HEB

A REVIEW ON WILSON'S DISEASE

JOHP

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ABSTRACT

Wilson's disease is a gentic disorder in which copper biluds up in the body and is an autosomal recessive condition due to mutation in the ATP7B . Symptoms are typically related to the brain & liver . Liver related symptoms include vomitings ,weakness , fluid buildup in the abdomen , swelling of legs, yellowish skin & Itchiness . Brain related symptoms include tremors, muscle stiffness ,trouble speaking ,personality changes , anxiety. Diagnosis include liver function test , Magnetic Resonance Imaging (MRI), Liver biopsy , CT scan , Urinary copper excretion, serum copper , serum ceruloplasmin ,liver copper concentration . Treatment includes dietary changes , chelating agents, zinc supplements , liver transplantation

KEY WORDS: Mutation , ATP7B , chelating agents , serum ceruloplasmin.

Introduction

Wilson's disease is a genetic disorder in which copper builds up in the body &it is an autosomal recessive condition due to mutation in Wilsons disease protein gene (ATP7B) For a person to effect they must inherit and effected copy of gene from each parent . Wilson's disease are also known as hepatolenticular degeneration . The Wilsons disease gene (ATP7B) has been mapped to chromosome 13 (13q14.3) & is expressed primarily liver ,kidney &placenta .

Signs and symptoms :

Signs and symptoms are related to the brains and liver

Liver:

- Vomiting
- Weakness
- Fluid buildup in the abdomen
- Swelling of legs
- Yellowish skin
- Itchiness

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