Copper Overload: Understanding Wilson Disease A Guide for Teens

By: Riddhi Ramesh, Wilson Disease Patient Volunteer

Have you ever thought about how the food we eat can affect our bodies in surprising ways? One fascinating example is Wilson disease, a rare genetic disorder that reveals how important our livers are in dealing with a mineral we often overlook: copper. Did you know we all need copper to be healthy? Some high copper food sources are chocolate, shellfish, nuts and mushrooms.

Wilson disease is caused by a mutation in the *ATP7B* gene, which is responsible for managing copper in our bodies. In a healthy person, the liver is able to control copper levels by incorporating it into needed proteins and safely removing any excess. However, people with Wilson disease are born with a broken copper regulator. The liver struggles to get rid of excess copper, leading to copper accumulation. That excess copper not only causes liver damage but can also damage to other organs—especially the brain.

As copper accumulates in the liver, people with Wilson disease may start to experience symptoms like fatigue, jaundice (which makes the skin and eyes yellow), and pain in the abdomen. These initial signs can be puzzling, often mistaken for more common illnesses. As the disease progresses, serious liver problems like liver inflammation or cirrhosis can occur. If the copper settles into the brain, tremors, difficulty speaking and walking, drooling and behavioral problems can develop. In many cases, people can experience bouts of depression. It's been called "Great Masquerader" because it can look like a liver, neurological or psychiatric disorder or a mix of these symptoms.

So, how do doctors figure out if someone has Wilson disease? They use a mix of laboratory tests, imaging and sometimes a liver biopsy. An eye exam may reveal tell-tale copper rings in the eyes. Early detection is crucial because treating this disease is all about removing excess copper. The treatment for Wilson disease often includes medications that remove excess copper from the body and limiting how much copper you absorb from your diet. In more extreme cases, a liver transplant might be required.

Understanding Wilson disease offers a valuable opportunity to explore rare liver conditions and their impact on overall health. This genetic disease underscores the importance of early diagnosis and the serious consequences of untreated health issues. Rare liver diseases like Wilson disease remind us that early symptoms can often go unnoticed or be attributed to more common illnesses.

For young adults, who may feel invincible, it's easy to ignore subtle signs that something is wrong. And, rare diseases are often unfamiliar to doctors, making it essential to stay persistent when faced with unexplained symptoms. By increasing awareness and advocating for our health, we can help ensure that these conditions are identified and treated in a timely manner and lives can be saved.