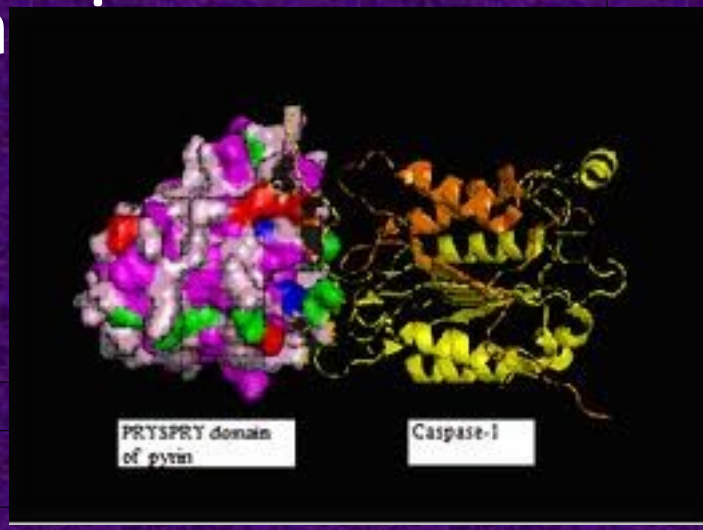
Genetics Better Diagnosis?

- Yes
- Classical diagnosis confirmation
- Targeted mutation analysis (Exon 2: p.Glu148Gln, Exon 10: p.Met694Val)
- For non-classic or mild cases, additional sequence analysis (inside or outside exon 10)
- Carrier testing (for heterozygous parents)
- Prenatal diagnosis


MEFV GENE LOCUS
(a)
1
2
3
4
5
6
7
8
9
10
(b)
1
148
680-761
761
(c)
M680I
METHIONINE
ISOLEUCINE
Amino Acid Substitution

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 diagn
- Family planning



References

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