

## About Wilson Disease

Wilson Disease is a genetic disorder that is fatal unless detected and treated before serious illness from copper poisoning develops. Wilson disease affects approximately 1 in 30,000 people worldwide. The genetic defect causes excessive copper accumulation in the liver and/or brain.

Small amounts of copper are as essential as vitamins. Copper is present in most foods, and most people have much more copper than they need. Healthy people excrete copper they don't need but Wilson disease patients cannot.

Copper begins to accumulate immediately after birth. Excess copper attacks the liver and/or brain, resulting hepatitis, psychiatric, or neurological symptoms. The symptoms usually appear in late adolescence, but can manifest at any age. Patients may have jaundice, abdominal swelling, vomiting of blood, and abdominal pain. They may have tremors and difficulty walking, talking and swallowing. They may develop all degrees of mental illness including homicidal or suicidal behavior, depression, and aggression. Women may have menstrual irregularities, absent periods, infertility, or multiple miscarriages. No matter how the disease begins, **it is always fatal if it is not properly diagnosed and treated.**

The first part of the body that copper affects is the liver. In about half of Wilson disease patients the liver is the only affected organ. The initial physical changes in the liver are only visible under the microscope. When hepatitis develops, patients are often thought to have infectious hepatitis or infectious mononucleosis when they actually have Wilson disease hepatitis. Testing for Wilson disease should be performed in individuals with unexplained, abnormal liver tests.

## How is Wilson Disease Diagnosed?

The diagnosis of Wilson disease is made by relatively simple tests. The tests can diagnose the disease in both symptomatic patients and people who show no signs of the disease. These tests can include:

- Ophthalmologic slit lamp examination for Kayser-Fleischer rings (copper rings around the eyes)
- Serum ceruloplasmin and serum copper blood tests
- 24-hour urine copper test
- Liver biopsy for histology and histochemistry and copper quantification
- Genetic testing, haplotype analysis for siblings, and mutation analysis.

It is important to diagnose Wilson disease as early as possible, since severe liver damage can occur before there are any signs of the disease making a liver transplant necessary.

## Wilson Disease is a Multisystem disorder.

Systems that can be affected are:

### Hepatic

- Asymptomatic hepatomegaly
- Isolated splenomegaly
- Persistently elevated serum aminotransferase activity (AST, ALT)
- Fatty liver
- Acute hepatitis
- Resembling autoimmune hepatitis
- Cirrhosis: compensated or decompensated
- Acute liver failure

### Neurological

- Movement disorders (tremor, involuntary movements)
- Drooling, dysarthria
- Rigid dystonia
- Pseudobulbar palsy
- Dysautonomia
- Migraine headaches
- Insomnia
- Seizures

### Psychiatric

- Depression
- Neurotic behaviours
- Personality changes
- Psychosis

### Other systems

- Ocular: Kayser-Fleischer rings, sunflower cataracts
- Cutaneous: lunulae ceruleae
- Renal abnormalities: aminoaciduria and nephrolithiasis
- Skeletal abnormalities: premature osteoporosis and arthritis
- Cardiomyopathy, dysrhythmias
- Pancreatitis
- Hypoparathyroidism
- Menstrual irregularities; infertility, repeated miscarriages

## **About Our Association**

The Wilson Disease Association (WDA) is a 501(c)(3), all-volunteer organization striving to promote the well being of patients with Wilson disease and their families and friends. We rely on donations to achieve our Mission.

## **Our Mission**

The Wilson Disease Association funds research and facilitates and promotes the identification, education, treatment, and support of patients and other individuals affected by Wilson disease.

## **Our Vision**

It is our belief that nobody need suffer from Wilson disease. Our vision is to be recognized for excellence as an international, non-profit health organization. We will:

- Conduct our business in a professional manner.
- Raise funds and responsibly manage our resources in order to provide and sustain high-quality program services.
- Support research and clinical investigation that are in the best interest of patients and families.
- Develop educational programs for healthcare professionals and patients to assure proper early diagnosis, treatment, and rehabilitation. Patient care will be accessible, affordable, and of high quality.
- Foster a comfortable, supportive environment for Wilson disease patients and families.

We are committed to this vision for the benefit of all affected by Wilson disease.

## **WDA's Patient Support Efforts**

- Maintain a membership list that includes patients, their families, their friends, medical personnel, and other individuals and organizations interested in the disease
- Send a regular newsletter containing information about the disease, issues of interest to the membership, and other related matters
- Provide a network for written, phone, and electronic communication between affected individuals and their families
- Provide referrals to physicians, other health care providers, and other assistive resources; facilitate consultation with Wilson disease expert physicians

## **WDA's Education and Outreach Efforts**

- Answer questions from the public about the disease
- Participate in political activities to promote the interests of Wilson disease patients
- Provide money to needy patients for travel, equipment and medication when necessary
- Attend professional meetings, distribute educational materials, and promote media coverage to educate the public and professionals about Wilson disease

## **Clinical and Research Efforts**

- Raise funds to support the goals of the WDA's research program
- Cooperate with investigators and clinicians to advance medical knowledge on the disease, and develop more accurate diagnostic methods, life-improving therapies, and ultimately a cure
- Accept grant proposals on an annual basis for innovative basic, clinical, or translational research relevant to the cause or treatment of Wilson disease
- Award annual grants to researchers based on the scientific merit of the proposed study and the research priorities established by the Wilson Disease Association



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*Looking Toward the Future*