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MADISON NEUROLOGY LLC

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Neurology Care 7 days a Week. Because Your Brain Matters.

Wilson's Disease

Patient education guide · Movement Disorders · Madison Neurology LLC

What is it?

Wilson's disease is an inherited disorder of copper handling in which copper accumulates to toxic levels in the liver, brain, and other organs. Neurological Wilson's causes tremor, stiffness, slurred speech, swallowing difficulty, and psychiatric changes — typically beginning between the teens and 40s. It is one of the most important diagnoses in neurology because it is completely treatable, and fatal if missed.

How does it happen?

Mutations in the ATP7B gene (autosomal recessive — both parents carry a copy) impair copper excretion into bile. Copper builds up first in the liver, then spills into the bloodstream and deposits in the brain (especially the basal ganglia), corneas (Kayser-Fleischer rings), kidneys, and joints.

Common symptoms

- Tremor (classically a coarse 'wing-beating' tremor), stiffness, and slowness
- Slurred speech, drooling, and swallowing difficulty
- Psychiatric symptoms: personality change, depression, impulsivity — often preceding neurological signs
- Signs of liver disease (may be silent)
- Kayser-Fleischer rings: copper-colored rings at the edge of the cornea, seen on eye exam

How is it diagnosed?

Blood tests show low ceruloplasmin and abnormal copper studies; 24-hour urine copper is elevated; a slit-lamp eye exam looks for Kayser-Fleischer rings. Liver ultrasound/biopsy and genetic testing complete the picture. Every patient under 40-50 with unexplained movement or liver disease should be screened.

How is it treated?

Lifelong copper-removing or copper-blocking medication: chelators (trientine, penicillamine) remove copper; zinc blocks its absorption. A low-copper diet (limiting shellfish, liver, nuts, chocolate initially) helps. Treatment must never be stopped — interruption can cause fatal liver failure. Neurological symptoms often improve over months of treatment, though they may briefly worsen when starting chelators. Siblings must be screened, since catching the disease before symptoms is fully preventive.

What does the future hold?

With consistent lifelong treatment, most patients live normal lives with normal life expectancy; neurological symptoms improve substantially, though severe deficits may only partly reverse. Untreated Wilson's is fatal — the contrast makes adherence the whole game. Family screening turns a dangerous disease into a prevented one.

Questions patients commonly ask

Q: Is it really curable?

A: Effectively yes — treatment removes and blocks copper, preventing further damage and allowing substantial recovery, as long as it is never stopped.

Q: Should my family be tested?

A: Yes — siblings have a 25% chance of having it, and treatment before symptoms begin prevents the disease entirely.

Q: Why did my tremor get worse right after starting treatment?

A: Copper mobilization can transiently worsen neurological symptoms in the first weeks; doctors manage this by adjusting agents and dose.

National support groups & resources

- **Wilson Disease Association** — wilsonsdisease.org
- **National Organization for Rare Disorders** — rarediseases.org — 1-800-999-6673
- **American Academy of Neurology — Brain & Life patient resources** — brainandlife.org

(Organization contact details were current at the time of compilation; please verify online.)

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