Medical Xpress

Pushing the boundaries of treatment for Wilson disease

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Uyen To, MD. Credit: Yale University

Uyen To, MD, assistant professor of medicine (digestive diseases) and transplant hepatologist, discusses how she first became interested in studying Wilson disease, the wide spectrum of symptoms caused by the rare condition, and what's next in this fascinating field of research.

What is Wilson disease?

Wilson disease, also known as Wilson's disease, is a genetic disorder in which large amounts of copper accumulate in the body. Typically, excess copper is excreted into the gut through bile formed by the liver. If you have Wilson disease, the copper accumulates in your liver and is released directly into your bloodstream, damaging your liver, brain, and other organs. Because the condition is so rare, it is often misdiagnosed.

How did you first become interested in studying this rare condition?

When I was a transplant fellow at YSM, Dr. Michael Schilsky invited me to come by his Wilson disease clinic. There was a wide span of patients in the clinic, from young children who had been diagnosed in utero to 80-year-olds. Patients can present with neurological issues, hepatic issues, psychiatric issues, or a combination of all three. The breadth was amazing. I was fascinated by how the disease affected systems beyond the liver, which is my favorite organ.

During my fellowship, one of our patients went from not being able to eat or speak, to walking the hallways with his mom and telling me about his favorite TV shows and what he was doing at home. This person, who couldn't even swallow when I met him, came to life. That was the moment I knew this was my life's work.

I enjoy the multidisciplinary approach to taking care of patients with this disease—coordinating with our pharmacists, psychiatrists, nutritionists, and neurologists—a strategy that we borrow from my other passion, transplant. I'm lucky to come into the field when there's so much research being done about the therapies, the diagnostic criteria, and what we can do for these patients. Dr. Schilsky, my mentor, is a pioneer on that front.

What do you find intriguing about the disease?

It's amazing that one protein malfunction can cause such a spectrum of disease. Patients can have a wide variety of neurological issues, especially when copper deposition happens in the brain. We've had patients who are manic and bipolar. We've had patients who have Parkinsonian symptoms. They can barely walk. They're drooling. They can't speak. People often don't think about Wilson's—they think the symptoms are related to early onset Parkinson's, or that maybe a young child or teenager is going through a tough time.

There is still so much to learn. There is a great need not only for better diagnostic criteria, but also a better test to assess a patient's copper balance. Currently we piece together a combination of the blood copper and the urine copper and a certain type of protein in the blood to calculate what potential amount of free copper is in the blood. And the assays are not perfect. Copper hides in different pockets. It hides in the liver. It hides in the brain.

Looking forward, what excites you about your research?

For so many years, we had only limited therapy. It was either zinc, trientine, or penicillamine, and a low-copper diet that we were telling people to adhere to. These patients took their medications multiple times a day on an empty stomach, and some of these medications can cause discomfort and side effects. Compliance and cost are big issues. To his credit, Dr. Schilsky has worked with the pharmacists here to help our patients get the medications they need without paying outrageous fees.

Now, not only are we looking at new medications that remove copper from the body, but also different methods to assess copper balance. The newest and most exciting treatment is gene therapy. We just enrolled our first patient, and we're hoping to enroll more.

Our work in gene therapy can be translated to other conditions, even more common ones. It's exciting because we are pushing the boundaries of treatment and finding a cure. This is the wave of the future.

Provided by Yale University

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