Common symptoms of Wilson's disease

- **Liver disease or liver function test abnormalities**
- **Haemolytic anaemia**
- **Tremors or chorea/Parkinsonism symptoms**
- **Gait disturbances**
- **Balance disorders**
- **Stiffness or rigidity (dystonia)**
- **Abnormal reflexes**
- **Abnormal speech (slurring)**
- **Drooling**
- **Difficulty swallowing – frequent choking**
- **Psychiatric disturbances**

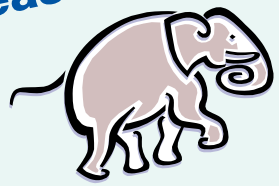
Any patient with an unexplained abnormal liver function test or presenting with one or more of the above symptoms should prompt the physician to consider the diagnosis of **Wilson's disease.**

Unless Wilson's disease is correctly diagnosed and treated in good time, **DISABILITY**, **SUFFERING** and **PREMATURE DEATH** are inevitable.

Aims of the Wilson's Disease Support Group – UK (WDSG-UK)

- To provide support for patients, their families and friends
- To provide a communications network for affected individuals, their families and friends
- To produce and distribute an interesting and informative annual newsletter for members and for Neurology and Hepatology clinics around the UK
- To provide and maintain an up-to-date website with information about the disease and details of how to join the Group
- To hold an informal annual meeting for medical professionals, new and existing patients, and their families and friends
- To raise awareness of Wilson's disease, especially amongst medical professionals
- To collect up-to-date information on research, treatment and management of the disease
- To raise funds for the Group's activities and to support research into Wilson's disease

www.wilsonsdisease.org.uk



WILSON'S DISEASE

WILSON'S DISEASE SUPPORT GROUP – UK

Affiliated to the British Liver Trust

www.wilsonsdisease.org.uk

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This pamphlet has been prepared by WDSG-UK and contains important information on **WILSON'S DISEASE**, a **RARE GENETIC DISORDER** which is **FATAL** if not correctly diagnosed and treated in time

What is Wilson's disease?

Wilson's disease (WD) is a rare autosomal recessive inherited disorder of copper metabolism, which is characterised by hepatic, neurological, and psychiatric symptoms. Worldwide, the disease affects between one in 30,000 and one in 100,000 individuals. **It is fatal unless diagnosed and treated in time**.

Copper is present in most foods and is an essential element for humans. In WD patients, the biliary route for excretion of dietary copper is impaired and copper accumulates in the tissues: in the liver leading to progressive hepatic damage; in the brain leading to disturbances of the personality and destruction of motor function; in the cornea producing Kayser-Fleischer rings; and in the kidney giving rise to functional disturbances.

When does Wilson's disease present?

Wilson's disease usually presents between the ages of 6 and 40, **but should not be ruled out at any age**.

Although the accumulation of copper begins at birth, progression is insidious and early symptoms may appear quite trivial e.g. deterioration in handwriting.

What symptoms and signs should make the GP consider the diagnosis of Wilson's disease?

Hepatic abnormalities

Patients may present with jaundice, abdominal swelling, abdominal pain, vomiting blood, haemolytic anaemia and abnormal liver functions.

Wilson's disease hepatitis is often mistaken for infectious hepatitis or infectious mononucleosis.

Neurological abnormalities

Neurological abnormalities are normally always motor and not sensory disturbances. Kayser-Fleischer rings are invariably present when there are neurological signs. While these are sometimes visible to the naked eye, they are best found by a slit lamp examination.

Motor abnormalities

Patients may have progressively severe motor tremors, gait disturbances, balance disorders, incapacitating rigidity of the head, arms, hands, legs and/or tongue (often with profuse drooling and slurred speech).

Psychiatric disturbances

Symptoms may include an abrupt personality change, an inexplicable deterioration in school work, depression, aggression and/or psychosis.

How is Wilson's disease diagnosed?

Once suspected, the diagnosis of WD can usually be confirmed or ruled out by blood tests for serum caeruloplasmin, a 24 hour urinary copper excretion measurement and an ophthalmologist's examination of the eyes for Kayser-Fleischer rings. In some cases further tests may be needed.

The earlier the diagnosis the better the chances of recovery.

How is Wilson's disease treated?

WD is a very treatable condition. With correct diagnosis and immediate therapy, disease progress can be halted and often symptoms can be improved and reversed.

Treatment is aimed at removing excess accumulated copper and preventing its re-accumulation. Therapy for WD is therefore lifelong and the available drugs include:

Trientine – a chelating agent, induces cupruria

D-penicillamine – also a copper chelating agent which induces cupruria

Zinc acetate – blocks intestinal absorption of copper

Tetrathiomolybdate (TTM) – blocks copper absorption; if absorbed, forms a TTM-copper-albumin complex