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WILSON'S DISEASE

Overview

Wilson's disease is a hereditary disorder that causes too much copper to accumulate in your liver, brain and other vital organs. This potentially fatal disease shows up in a variety of different ways, but can remain silent for years.

Bile is the brown liquid produced by your liver to aid in digestion. Bile normally carries away excess copper from your liver. The liver of someone with Wilson's disease doesn't release copper into the bile as it should. The resulting buildup of copper in the liver causes injury to the tissue. Eventually, some of the excess copper travels throughout the body, where it may damage your brain, eyes, kidneys and red blood cells.

Left untreated, the liver damage can lead to cirrhosis, an irreversible scarring of the liver that seriously disrupts its normal functioning. Treatment involves taking medications and avoiding certain high-copper foods.

Wilson's disease is rare, affecting about one in 30,000 people in the United States.

Signs and symptoms

Signs and symptoms of Wilson's disease usually appear between the ages of 6 and 20, but they can appear as late as age 50. The symptoms of Wilson's disease fall into three main categories:

Liver problems. Wilson's disease can mimic any form of acute or chronic liver disease. Possible signs and symptoms include abdominal pain and yellowing of the skin (jaundice). Sometimes, the liver damage progresses without any obvious symptoms until cirrhosis develops. In the advanced stages of disease, more serious signs and symptoms, such as anemia or the vomiting of blood, can occur.

Neurological problems. The most common neurological signs and symptoms in people with Wilson's disease are tremors and muscle spasticity. Speech problems may also occur.

Behavioral or psychological problems. Wilson's disease can cause abrupt personality changes and inappropriate behavior. Sometimes, signs and symptoms in school-age children, such as depression, erratic behavior or failing school performance, are mistaken for behavioral problems.

Some people with Wilson's disease are diagnosed incidentally during an eye exam after a doctor notices a brown, ring-shaped pigmentation, called a Kayser-Fleischer ring, in the cornea.

Causes

An abnormal gene inherited as a recessive trait causes Wilson's disease. This gene controls a protein that transports copper in the liver. For people to exhibit signs and symptoms of the disease, they must inherit two copies of the abnormal gene, one from their mother and one from their father. Children won't develop signs and symptoms of the disease if they inherit the gene from only one parent. Instead, they become carriers of the disease, meaning they can pass on a copy of the abnormal gene to their offspring. As many as one in 100 people has an abnormal copy of the Wilson's gene.

Siblings of people with Wilson's disease have a one in four chance of carrying the same gene pattern and should be tested for Wilson's disease. Children of someone with Wilson's disease have a one in 200 chance of having the disorder. Wilson's disease is more common among people of European descent.

When to seek medical advice

Many signs and symptoms of Wilson's disease can be seen and evaluated only with a doctor's help. However, if you notice any of the signs of the disease — difficulty speaking, problems with balance, tremors in your arms and hands, or yellowing of the skin — go to a doctor to see what the problem is. Your doctor can then conduct the proper tests to establish the cause of the signs and symptoms.

Screening and diagnosis

Diagnosing Wilson's disease can be difficult. There is no single test — even with genetic testing — that can diagnose Wilson's by itself. Doctors rely on a combination of symptoms and test results to make the diagnosis. Some tests you might need include:

Blood and urine tests. Your doctor may have you submit blood and urine samples. Ceruloplasmin, a protein in blood that binds copper, is usually quite low in people with Wilson's disease.

Liver biopsy. This procedure requires local anesthesia. As you lie on an examining table, a needle is inserted into the right side of your abdomen to obtain a small sample of liver tissue. The tissue is analyzed in the laboratory for excess copper.

Eye exam. An eye specialist examines your eyes with a special light to look for brown, ring-shaped pigmentation in the cornea.

Complications

One of the most serious complications of the disease is liver damage, which may be so severe that a liver transplant is needed.

Other complications may include:

Damage to the central nervous system. This damage may cause uncontrollable repetitive movements, stiffness, speech problems, and the loss of the ability to function at work or at home. Coordination may also be affected, resulting in clumsiness and awkwardness in mobility.

Psychological problems. These sometimes occur in some people with Wilson's disease. The most common psychological complications include mood swings, depression, inappropriate behavior, agitation, loss of memory and confusion.

Greater risk of bone fractures.

High susceptibility to infection.

Impaired kidney function.

Untreated, Wilson's disease is fatal.

Treatment

Treatment continues for life and involves taking medications that either remove the deposited copper from tissues or render it harmless. The two medications approved for this purpose are penicillamine (Cuprimine, Depen) and trientine (Syprine). Taking zinc acetate (Galzin) helps block copper absorption from the stomach and intestine and may be an alternative treatment for pregnant women and people without symptoms or organ damage.

If you have Wilson's disease, you don't need to follow a rigid diet, but you should avoid high-copper foods, such as nuts, chocolate, dried fruit, liver, shellfish and mushrooms.

With early diagnosis and proper treatment, the long-term outlook and life expectancy are generally good for people with Wilson's disease.