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what's Wilson disease

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Introduction

Wilson disease is an inherited autosomal recessive disease which results in an excess copper build up in the body. It primarily affects the liver and basal ganglia of the brain, but it can affect other organ systems too
There are about 30000 cases world wide, it effect males and females equally, but females are more likely than males to develop acute liver failure due to Wilson disease

Signs and symptoms

The signs and symptoms of Wilson's disease vary widely, depending on which organ is affected. It can be:
Liver-related
The following symptoms may indicate copper accumulation in the liver:
weakness
feeling tired
weight loss
nausea
vomiting
loss of appetite

Neurological
Copper accumulation in the brain can cause symptoms such as:
memory, speech, or vision impairment
abnormal walking
migraines
drooling
Insomnia
Other symptoms
The buildup of copper in other organs can cause:
bluish discoloration in the nails
kidney stones
low blood pressure

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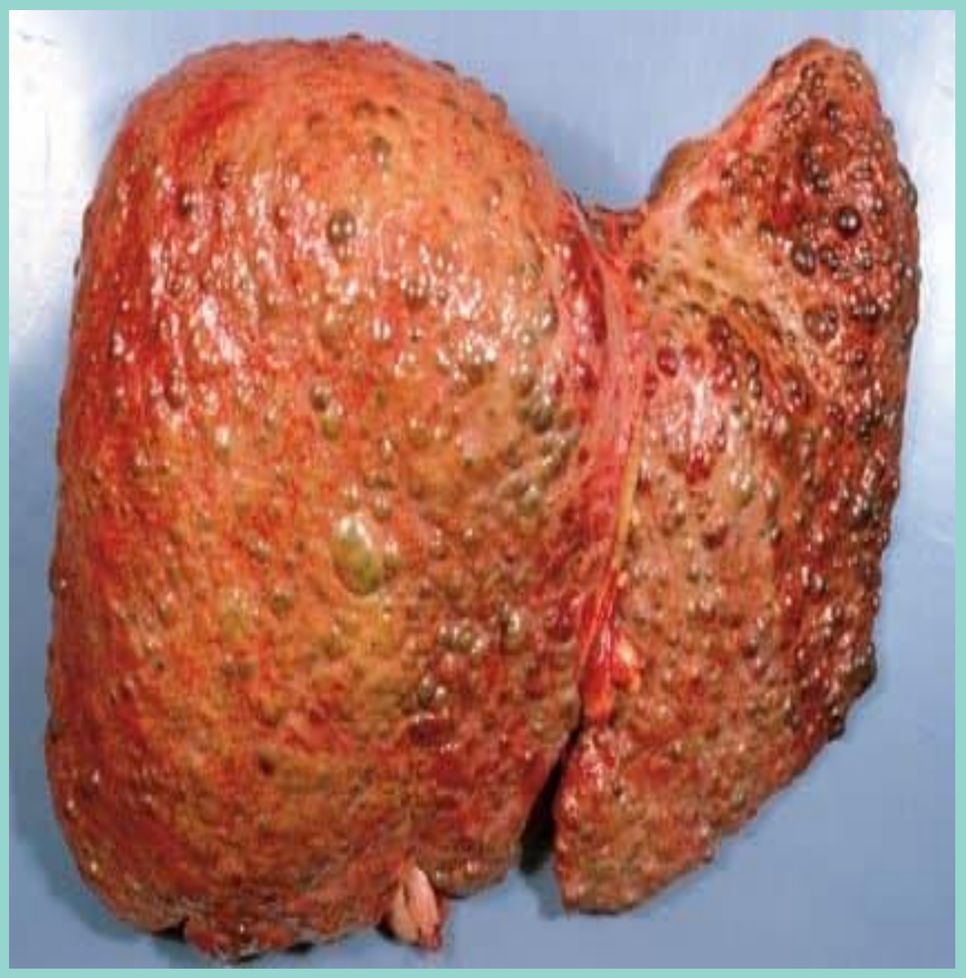

Causes

Wilson disease is inherited as an autosomal recessive. It caused by disruption or changes (mutations) of the ATP7B gene, which plays an important role in the movement of excess copper from the liver to the bile to eventually be excreted from the body through the intestines. More than 300 different mutations of the ATP7B gene have been identified.
males and females in equal numbers. The disease is found in all races and ethnic groups. occurs in approximately one in 30,000 to 40,000 people worldwide. Approximately one in 90 people may be carriers of the disease gene.

diagnosis

Blood and urine tests. Blood tests can monitor your liver function and check the level of a protein that binds copper in the blood (ceruloplasmin) and the level of copper in your blood.
Eye exam. Using a microscope with a high-intensity light source (slit lamp), an ophthalmologist checks your eyes for Kayser-Fleischer rings, which is caused by excess copper in the eyes.
Liver biopsy: laboratory tests the tissue for excess copper
Genetic testing: A blood test can identify the genetic mutations that cause Wilson's disease.
Ultrasound: which show accumulation of copper in liver(cause liver crihosses and liver failure) and brain

treatment

The goal of treatment is to preventing copper from building up again.
The drugs that used in treatmentis:
Penicillamine (Cuprimine, Depen).
Trientine (Syprine).
Zinc acetate (Galzin)

Surgery:
If your liver damage is severe, you might need a liver transplant. During a liver transplant, a surgeon removes your diseased liver and replaces it with a healthy liver from a donor.

reference

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