Primary haemochromatosis at a glance

Epidemiology

Prevalence

0.4%, but the prevalence of identified cases is much less

Age

Usually 40–60 years of age

Sex

Males usually present before females

Genetics

Autosomal recessive

Fewer than 5% with genetic haemochromatosis develop iron overload

Investigation

Biochemistry

Liver tests may be normal

Serum ferritin is increased, often exceeding 1000 μg/l

Serum iron is high

Iron saturation (serum iron : total iron binding capacity) exceeds 75%

Liver iron exceeds 180 μg/g liver (dry weight)

Genetics: shows *HFE* genes in >95% cases

Genetics

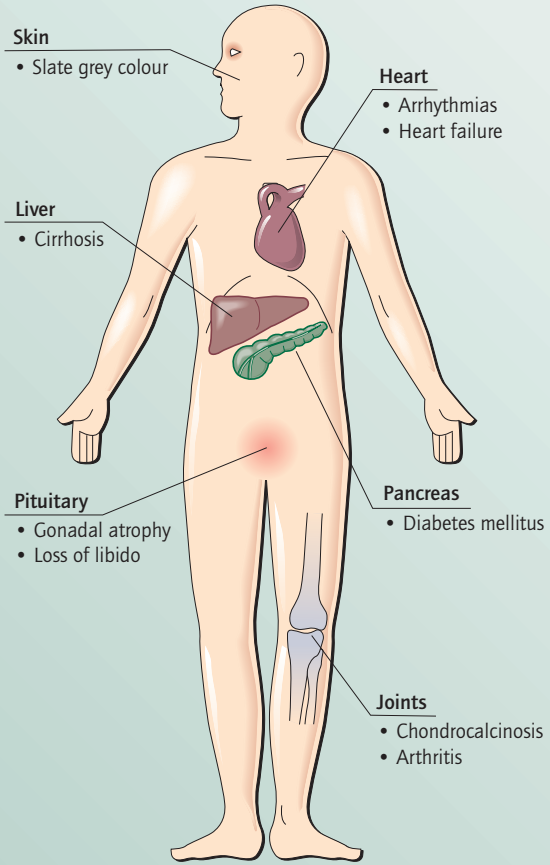
Recognized mutations in *HFE* are C282Y and H63D

Histopathology

Liver biopsy may reveal an established cirrhosis. There is excess iron deposition in the hepatocytes and biliary epithelial cells

Management

Venesection



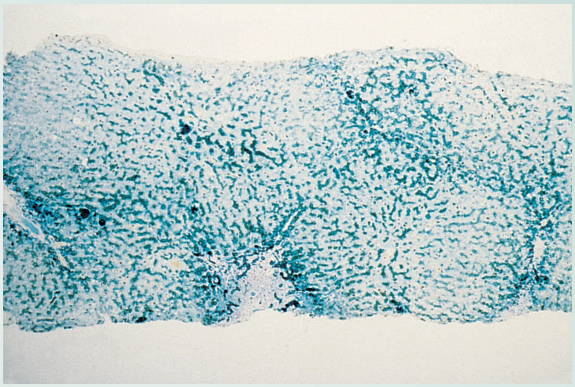


Fig. A Perls' stain showing grade 4 siderosis with normal architecture.

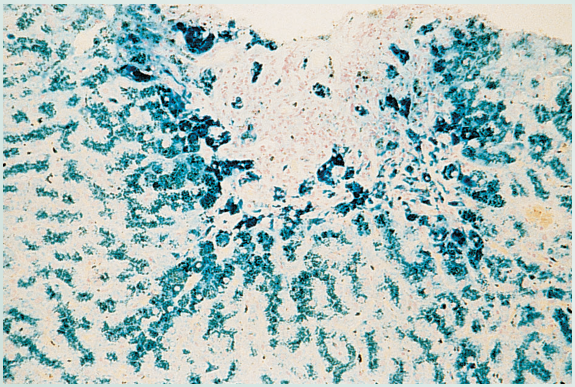


Fig. B Perls' stain showing periportal deposits, heavier pigment and biliary epithelium.