Familial Mediterranean Fever

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FMF :

It’s characterized by auto inflammatory condition that causes recurrent fevers and painful inflammation of the abdomen, lungs, joints, etc.

It’s an autosomal recessive disease that mostly occurs in certain ethnic groups such as Arabs, Turks, Armenian, Sephardic Jews and those with Mediterranean origin.

Causes:

It’s caused by mutations in the MEFV gene located on the short arm of chromosome 16 ,encoding a protein called Pyrin.

Pyrin involves in the regulation of inflammation, by indirectly suppressing Caspase1 and the signaling of IL-1, resulting in inflammation suppression, and when it’s mutated, auto inflammatory attacks take place.

-Patients with FMF usually have their first attack before the age of 20.

-more in males.

-The frequency of the attacks may vary from several times in a month to once every few years.

-With attacks' duration ranging from several hours to several days.

Clinical features:

-Fever

-Abdominal pain, it’s the most common symptom happens in almost 95% of patients, varies in intensity and localization and it can mimic acute abdomens.

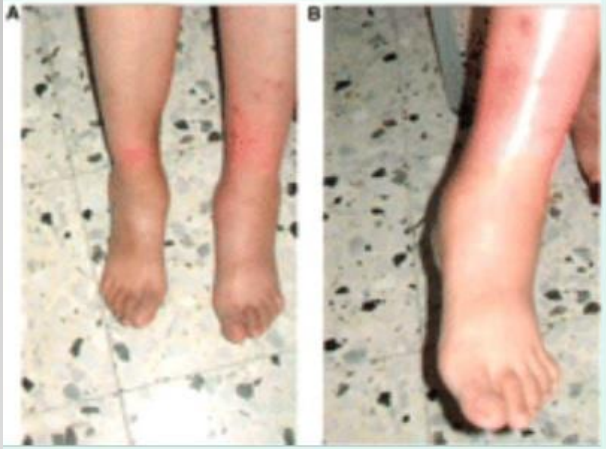
-Chest pain, reported in 30% of cases, presented mostly as plueritis and rarely as pericarditis.

-Joints: comes more as monoarthritis, involves mostly ankles and knees, it’s considered as non erosive arthritis and with no deformities.

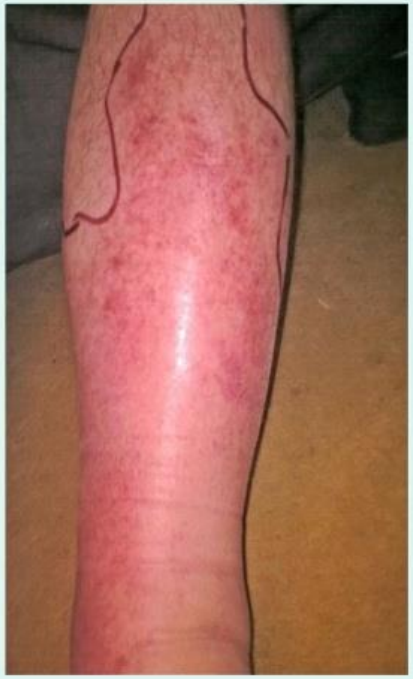
Manifested as hotness, redness, pain, and swelling on the joint.

-Skin: Red rash on legs especially below knees that can appear like erysipelas.

- Familial Mediterranean Fever



- Cellulitis



Diagnosis:

-History of similar attacks.

-Physical examination.

-Family History.

-Blood Test: (During attacks) Elevated WBC counts. elevated acute phase reactants in the serum (C-reactive protien, Amyloid A protien, ESR).

-Gene Test: it may detect the mutated gene but not reliable.

So, the diagnosis is based on clinical picture and exclusion of other diseases.

Complications:

-Amyloidosis:

Secondary to chronic inflammatory state that leads to deposition of abnormal proteins in tissue of many organs leading to organ failure.

AA type amyloidosis, which we have a deposition of Serum Amyoild A protein (Acute phase reactant).

Binds with other proteins (GAG,SAP) and deposits as insoluble protein fibrils.

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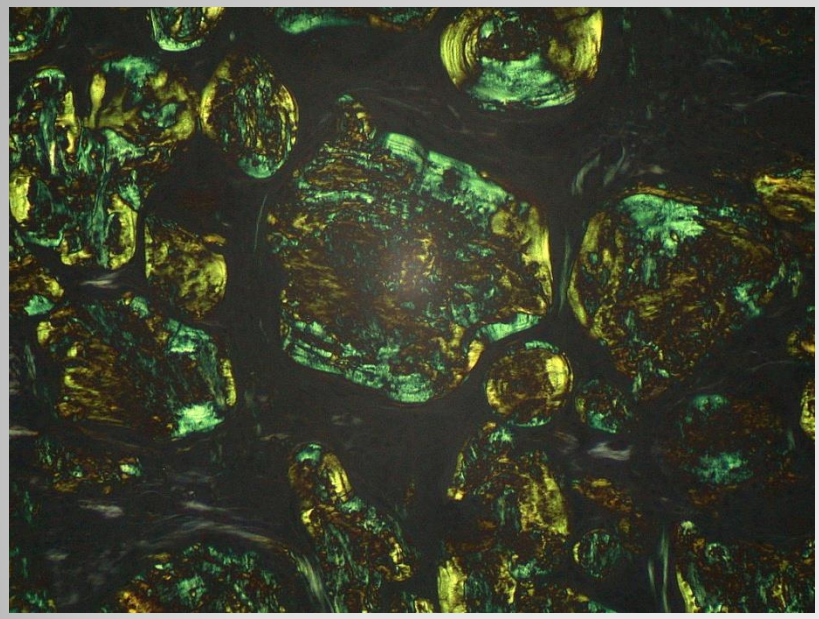
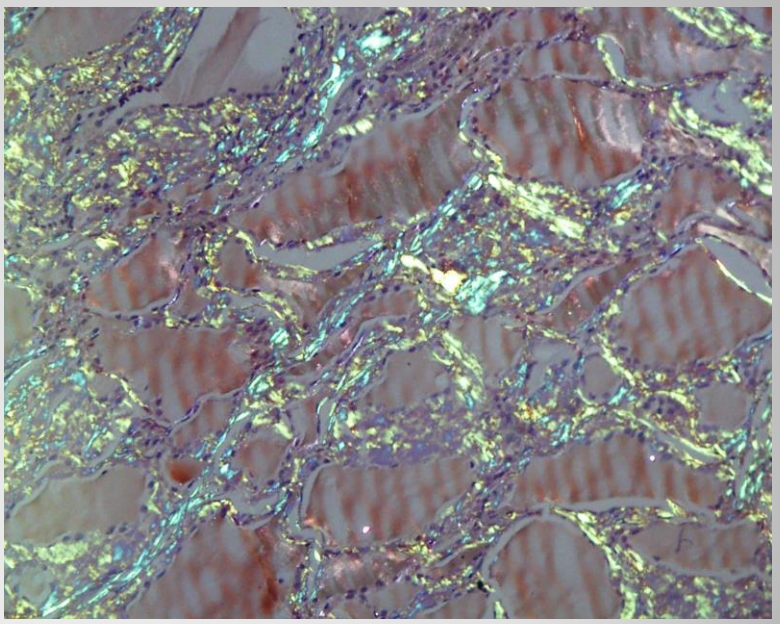
Deposition in the kidney, presenting as nephrotic syndrome or lately as a Kidney failure.

For diagnosis, biopsy is required, can be from the affected organ, rectal, or subcutaneous fat biopsies.

Can use scientigraphy with radio-labelled SAP to check for the overall loading and distribution of the amyloid deposits.

The pathgnomonic histological feature is Apple-Green Birefringence of amyloid deposits when stained with Congo red dye and viewed under polarized light.





Other complications :

-Increase the risk or vasculitis-related diseases (Henoch-schonlein pupura).

-Prolonged Arthritis of certain joints.

Treatment:

There's no cure for familial Mediterranean fever. However, treatment can help prevent signs and symptoms.

The Drug of choice is Colchicine, used mainly to reduce the Frequency of the attacks and the build up of amyloids, but it’s ineffective once the symptoms start, so it’s given to prevent not to suppress the attack.

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In cases of resistance, other treatments are indicated such as Anakinra, Canakinumab.

For amyloidosis, Epridosate has been proven to reduce the renal deterioration,

It’s designed to interfere between the amyloidogenic proteins and GAG.

Analgesics and NSAIDs are indicated as well.

References:

-Kumar and clarck
-Davidson
-Mayoclinic.org
-WiKi
-Nejm.orj

THANK YOU