Diagnostic tests for Wilson's disease

Download booklet

The diagnosis of Wilson's disease is made by relatively simple tests. These tests can diagnose the disease in both symptomatic patients and people who show no signs of the disease "pre-symptomatic"

The copper accumulation in the eye in Wilson's disease may cause a diagnostic golden-brown ring to form around the edge of the iris, called a Kayser-Fleischer ring. This ring is only visible using a special instrument (slit-lamp) and is rarely present before the age of 10 years.

Listed below are the standard laboratory tests used to diagnose Wilson's disease:

- Urine copper is high; this should be measured in a 24 hour urine collection.
- "Caeruloplasmin", a copper-containing protein in blood plasma is usually low.
- The copper concentration measured in a liver biopsy specimen will be high.
- The cerebral imaging (MRI) may be abnormal.
- In cases which are difficult to diagnose, copper isotope studies (more complex copper tests) may be performed.

Parameter	Normal	Wilson's
Plasma caeruloplasmin	>200 mg/l	<200 mg/l
Urine Cu	<0.6 µmol/24 h	>1.6 µmol/24 h
Hepatic copper	< 250 µg/g dry wt	> 250 µg/g dry wt
Kayser-Fleischer rings	Absent	Present in neurological cases, but may be absent in younger children
Cerebral imaging (MRI)	Normal	May be abnormal