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Genetic disorders affecting proteins of iron and copper metabolism: clinical implications

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Abstract

Iron and copper are essential transition metals that permit the facile transfer of electrons in a series of critical biochemical pathways. Recent work has identified the specific proteins involved in the absorption, transport, utilization, and storage of iron and copper. Remarkable progress is being made in understanding the molecular basis of disorders of human iron and copper metabolism. This review describes these proteins and examines the clinical consequences of new insights into the pathophysiology of genetic abnormalities affecting iron and copper metabolisms. Hereditary hemochromatosis is the most common genetic disorder of iron metabolism caused by mutations in the HFE gene. Aceruloplasminemia is a rare iron metabolic disorder that results from deficiency of ceruloplasmin ferroxidase activity as a consequence of mutations in the ceruloplasmin gene. Menkes disease and Wilson's disease are inherited disorders of copper metabolism resulting from the absence or dysfunction of homologous copper-transporting ATPases.

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