

HAEMOCHROMATOSIS

4th year MBCHB 2021

Haemochromatosis

What is it?

- ❖ This is a disorder where too much iron builds up in the body →
Iron overload
- ❖ The body absorbs too much iron & has no way to get rid of it.

Two types exist:

- ❖ 1^o haemochromatosis:
Is hereditary, runs in families & one gets genes from both parents
- ❖ 2nd. Haemochromatosis: Secondary to other conditions

Haemochromatosis: Presentation

- ❖ In males symptoms occur at ages 30-50
- ❖ In females occurs >50yrs

Symptoms include:-

- ❖ Joint pains (arthralgia) 44%
- ❖ Fatigue 74%
- ❖ Unexplained weight loss
- ❖ Skin with grey or brown coloration
- ❖ Loss of sex drive (impotence) 45%
- ❖ Heart: Flutter
- ❖ Memory: Foggy
- ❖ Abdominal pain
- ❖ **75% are asymptomatic**

Haemochromatosis: Clinical manifestation

- ❖ Liver disease:

 - Hepatomegaly. 13%

 - Cirrhosis. 13%

- ❖ Skin bronzing & hyperpigmentation: 70%

- ❖ Diabetes mellitus 48%

- ❖ Arthropathy

- ❖ Amenorrhea, impotence, hypogonadism

- ❖ Cardiomyopathy

- ❖ Osteopenia & osteoporosis

- ❖ Hair loss

- ❖ Koilonychia

Haemochromatosis: Organs most involved:

- ❖ Liver: Cirrhosis
- ❖ Pancreas: Diabetes
- ❖ Heart: Arrhythmias/ Failure
- ❖ Joints: Arthritis
- ❖ Sex organs: Erectile dysfunction

Haemochromatosis: Diagnosis

Tests to detect iron overload:

1) Serum transferrin saturation:

It measures the amount of iron bound to a protein (transferrin)
levels >45% significant

2) Serum ferritin:

Measures the amount of iron stored in the liver

Both blood tests are abnormal in haemochromatosis

3) Additional tests:

Haemochromatosis: Diagnosis

Additional tests:

- 1) Liver biochemistry: shows liver damage
- 2) MRI: More sensitive > CT: Noninvasive way to measure iron overload in the liver
CXR & Echocardiogram & ECG to exclude cardiac disease
- 3) Genetic testing: Test for gene mutation:
 - ❖ Test DNA for mutation in the HFE gene : Two most common are: -C282Y & H63D: They are pivotal in diagnosis of haemochromatosis
 - ❖ Genetic testing is also recommended for healthy people for haemochromatosis
 - ❖ Genetic testing is recommended for all 1st. Degree relatives (parents, siblings, children of anyone diagnosed with haemochromatosis)

NOTE: If mutation is found in only one parent then children don't need testing

Haemochromatosis: Treatment:

- ❖ Phlebotomy on regular basis, to reduce level of iron

- ❖ Chelation:

 - Deferoxamine, Deferasirox

- ❖ Liver transplant in liver failure