

# What doctors want patients to know about Wilson disease

DEC 26, 2025



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Wilson disease is a rare genetic condition that causes copper to build up in the body, often damaging the liver, brain and other organs before symptoms are recognized. Early signs of the condition—also called Wilson’s disease and [named for the British neurologist](#) who described it in 1912—can be subtle or mistaken for more common conditions. Because of that, many people live with the disorder for years before receiving the diagnosis that can change the course of their health.

The AMA’s What Doctors Want Patients to Know™ series gives physicians a platform to share what they want patients to understand about today’s health care headlines.

In this installment, two physicians took time to share what patients should know about Wilson disease. They are:

- Ryan W. Himes, MD, a pediatric hepatologist at Ochsner Children’s Hospital in New Orleans.
- Hrishikesh V. Samant, MD, a hepatologist at Ochsner Health in Baton Rouge, Louisiana.

[Ochsner Health](#) is part of the [AMA Health System Member Program](#), which provides enterprise solutions to equip leadership, physicians and care teams with resources to help drive the future of medicine.

## Wilson disease is a rare disorder

“Wilson disease is a rare, inherited disorder where the body cannot properly eliminate excess copper,” said Dr. Samant, noting “the prevalence of Wilson disease is about one in 30,000 people in the U.S.”



Hrishikesh V. Samant, MD

“It leads to accumulation of copper in various organs such as the brain, liver and sometimes the heart and eyes,” he said, which “is due to a mutation in the gene called *ATP7B*. This gene is responsible for copper export.”

## The liver and brain are affected the most

“In kids, the first indications of Wilson disease often present in the liver,” Dr. Himes said. “Most commonly, it’s minor elevation in blood tests for liver function, and very often the pediatric patient doesn’t have any outward clinical symptoms.”

For adults with Wilson disease, patients “have more neuropsychiatric and movement symptoms—they experience tremors and personality changes,” said Dr. Samant. “They’re referred to us with increased liver enzymes, sometimes they are referred for fatty liver disease and if they have associated neurological symptoms, we start thinking about Wilson disease.

“They may have some mood swings, mood disorders, depression or anxiety as well,” he added.

Additionally, “very sick adults and children can present with liver failure and are often jaundice, with very yellow skin and eyes, bleeding and bruising easily,” said Dr. Himes. “These are patients who are in the ICU, they are quite sick and need urgent access to liver transplantation in this advanced stage.”

## **Wilson disease can appear as young as 5 years old**

“Wilson disease can present from 5 to 35 years old. This is the most common age group for diagnosis, but there can be some exceptions,” said Dr. Samant. “Normally kids are the first to be diagnosed because they can have scholastic issues related to neurological symptoms, such as handwriting deterioration, poor coordination, slow or slurred speech and cognitive impairment.”

“It’s genetically determined, so these patients have had it from the moment they were conceived,” said Dr. Himes. “It’s just a question of when it expresses itself, which could be at least a few years for copper accumulation to start causing problems and have some detectable features such as elevated blood tests.”

## **Fatty liver disease can be mistaken for Wilson disease**

“Often times Wilson disease is misdiagnosed,” said Dr. Samant. “For example, it may be seen as fatty liver, but it is actually Wilson disease.”

Additionally, some patients “may have some neurological issues that are labeled as depression or anxiety when they may have underlying Wilson disease,” he said, because “the disease involves so many organs.”

## **There are several diagnostic tests**

“The standard first-line tests are blood tests,” said Dr. Himes. “There is a ceruloplasmin level, which is the universal screening test. Low ceruloplasmin level would get you thinking along the lines of Wilson disease and would generally result in someone requesting that you check the urine for copper.”



Ryan W. Himes

“We do that by collecting the urine for 24 hours and sending it to a special lab for measurement of copper,” he said. “Often, at the same time, we’re going to send them to the eye doctor to look in the eyes and look for that telltale Kayser-Fleischer ring, which is seen in many patients who have Wilson disease.”

“From there, sometimes a liver biopsy can be helpful to look both for the changes under the microscope, fat and inflammation, and fibrosis, but also you can send a piece of the liver off for the measurement of copper in the liver,” Dr. Himes said. “There are some well-established thresholds over which Wilson disease is almost certainly the diagnosis.”

## Get gene testing early

“Gene testing is also part of the armamentarium, and you can do that up front as we increasingly do in pediatric hepatology,” said Dr. Himes, emphasizing that getting gene testing early can be very helpful for the patient and the family.

“One of the benefits of doing gene testing is once you’ve determined the variant in the family of the patient with Wilson disease, it makes it somewhat easier to screen other family members for Wilson disease,” he said.

Wilson disease is “an autosomal recessive condition. The autosomal gene passes from one generation to the other,” said Dr. Samant. “With Wilson disease, some people may be a carrier for the gene and they may not present with Wilson disease.”

“For those who have pathogenic variants in the *ATP7B* gene, we have to monitor them for their liver enzymes. Sometimes we start them on treatment just to prevent them from copper accumulation,” he said.

## There are two pillars to treatment

“The management of Wilson disease can be divided into two pillars. One is the primary pillar—which is to control the copper,” said Dr. Samant. “The secondary is to manage complications associated with Wilson disease such as neurological issues or psychiatric issues.”

“Under the primary pillar, we have diet and maintenance therapy such as zinc, which reduces the absorption of copper,” he explained. “When a patient presents with acute liver failure, the only option is liver transplant. Also, for some patients with decompensated cirrhosis, liver transplant is the only option.”

“Zinc chelation with one of the chelator medicines is the most potent form of treatment—more potent than the zinc salts, which are used to prevent absorption of copper,” Dr. Himes said. “Kids who are not as advanced as adult patients and asymptomatic relatives of the patient who has been identified are often good candidates for zinc treatment up front, skipping over the chelators if their disease is quite early in the spectrum.”

## Wilson disease requires ongoing monitoring

Ongoing monitoring is needed for patients with Wilson disease, said Dr. Himes. “We look at blood tests and we look at the urinary copper levels over time as our two main indicators of how we’re doing with the total body copper levels.”

“In kids, though, you do have to account for growth. Kids’ body weight is always changing and the dosing is based on body weight,” he said. “The other thing is adherence to the medication. These drugs only work when you take them.”

“We struggle a little bit when things are going in the wrong direction, whether the disease is really getting worse because the medicines are not working like we think or just the patient is not taking them,” said Dr. Himes, who noted medication side effects include rash allergies, gastritis and bone-marrow suppression. “So, those can be tricky to sort through if patients aren’t being forthright about their medicine taking.”

“The other thing we monitor in adults is organ damage. We monitor for complications, such as developing fibrosis or to see whether they’re developing any neurological issues,” Dr. Samant said. “We also need to monitor any side effects of these medications.”

## Organ damage is the main complication

“The main complication of Wilson disease is organ damage. Once the copper accumulates, there is permanent damage to the organ, mainly the liver,” said Dr. Samant. “The liver develops cirrhosis, it can get decompensated in portal hypertension and there are also complications in neurology with Wilson disease.

“In neurology, we see more of [Parkinson’s disease](#) symptoms such as tremors, dysarthria is most common in about 90% of people and that is signs of permanent organ damage,” he said. “So, even if at that point we start copper chelation, it may not be completely reversible.”

“We certainly worry about liver failure in children. But abruptly stopping taking medications, for example, is a well-known precipitant to liver failure in patients with Wilson disease,” said Dr. Himes. “We’re a little luckier taking care of kids in that they rarely have advanced neurological features and we are much more in a preventative stance to be taking the medicines to prevent the central nervous system problems.”

## Early detection improves prognosis

“Identifying Wilson disease in kids is really beneficial because we’re going to find it before it becomes a catastrophe, before it lands them in the ICU with liver failure,” Dr. Himes said. “And knowledge is power.”

“In putting together Ochsner Health’s center of excellence for Wilson disease, every member of the team is bringing compassion and knowledge to help the patient and the family,” he said. “We want to make sure they understand the disease and understand the steps they can take to keep their health now and for the future.”

## Diet and lifestyle changes are important

“We have registered dietitians who are involved in patient care. In the beginning phases of therapy, everyone should restrict the copper in the diet. At this phase, we’re trying to get rid of years of accumulated copper,” said Dr. Himes. “It used to be that people thought that was a lifetime and absolute thing, and most centers now liberalized that a little bit.”

“Once you get out of the initial phase of reducing copper intake, you enter a maintenance phase,” he said. “It is so invaluable to have dietitians outline foods rich in copper—some of them expected and some of them quite unexpected.” he said. “Examples include organ meats—such as beef liver—and sunflower or sesame seeds, nuts, chocolates and shellfish. Other surprising sources are whole grains, legumes and some vegetables, such as avocados, potatoes and leafy greens.

For adults, “we tell them to avoid alcohol, which can damage the liver,” said Dr. Samant.

“The other part of [eating and nutrition](#) is keeping a healthy body weight. By far the most common chronic liver disease in young people and adults is fatty liver disease and that’s totally preventable,” Dr. Himes said. “That has to do with obesity and consuming too many calories or the wrong types of calories. So, good lifestyle advice to help prevent that really common liver disease being superimposed on this uncommon liver disease is quite important.”

“You cannot control Wilson disease only on diet,” said Dr. Samant. “It’s important to know that even if you have a diet restricted in copper, you might still need medication because copper is present everywhere.”

## Wilson disease is a lifelong condition

“Gene testing, whether it’s for Wilson disease or other genetic disorders, is often a really personalized decision,” Dr. Himes said. “Some people, particularly if they’re outwardly well, prefer not to know their risk for future disease, so I always have a conversation to try to get a sense for where the family’s head is at and what they want to know.”

“It’s good practice to screen first-degree relatives of a patient with Wilson disease, and you can do that with standard blood and urine testing, or you can do it if you’ve established genetic changes in the patient,” he said. “You can look for that exact same gene change in the family members, which is a relatively simple and rapid way to approach it.”

“That’s the recommendation, because you may uncover other members of the family who have the disease but haven’t yet manifested the first symptoms,” Dr. Himes said. “And it provides an opportunity to intervene earlier on those patients before they become unwell.”

“Even if they have Wilson disease, if we control the copper overload, they can have a normal or near normal lifespan,” Dr. Samant said.

## Connect with other Wilson disease patients

It is important to “find community among other patients with Wilson disease,” said Dr. Himes. “This is a rare condition and navigating it alone is lonely.

“You can learn from other people in the community who have Wilson disease and figure out things that have worked for them and probably tap into their network of experts who they may have curated through their own care,” he added.

## More awareness is needed

“[There is a study](#) that showed about 70% of people are misdiagnosed who have Wilson disease,” said Dr. Samant, adding that “the median time of diagnosis is two years from symptoms to the diagnosis.”

That is why “we need more awareness and then we need to have more centers of excellence like at Ochsner Health where we can have a team approach where everybody is aware and committed to treating Wilson disease,” he said.