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Inherited metabolic disorders of copper transport —Clinical features and pathophysiology of Wilson disease, Menkes disease and aceruloplasminemia—

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Abstract:
Copper is an essential trace metal. Proteins exploit the unique redox nature of this metal to undertake a series of facile electron transfer reactions using copper as a cofactor in a select number of critical enzymatic pathway. The signs and symptoms of copper deficiency are the results of impaired function of these cuproenzymes.
Copper homeostasis is maintained entirely by gastrointestinal absorption and biliary excretion. Biliary copper is not absorbed from the gastrointestinal tract, so there is no enterohepatic circulation of this metal.
Wilson disease and Menkes disease are inherited metabolic disorders of copper transport. Each disease results from the absence or dysfunction of homologous copper-transporting ATPase present in the trans-Golgi networt of cells. The Wilson disease ATPase transports copper into the hepatocyte secretary pathway for incorporation into ceruloplasmin and excretion into the bile. Thus, individuals with this autosomal recessive disease present with signs and symptoms arising from impaired biliary copper excretion.
The Menkes disease ATPase transports copper across the placenta, gastrointestinal tract, and blood-brain barrier, and the clinical features of this X-linked disease arise from copper deficiency. Despite striking differences in the clinical presentation of these two disease, the respective ATPase function in precisely the same fashion within the cell. The unique clinical features of each disease are the results of the tissue specific expression of these ATPase.
Aceruloplasminemia is an autosomal recessive disease characterized by absent serum ceruloplasmin and progressive neurodegeneration of the basal ganglia in association with specific inherited mutation in the ceruloplasmin gene. Although the basal ganglia symptoms and a lack of serum ceruloplasmin may lead to diagnostic confusion with Wilson disease,

magnetic resonance imaging reveals the presence of iron the basal ganglia.

Key words: ATP-7A, ATP-7B

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