WILSON DISEASE

AN OVERVIEW

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Introduction

Wilson disease (WD) is an inherited disorder of copper metabolism, transmitted as an autosomal recessive trait. This type of inheritance means unaffected parents who each carry the WD gene have a 25% risk in each pregnancy of having an affected child.

The disorder is caused by a defective copper-binding protein found primarily in the liver, which leads to excess copper circulating through the bloodstream.

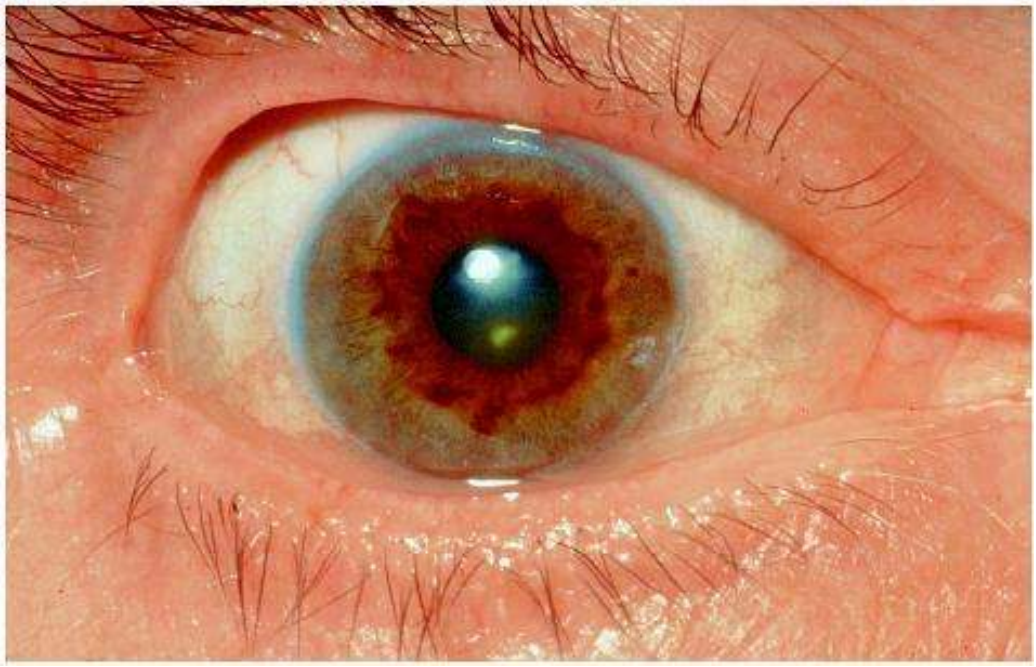
Over time, the copper is deposited and increased to toxic levels in various body tissues, especially the liver, brain, kidney, and cornea of the eye. Left untreated, WD is invariably fatal.

The classic triad of signs for WD includes lenticular degeneration, cirrhosis of the liver, and neuropsychiatric symptoms. Errors in a specific gene produce a defective copper-binding protein in the liver, which results in an inability to excrete excess copper. While some copper is necessary for normal metabolic processes in the body, too much can be toxic.

The disease is present at birth, but symptoms typically do not show until years later. WD is progressive because the underlying cause cannot be corrected. Effective treatments are available, but without treatment, people with WD will eventually die of liver failure.

An easily detectable physical sign is the presence of Kayser-Fleisher rings in the eye, which are bluish rings around the iris, caused by copper deposition in the cornea.

(Picture Below - blue Kayser-Fleisher rings in the eye)



Symptoms

Neurological symptoms are primarily the result of copper's toxic effects in the basal ganglia, a portion of the brain that controls some of the subconscious aspects of voluntary movement such as accessory movements and inhibiting tremor. These symptoms include:

- Dystonia - Prolonged muscular contractions that may cause twisting (torsion) of body parts, repetitive movements, and increased muscular tone
- Dysarthria - Difficulty in articulating words, sometimes accompanied by drooling
- Dysphagia - Difficulty swallowing
- Pseudosclerosis - Symptoms similar to multiple sclerosis

Treatment

Treatment of WD revolves around the process of copper chelation. A chelating agent binds to excess copper in the bloodstream so that it can be excreted from the body. Penicillamine is the most effective and commonly used medication, but about 20% of all patients suffer serious side effects, which may include joint pain, blood disorders, fever, an increase in neurologic symptoms, and systemic lupus erythematosus.

Trientine and zinc salts given orally are somewhat less effective, but have fewer side effects than penicillamine.

In addition, zinc salts may take several months to have any noticeable effect. A diet low in copper will also have some preventive effect. Finally, for those patients in advanced stages of liver disease, liver transplantation may be the only method of averting liver failure and death.

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