

# Wilson's Disease





# Summary



Genetic disorder, where there is too much copper in the body.

Caused by a mutation located on the 13th chromosome called ATP7B

Autosomal recessive disease

Wilson's disease first attacks the liver, the central nervous system, or both.

Treatment : Drugs, Zinc acetate, liver transplant or healthy diet.

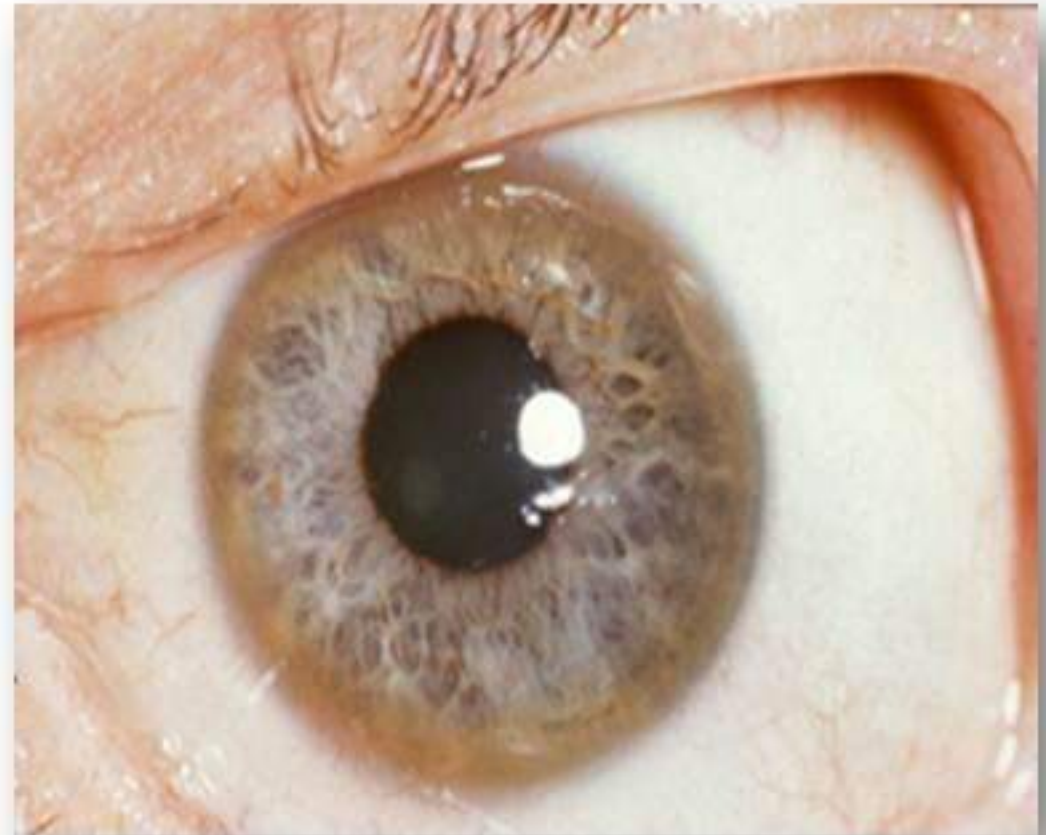


# Definition



Wilson's disease is a genetic disorder, where there is too much copper in the body.

Small amounts of copper are needed in the body, but too much can be hazardous to the liver, brain, eyes, and other organs.



# Hepatolenticular degeneration



Another name for Wilson's disease is Hepatolenticular degeneration.

The special proteins in the liver are called Hepacytes

Degeneration of the liver and the central nervous system.

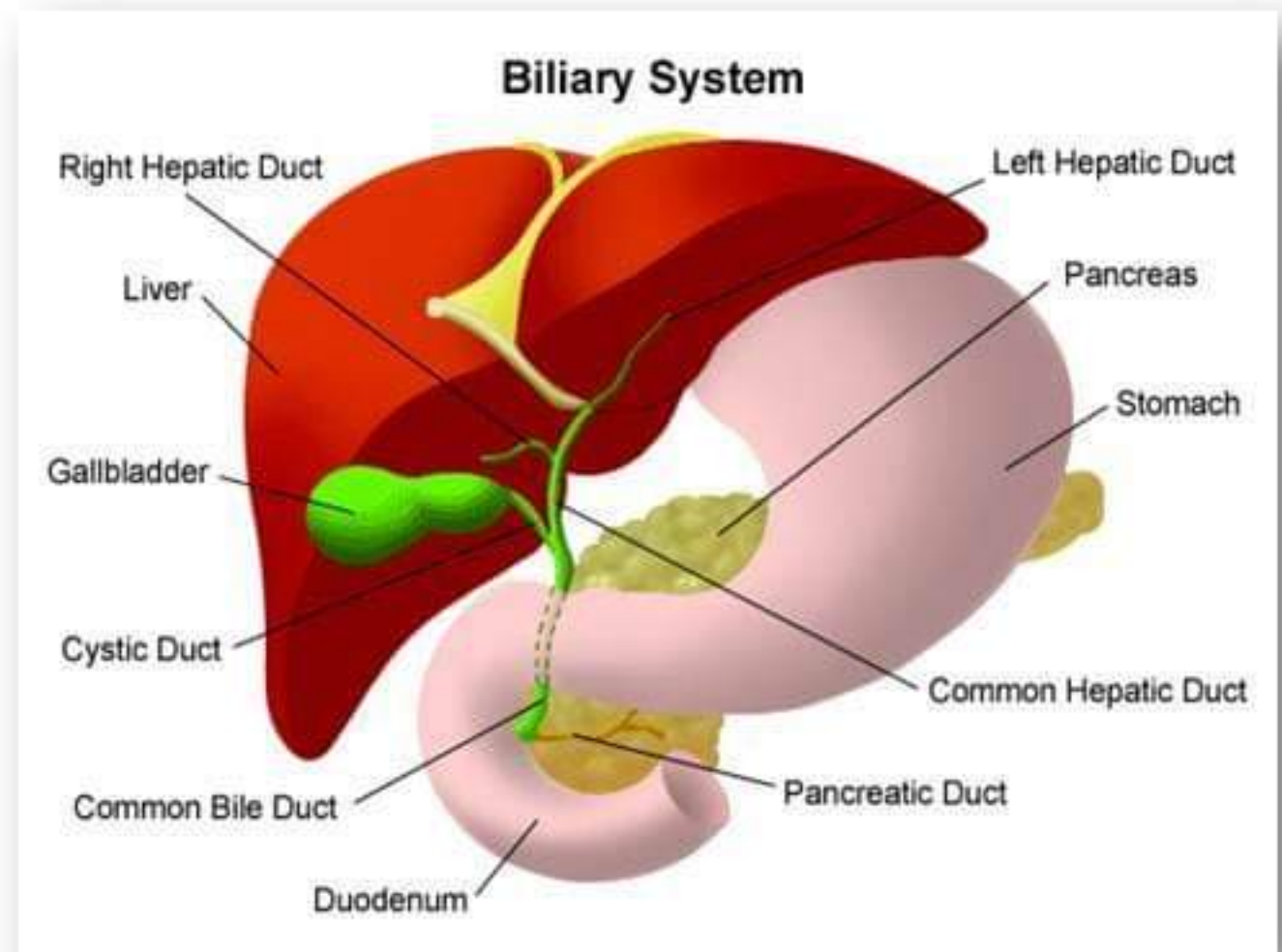


# Bile



Copper is absorbed from your food, and any excess is excreted through bile, required for the digestion of food, is excreted by the liver into passages

In Wilson disease, copper does not pass from the liver into the bile, but rather begins to accumulate within the liver.



# How it was found



Wilson's disease is named after Dr. Wilson, a doctor at the National Neurology Hospital in London.

He discovered the disease in 1911





# Mutations



Wilson's disease is caused by a mutation located on the 13th chromosome called ATP7B.

Mutations in the ATP7B gene produce a protein with decreased ability to bind copper.

# Inheritance



Autosomal recessive disease

In order to inherit Wilson disease, both parents must carry one genetic mutation

Wilson disease carriers, who have only one copy of the abnormal gene, do not have symptoms



# Symptoms



Wilson's disease first attacks the liver, the central nervous system, or both.

Swelling of the liver or spleen

Jaundice

Fluid buildup in the legs or abdomen

Fatigue

a tendency to bruise easily



# Treatment



**Drugs : d-penicillamine and trientine**

**hydrochloride** → Both of these drugs release copper from organs into the bloodstream. Most of the copper is then filtered out by the kidneys and excreted moment.

Treatment for Wilson disease is a life long process

**Zinc acetate** → prevents your body from absorbing copper from the food you eat.

**Liver Transplant** → removes your diseased liver and replaces it with a healthy liver from a donor.

A tendency to bruise easily