Hemochromatosis Pathophysiology

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. Though the transferrin level decreases, the transferrin saturation % increases. 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.



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Pathophysiology

Hemosiderin Deposition
He-man-chrome depositing cinder blocks
Hemosiderin is an iron storage complex found in cells as opposed to circulating in the blood. Hemosiderin is usually found in macrophages and abundant in situations following hemorrhage but can accumulate in different organs in hemochromatosis.

Autosomal Recessive
Recessive-chocolate
Primary hemochromatosis is caused by a mutation in the HFE gene on chromosome 6. This mutation is inherited in an autosomal recessive fashion.

HLA-A3
HuLA (A) Apple (3) Tree
There is a higher frequency of HLA A3 serotypes in individuals with hemochromatosis.

Secondary to Transfusion Therapy
Transfusion-IV
Excessive iron accumulation can also occur secondary to parenteral iron administration for example with multiple transfusion therapies. Individuals at high risk for secondary hemochromatosis include individuals with chronic blood disorders like beta thalassemia major.

Increase Iron
Up-arrow Iron
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.

Increase Ferritin
Up-arrow Ferret-tin
Ferritin is a protein that stores and releases iron. Because hemochromatosis is characterized by increased iron absorption, there is increased ferritin levels in the body.

Increased Transferrin Saturation
Up-arrows on Train-ferrets %
Transferrins are blood plasma glycoproteins that bind iron and control the level of free iron in biological fluids. When there are increased iron stores in the body, transferrin levels decrease. Furthermore, an increase in iron means that transferrin is fully saturated, leading to a decreased total iron binding capacity in the blood (TIBC). Thus, patients have decreased transferrin levels, decreased TIBC, but increased transferrin saturation percentage.

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