ONLINE ISSN : 1880-1404
PRINT ISSN : 0916-717X

Biomedical Research on Trace Elements
Vol. 18 (2007) , No. 3 249-254

[PDF (1312K)] [References]

Copper Metabolism and Copper Transport Disorders

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(Received: October 4, 2006)
(Accepted: February 16, 2007)

Abstract:
Genetic disorders of copper metabolism are reviewed, particularly in Menkes disease and Wilson's disease. The responsible genes for Menkes disease and Wilson's disease are *ATP7A* and *ATP7B*, respectively, with the both proteins responsible for transporting copper from the cytosol to the Golgi apparatus in cells. However, the pathology of Menkes disease is completely different from that of Wilson's disease, with Menkes disease characterized by a copper deficiency and Wilson's disease by a toxic excess of copper. The reason for this difference is related to the particular cell types in which the ATP7A and ATP7B proteins are expressed. ATP7A is expressed in almost all cells, except hepatocytes, where ATP7B is expressed in hepatocytes.
Menkes disease is an X-linked recessive disorder characterized by copper deficiency. The typical features, such as neurological disturbances, connective tissue disorders and hair abnormalities, can be explained by the abnormally low activity of copper-dependent enzymes. The treatment is so far parenteral administrations of copper-histidine. When the treatment is started in patients more than 2 months of age, however, the neurological disturbances cannot be improved. Moreover, the treatment does not improve the connective tissue disorders. Thus an alternative treatment needs to be found.
Wilson's disease is an autosomal recessive disorder characterized by the toxic effects of copper. The clinical symptoms mainly appear as diseases of the liver and nervous system. However, various other symptoms can also be observed and can sometimes make an early diagnosis difficult. All the patients should be treated with chelating agents or zinc. However, the treatments are ineffective in patients with fulminant hepatic failure. Liver transplantation is accepted for these patients. In these cases also, the disturbances are prevented by early treatments. Thus, early diagnosis is important. Screening in early infants should be established for early diagnosis of this disease.

Key words: Menkes disease, occipital horn disease, Wilson's disease, ATP7A, ATP7B, copper

To cite this article:

Hiroko Kodama, Chie Fujisawa, Yan-Hong Gu and Katsuaki Shiga, “Copper Metabolism and Copper Transport Disorders”, Biomedical Research on Trace Elements, Vol. **18**, pp.249-254 (2007) .

JOI JST.JSTAGE/brte/18.249