The Organisation

- founded in 1990
- active throughout Germany
- maintaining contacts with partner organisations operating at national level
- international networking
- a non-profit healthcare organisation

Purpose of the Organisation

- Information, advice and support
- Networking amongst patients, relatives, doctors and interested parties
- Training events
- Public awareness campaigns
- Support for research

The organisation is supported by its Scientific Advisory Board on medical matters.

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MORBUS WILSON e.V.

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If you would like to support the work of Morbus Wilson e. V., we would be delighted to welcome you as amember or to receive a donation. Donations may be tax-deductible in Germany.

Our Bank Details

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Wilson’s Disease

Copper Storage Disorder

What is Wilson’s disease?

Which treatment options are available?

What are our objectives?

Wilson's Disease

is a rare, autosomal recessive disease affecting children, adolescents or adults, caused by copper overload in the body.

Cause

Due to a genetic defect, the excretion of copper via the bile ducts is impaired. The excess copper is initially stored in the liver. In the early stages of the disease, this leads to liver damage and elevated liver enzyme levels. When the liver can no longer store any more copper, it also enters the brain and other organs (e.g. the kidneys and the cornea of the eye). Thus, over the years, copper accumulates throughout the entire body.

Autosomal recessive inheritance means that both parents are carriers of the same defective gene, without themselves developing the condition. If the child inherits the defective gene from both parents, they will develop Wilson's disease.

According to current knowledge, the prevalence of the disease worldwide is 1 in 30,000. The Wilson's disease gene is located on chromosome 13 and exhibits many different mutations.

Symptomatology

The first symptoms usually appear between the ages of 5 and 45. In children, liver damage is usually the main symptom; this can lead to hepatitis or cirrhosis of the liver.

In adolescents and young adults, neurological deficits also begin to appear. These include, amongst other symptoms, speech and swallowing difficulties, tremors in the limbs, and problems with gait and handwriting. Psychological changes occur in isolated cases, too.

Copper deposits in the eye may manifest as a greenish-brown ring around the cornea, known as the Kayser-Fleischer corneal ring. This is more of a late-stage symptom than an early one and is therefore not always detectable in the early stages of the disease.

Diagnostics

- If Wilson's disease is suspected, it is advisable to consult a doctor with experience in Wilson's disease.
- Laboratory tests: liver function tests, complete blood count, coeruloplasmin and copper levels in the blood, copper excretion in a 24-hour urine collection
- Slit lamp examination in a specialised ophthalmological practice
- Ultrasound scan of the liver and the spleen
- If necessary, a liver biopsy to measure hepatic copper levels and/or a D-penicillamine loading test
- Molecular genetic testing
- Only a combination of several tests can confirm the diagnosis

Family Screening

- If a family member is diagnosed with Wilson's disease, it is strongly recommended that all first-degree relatives undergo a family screening.
- Genetic testing is essential for siblings.

Treatment

With early diagnosis and lifelong, consistent treatment, the prognosis is favourable. The aim of initial treatment is to deplete the body's copper stores. Maintenance therapy serves to maintain a balanced copper metabolism.

So far, the first-line treatments remain the chelating agents D-penicillamine and trientine.

Oral zinc therapy may be used as part of a maintenance therapy.

Avoiding foods high in copper is advisable as a complementary measure, particularly in the early stages of treatment.

If drug therapy fails, a liver transplant is the life-saving alternative.

Several companies are currently conducting clinical trials on gene therapies they have developed.

Treatment must not be interrupted during pregnancy. Morbus Wilson e.V. provides a separate leaflet for expectant parents.