

Wilson's disease (biochemical features)

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Wilson's disease disorder of metal transport and metabolism (copper).

The biochemical features of Wilson's disease are:

1. A reduced plasma caeruloplasmin concentration.
2. low- normal or low plasma copper (with increased binding to albumin).

- ▶ Caeruloplasmin It is a copper containing α_2 -globulin, a glycoprotein with enzyme activities. Molecular weight is $\approx 151,000$.
- ▶ It has eight sites for binding copper- contains about eight atoms of copper per molecule- $\frac{1}{2}$ as cuprous (Cu^+) and $\frac{1}{2}$ as cupric (Cu^{++}).

- ▶ It carries **0.35** per cent Cu by weight.
- ▶ Normal plasma contains approx. **30 mg/100** ml and about **75** to **100** µg of Cu may be present in 100 ml of plasma.
- ▶ It has enzyme activities, e.g. **copper oxidase, histaminase** and **ferrous oxidase**.

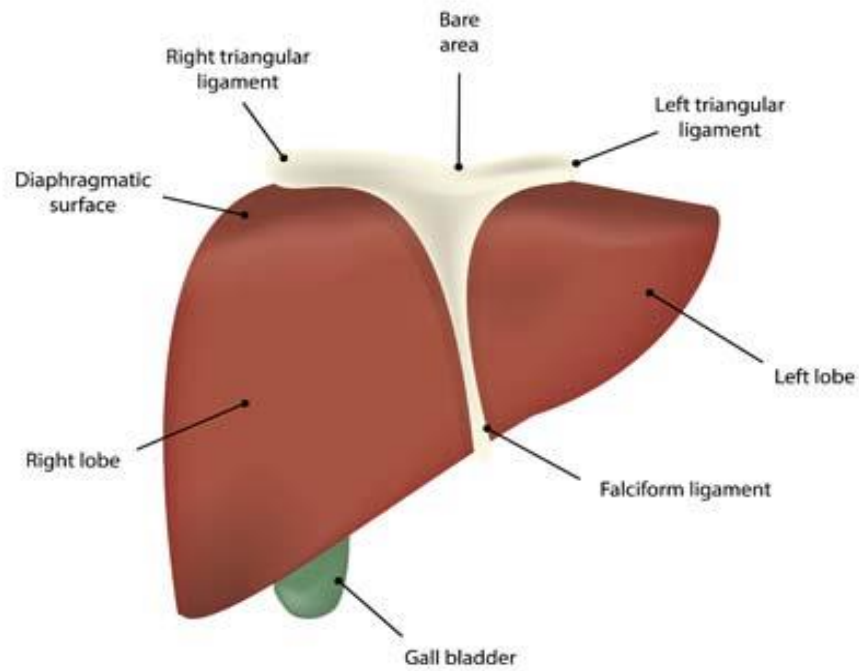
- ▶ **Site of Synthesis:** It is synthesised in liver, where **eight copper atoms** are attached to a protein, apocaeuroplasmin.
- ▶ **Level of caeruloplasmin with age and sex:** There is low concentrations at birth, gradually increases to adult levels, and slowly continues to rise with age thereafter.
- ▶ **Adult females have higher concentrations than males.**

► Functions of Caeruloplasmin

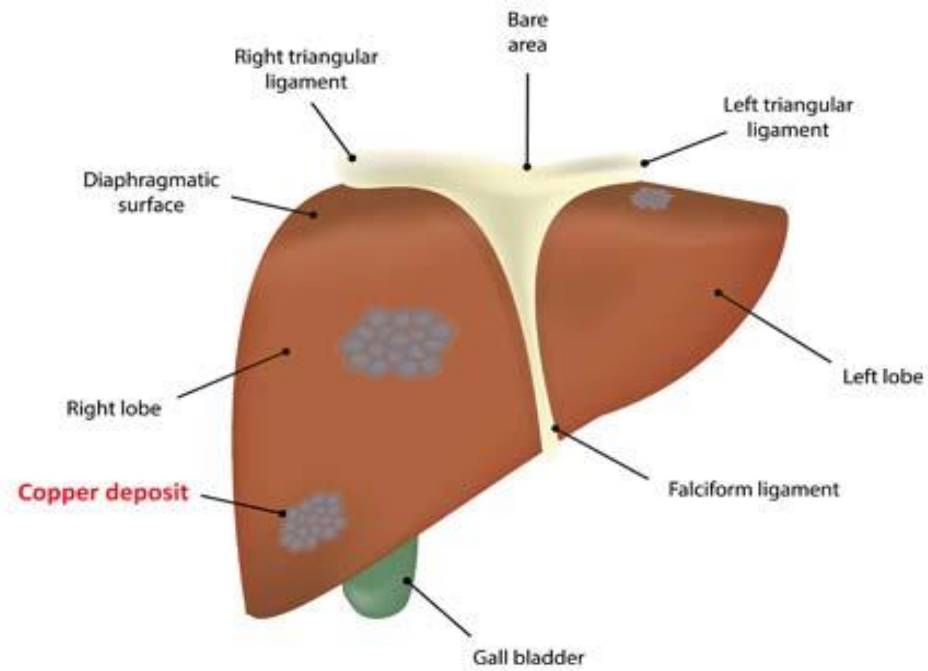
- Although caeruloplasmin is not involved in copper transport, 90 per cent or more of total serum copper is contained in caeruloplasmin.
- It mainly functions as a ferroxidase and helps in oxidation (conversion) of Fe^{++} to Fe^{+++} which can be incorporated into transferrin.

Wilson's Disease

Healthy Liver



Wilson's Disease

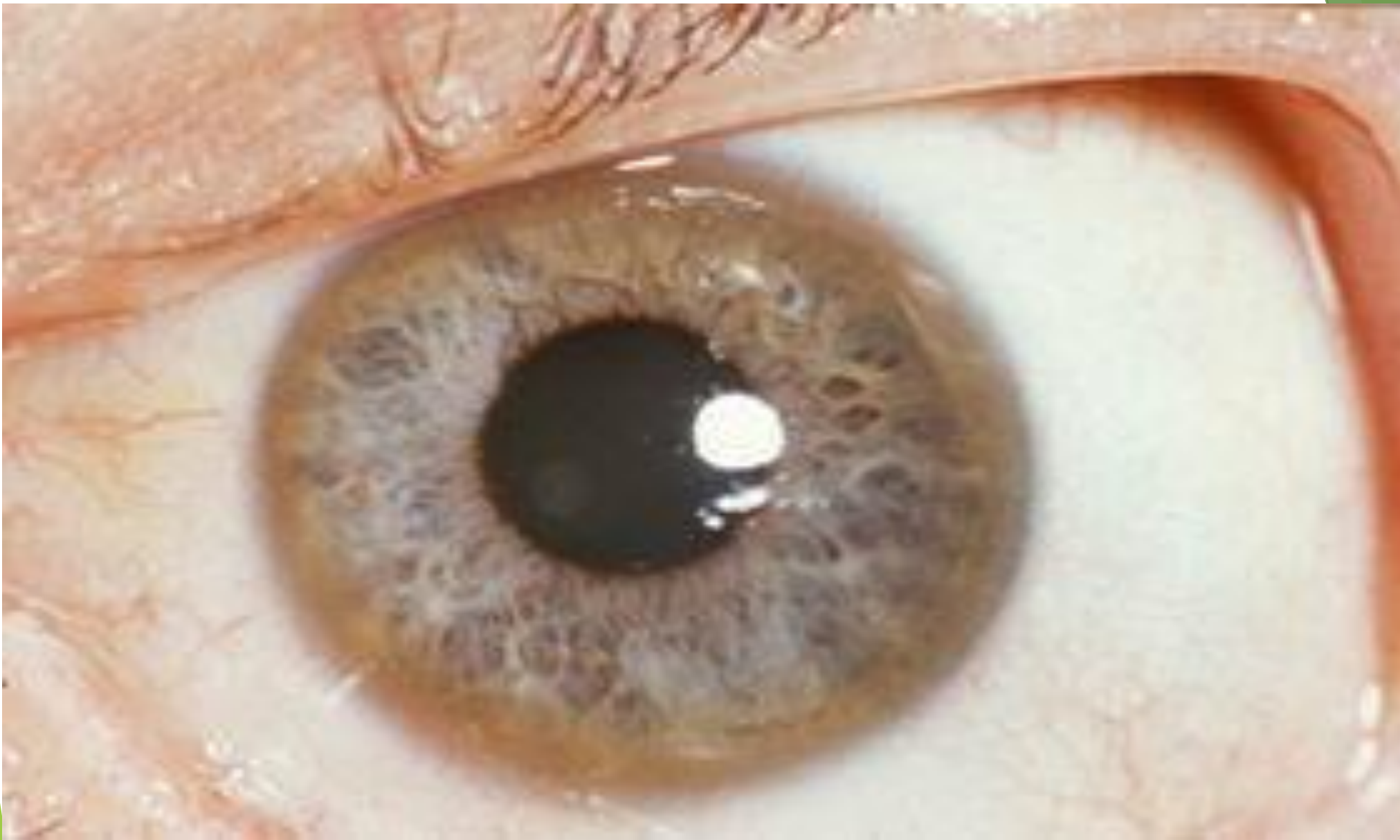


The decrease in caeruloplasmin is not unique to Wilson's disease, but also seen in

- ▶ chronic hepatitis
- ▶ malnutrition.

- ▶ Copper is an essential trace element that plays a fundamental role in biochemistry, permitting facile electron transfer reactions in diverse metabolic pathways.
- ▶ Despite this essential role, copper is highly reactive and potentially toxic, thus specialized pathways have evolved for the trafficking of this metal within cells.

- ▶ The most significant sign in the diagnosis of Wilson disease results from the deposition of copper in Descemet's membrane of the cornea.
- ▶ These golden-brown deposits can be seen with a slit-lamp and are Kayser-Fleischer rings (see image below).



Kayser-Fleischer rings

Thank you

The background features abstract, overlapping geometric shapes in various shades of green, ranging from light lime to dark forest green. These shapes are primarily located on the right side of the frame, creating a modern, layered effect against the white background.