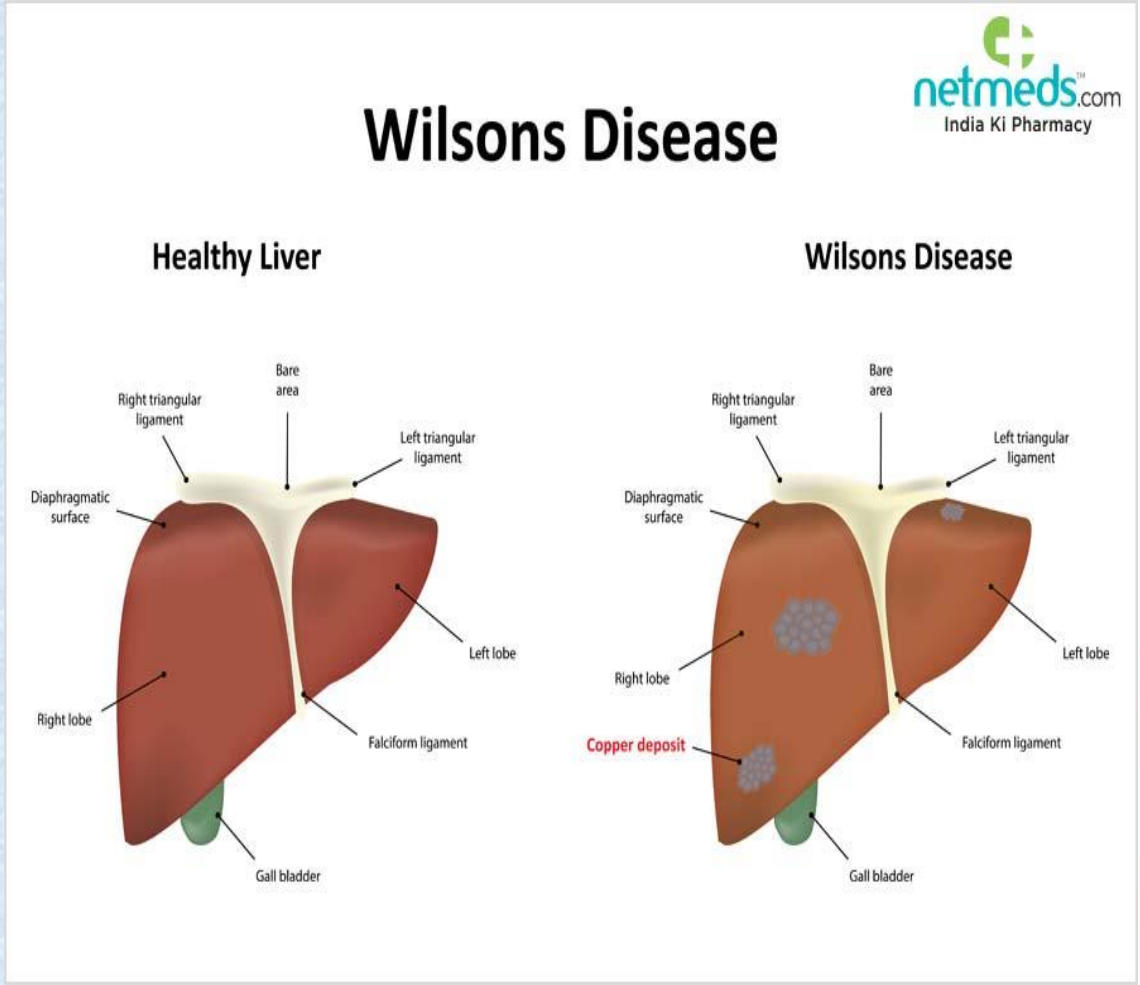
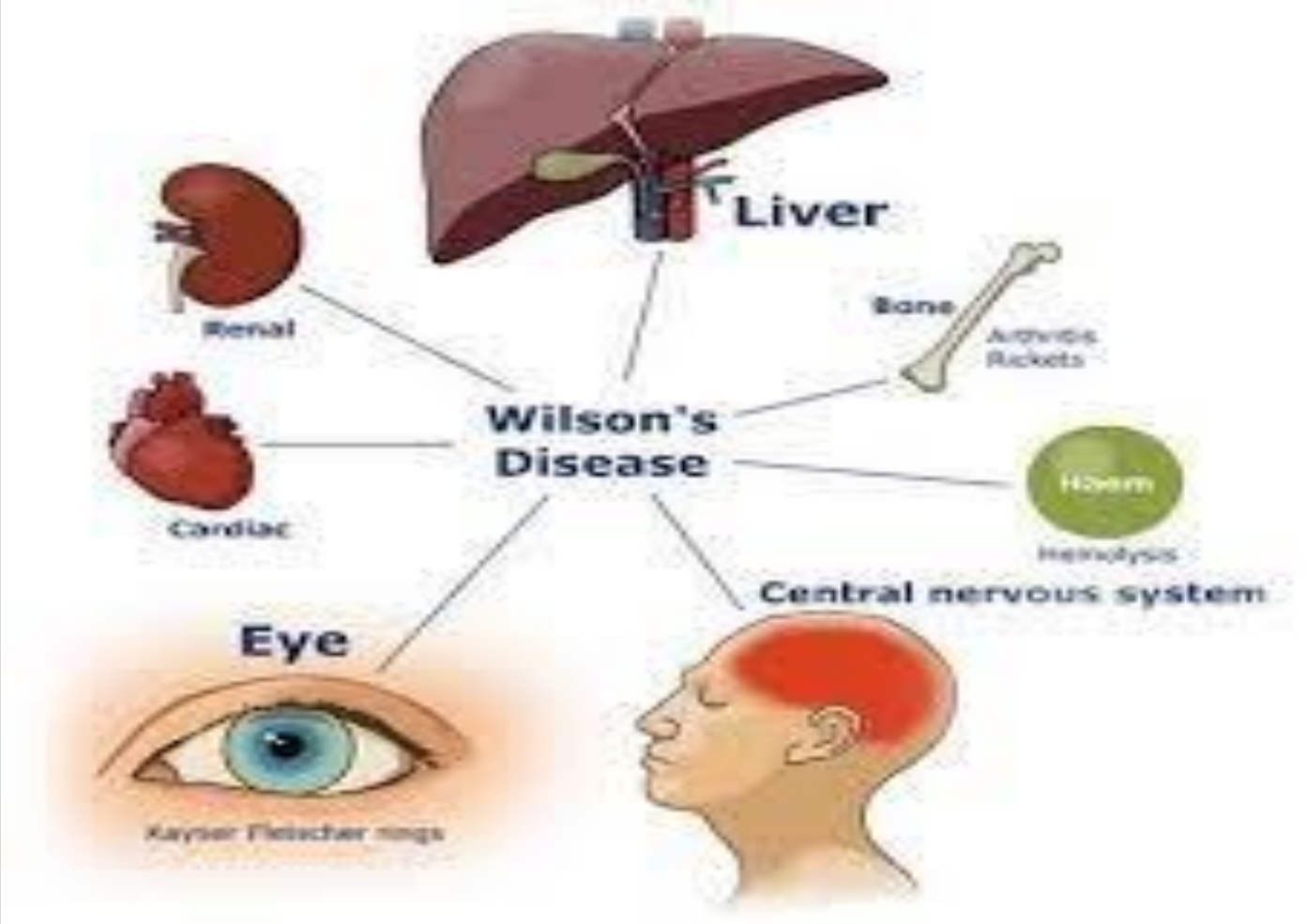
Assistant prof. Dr. heba fadhil hassan

- Synonyms of Wilson Disease
  - hepatolenticular degeneration
  - lenticular degeneration, progressive



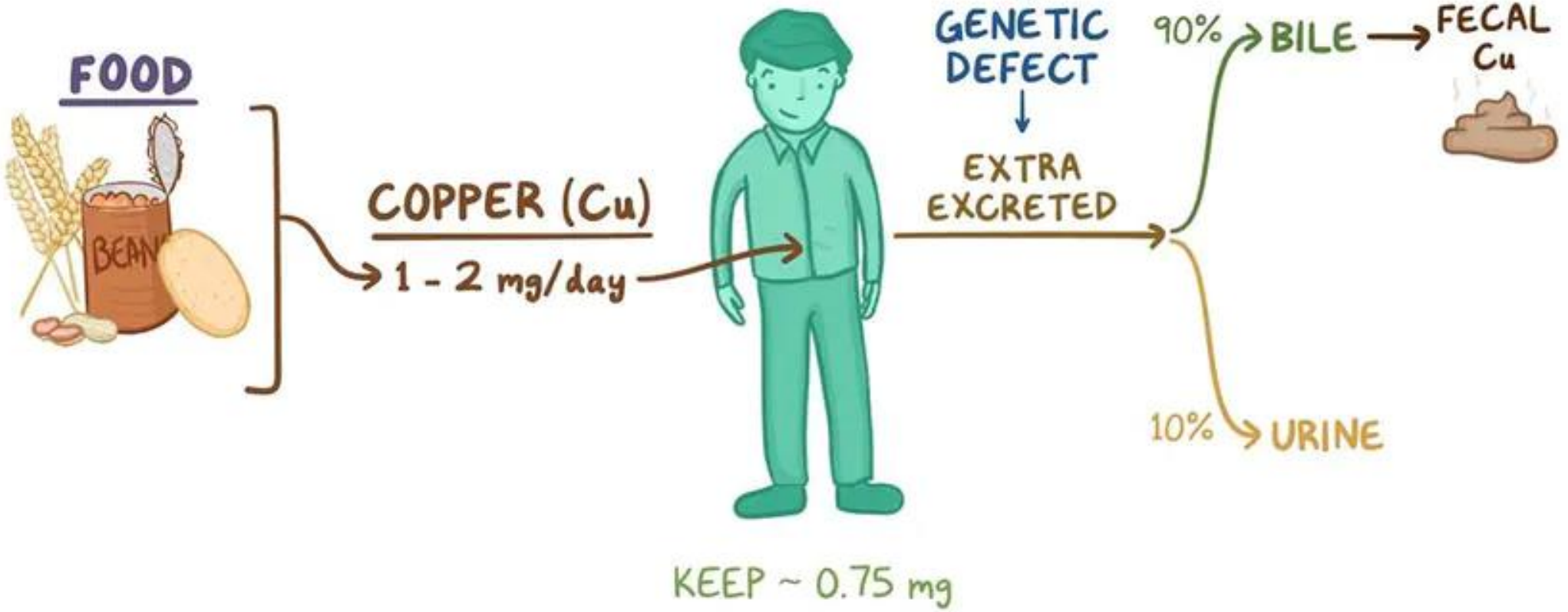


- Wilson disease is a rare genetic disorder characterized by excess copper stored in various body tissues, particularly the liver, brain, and corneas of the eyes.
- The disease is progressive and, if left untreated, it may cause liver (hepatic) disease, central nervous system dysfunction, and death.

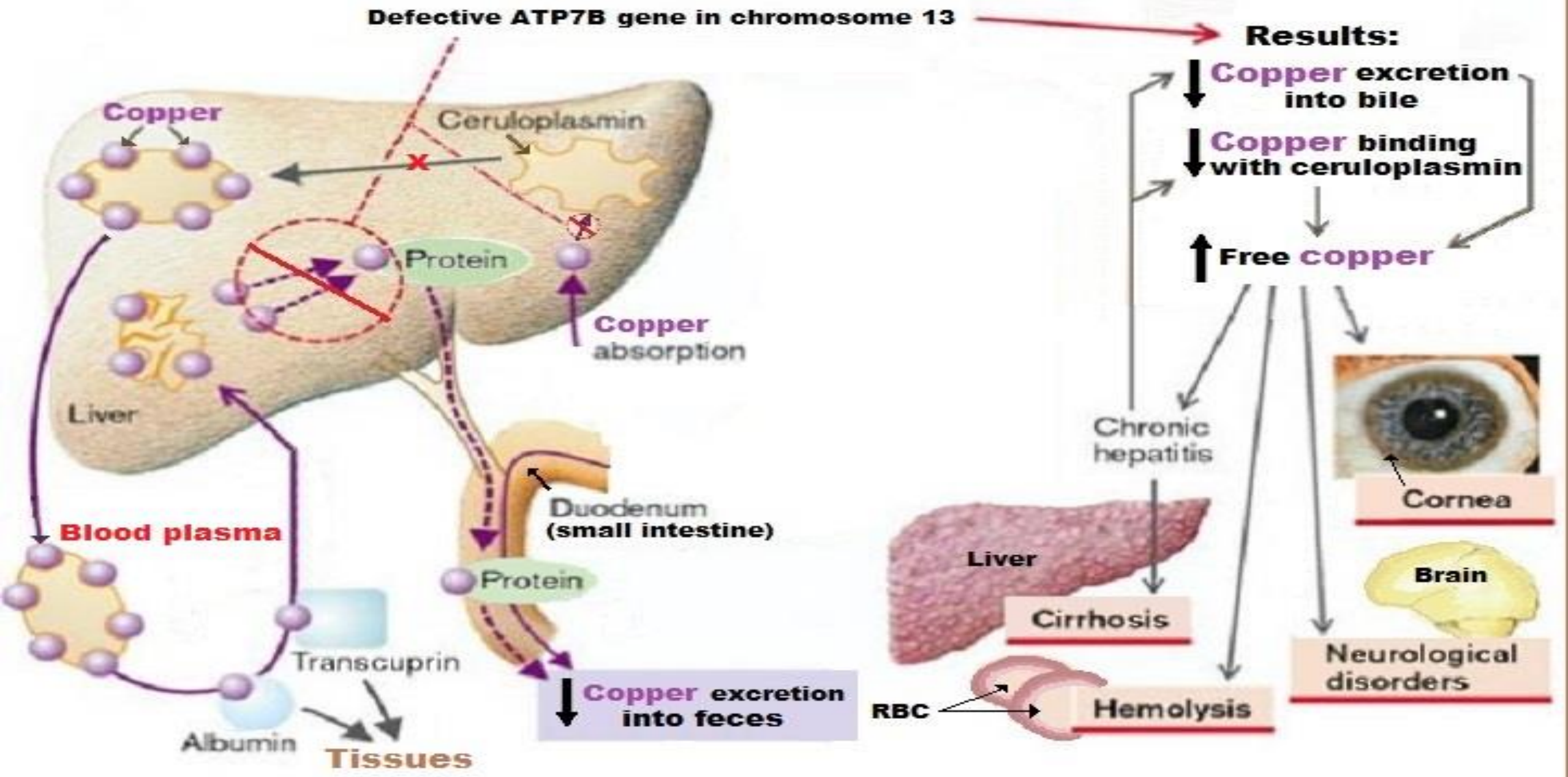
- Wilson’s disease is detected in its early stages in the patient, in people between the **ages of 5 and 35**.
- Wilson disease is a rare disorder that **affects males and females in equal numbers**. The disease is found in all races and ethnic groups.
- Wilson’s disease occurs in approximately **one** in 30,000 to 40,000 people worldwide. Approximately **one** in 90 people may be carriers of the disease gene.
- only about 2,000-3,000 cases have been diagnosed in the United States, other affected individuals may be misdiagnosed with other neurological, liver disorders.

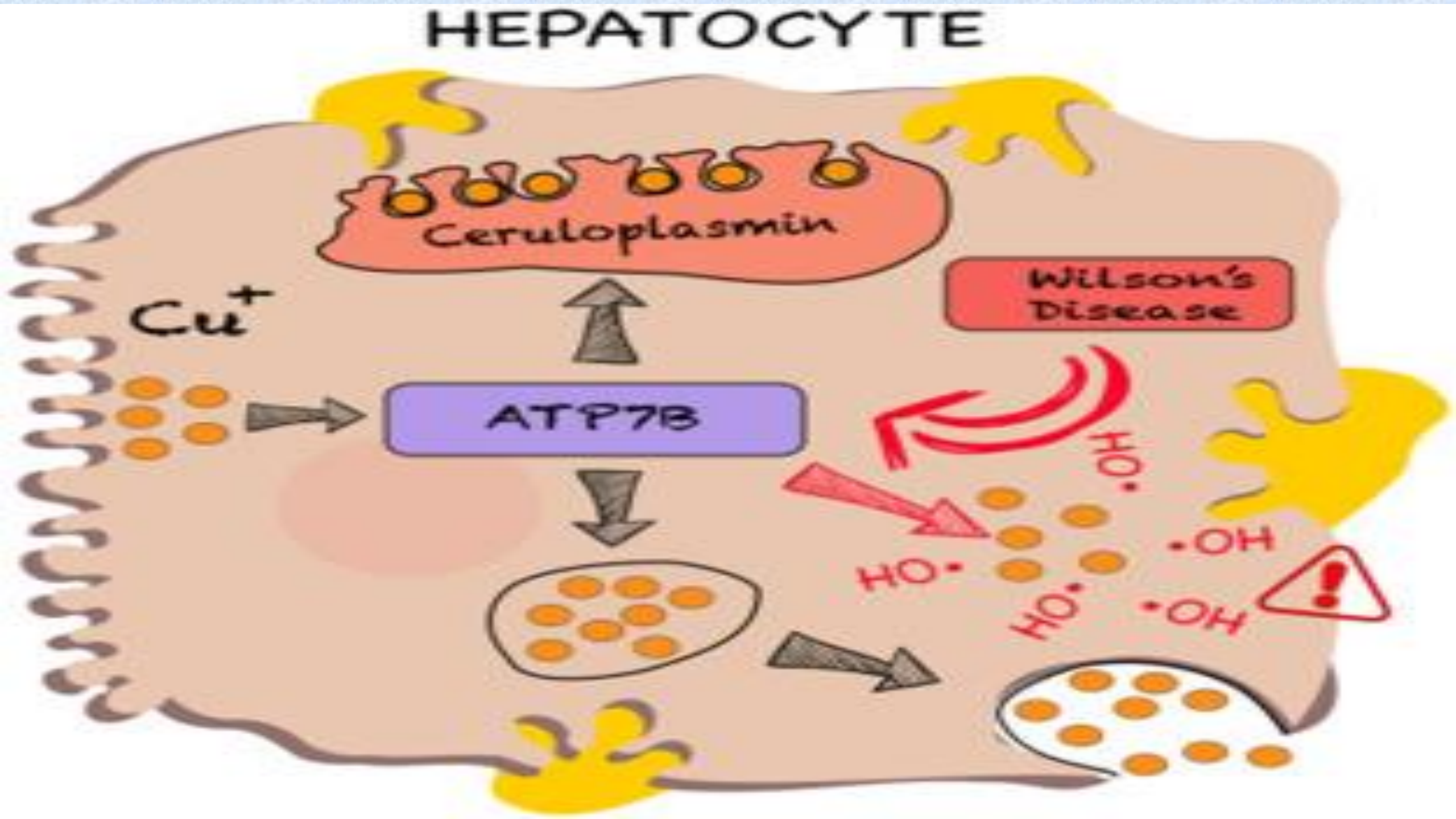
- Wilson disease is **caused by** mutations of the **ATP7B** gene on chromosome **13** , which plays an important role in the movement of excess copper from the liver to the bile to be excreted from the body through the intestines.
- More than **300** different mutations of the ATP7B gene have been identified.

WILSON DISEASE



Wilson's Disease



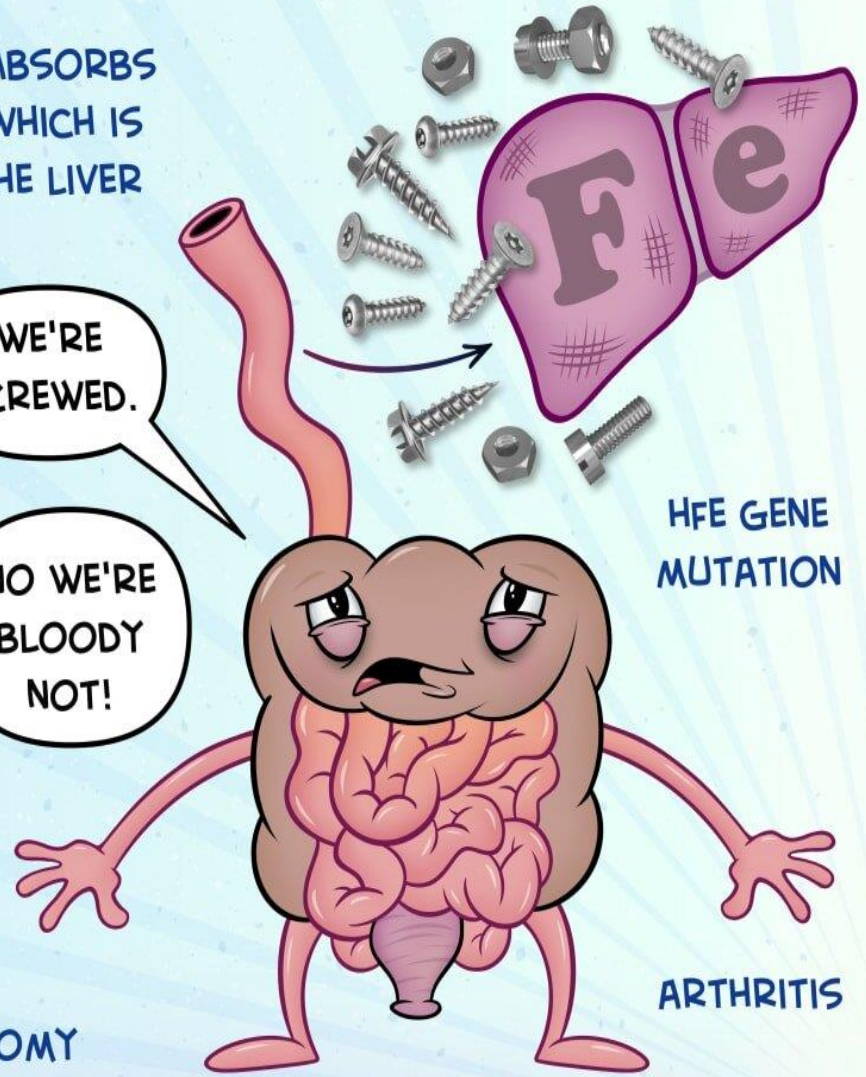

HEPATOCYTE
Ceruloplasmin
Wilson's Disease
Cu+
ATP7B
HO·
HO·
·OH
HO·
·OH

HEREDITARY HEMOCHROMATOSIS

WILSON DISEASE

BOTH ARE INHERITED AUTOSOMAL RECESSIVE DISORDERS THAT CAN CAUSE CIRRHOSIS AND ORGAN TOXICITY DUE TO EXCESS METAL

THE INTESTINE ABSORBS EXCESS IRON WHICH IS RECEIVED BY THE LIVER



BRONZE DIABETES

HFE GENE MUTATION

ARTHRITIS

TX: PHLEBOTOMY

HEMOLYTIC ANEMIA

Cu

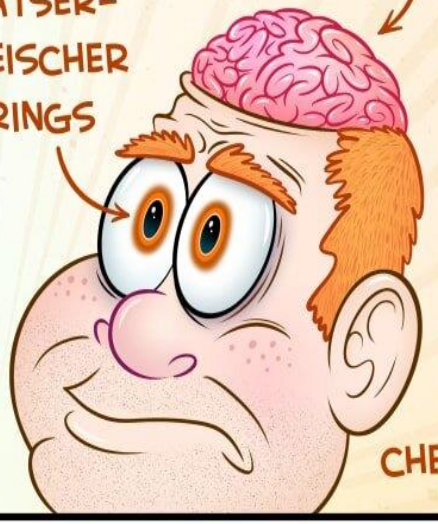
ATP7B GENE MUTATION

LIVER CELLS' ABILITY TO EXCRETE COPPER INTO BILE IS IMPAIRED

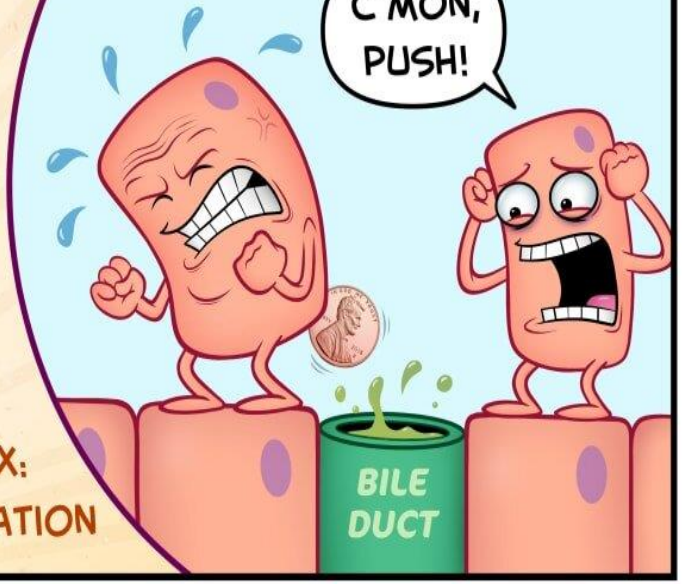
RENAL DISEASE

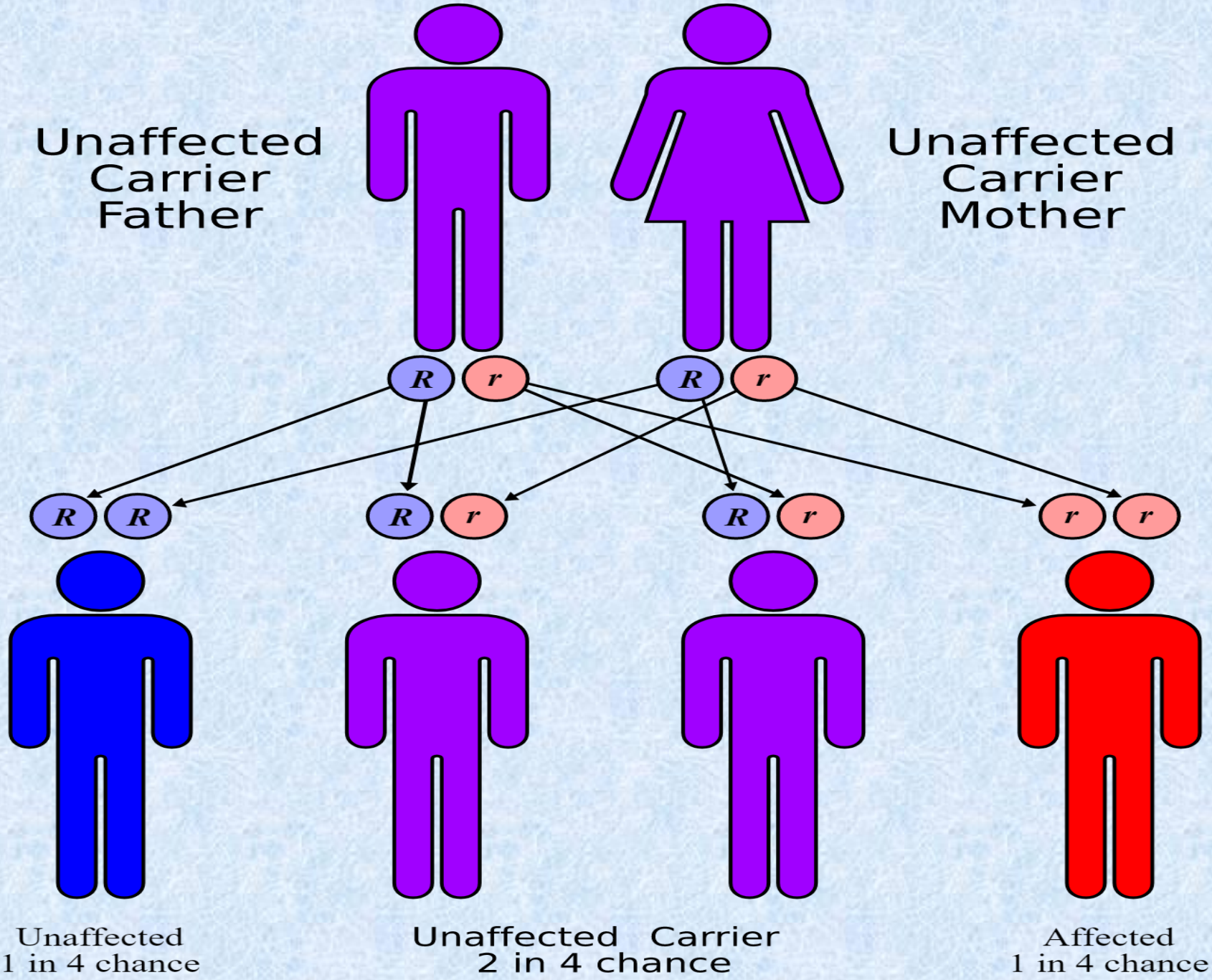
CNS DISEASE

KAYSER-FLEISCHER RINGS



TX: CHELATION




Unaffected
Carrier
Father
Unaffected
Carrier
Mother
R
r
R
r
R
R
R
r
R
r
r
r
Unaffected
1 in 4 chance
Unaffected Carrier
2 in 4 chance
Affected
1 in 4 chance

- **Common signs** of associated liver disease include a yellow discoloration (**jaundice**) of the skin, mucous membranes and the membranes (sclera) that line the eye.
- swelling (**edema)** of the legs and abdomen **(ascites**) due to abnormal retention of fluid and fatigue.
- symptoms of liver disease often associated with anemia due to breakdown of red blood cells **(hemolysis**) and mental confusion.
- In young patients, the characteristic rusty-brown deposits in the corneas of the eyes (**Kayser-Fleischer rings**).

Signs of Wilson's Disease

20,000 cases per year

Inhertited disorder

causes too much copper

to accumulate in organs


Thank You