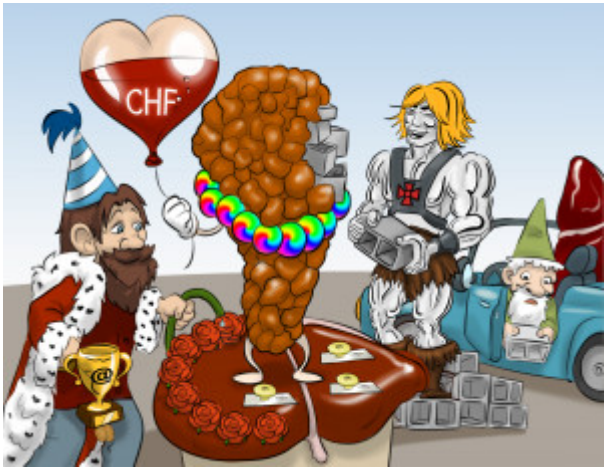
Hemochromatosis Signs and Symptoms

Hemochromatosis is a disease characterized by excessive accumulation of iron in the body, most of which becomes deposited in organs such as the liver, pancreas, joints and heart. Primary hemochromatosis is an autosomal recessive disorder caused by a gene mutation on chromosome 6 that causes excessive iron absorption. Accumulation of iron in tissues can also occur as a consequence of parenteral administration of iron, often secondary to multiple transfusion therapies. In primary hemochromatosis, the regulation of intestinal absorption of dietary iron is highly abnormal, leading to net iron accumulation of 0.5 to 1.0 gm/year. The increase in iron causes increase in ferritin stores as well as a decrease in transferrin. Excessive iron accumulation is directly toxic to host tissue due to formation of reactive oxygen species and stimulation of collagen formation. In the liver, the excessive iron accumulation can lead to lethal cell injury with predisposition for micronodular cirrhosis and hepatocellular carcinoma. Deposition of hemosiderin in the pancreas can cause diabetes and deposition in the skin can cause bronze skin discoloration. The heart, joints, and testes are also commonly affected leading to congestive heart failure, arthropathy, and testicular atrophy causing impotence.



PLAY PICMONIC

Signs and Symptoms

Micronodular Cirrhosis
Micro-knobs with C-roses-on-liver
The liver is the organ most severely involved in hemochromatosis. Initially, the hemosiderin is deposited in the cytoplasm of hepatocytes, and with increasing accumulation, there is progressive involvement of the lobes along with the bile duct epithelium. Fibrous septa can develop in the liver, leading to a micronodular pattern of cirrhosis.

Hepatocellular Carcinoma
Liver Car-gnome
Excessive iron accumulation is directly toxic to host tissue due to the formation of reactive oxygen species and stimulation of collagen formation. In the liver, excessive iron accumulation can lead to lethal cell injury with a predisposition for micronodular cirrhosis and hepatocellular carcinoma. Hepatocellular carcinoma is a significant cause of death in these individuals, with a 200-fold greater risk than in the general population.

Diabetes
Dyed-bead-pancreas
Hemosiderin is deposited in the pancreas in both the acinar and islet cells and can cause diffuse interstitial fibrosis. Destruction of the pancreatic islet cells can cause deranged glucose homeostasis or frank diabetes mellitus.

Bronze Skin Coloring
Bronze-statue
Skin pigmentation is a classic feature of hemochromatosis caused by both hemosiderin deposition in dermal macrophages and increased epidermal melanin production.

CHF
CHF Heart-balloon
The heart is often enlarged due to hemosiderin deposition and can lead to congestive heart failure.

Arthropathy
King-Arthur-party-hat
Excess iron can deposit in the joint synovial linings as well as articular cartilage, causing severe arthropathy.

Testicular Atrophy
Testicle @-trophy
The testes are commonly affected and can become small and atrophic, although usually not significantly pigmented. Testicular atrophy can lead to symptoms including impotence.

Impotence
Limp-hose
Impotence is defined as the failure to achieve and sustain an erection sufficient for mutually satisfactory intercourse and can be caused by hemosiderin deposition in the testes.