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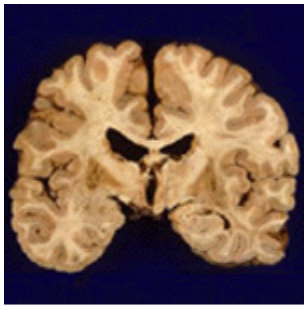
Wilson's disease



In Wilson's disease, toxic levels of copper accumulate and damage many tissues and organs, including the basal ganglia of the brain. [Image credit: Kevin Roth and Robert Schmidt, Washington University, St. Louis, MO, USA.]

Wilson's Disease is a rare autosomal recessive disorder of copper transport, resulting in copper accumulation and toxicity to the liver and brain. Liver disease is the most common symptom in children; neurological disease is most common in young adults. The cornea of the eye can also be affected: the 'Kayser-Fleischer ring' is a deep copper-colored ring at the periphery of the cornea, and is thought to represent copper deposits.

The gene for Wilson's disease (ATP7B) was mapped to chromosome 13. The sequence of the gene was found to be similar to sections of the gene defective in Menkes disease, another disease caused by defects in copper transport. The similar sequences code for copper-binding regions, which are part of a transmembrane pump called a P-type ATPase that is very similar to the Menkes disease protein.

A homolog to the human ATP7B gene has been mapped to mouse chromosome 8, and an authentic model of the human disease in rat is also available (called the Long-Evans Cinnamon [LEC][rat). These systems will be useful for studying copper transport and liver pathophysiology, and should help in the development of a therapy for Wilson disease.

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