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## EXPERT INSIGHTS ON WILSON DISEASE AND A HEALTHY PARFAIT RECIPE

*Michael Schilsky, M.D., shares crucial insights on advancements in Wilson disease, plus a liver-friendly parfait recipe.*

**By Lana Pine** | Published on March 17, 2025

6 min read

Wilson disease is a rare genetic disorder that affects copper metabolism, leading to severe liver and neurological complications if left untreated. In this Q&A, Michael Schilsky, M.D., medical director of liver transplantation at Yale New Haven Hospital, explains how Wilson disease is diagnosed, treated and managed.

He also shares the latest advancements in research and offers practical advice for newly diagnosed patients. Plus, enjoy a simple and nutritious Greek Yogurt Parfait recipe,



*Credit: Adobe  
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[originally](#) included in the [American Liver Foundation's \(ALF's\)](#)

30-day meal plan, a great addition to a liver-friendly diet.

## **Nutrition Facts Per Serving (1)**

- Calories: 130
- Total fat: 0 grams
- Saturated fat: 0 grams
- Trans fat: 0 grams
- Sodium: 60 milligrams
- Total carbohydrate: 10 grams
- Dietary fiber: 0 grams
- Sugars: 10 grams
- Protein: 18 grams

## **Greek Yogurt Parfait**

### **Ingredients:**

- 1 cup Greek yogurt (plain or flavored, depending on your preference)
- Honey (to taste)
- Optional toppings: fresh berries, nuts, granola, etc.

### **Instructions:**

1. Scoop 1 cup of Greek yogurt into a bowl or serving dish.
2. Drizzle honey over the Greek yogurt according to your taste preference. Start with a tablespoon and adjust based on how sweet you want it.
3. Optional: Top your Greek yogurt with fresh berries (such as strawberries, blueberries or raspberries), nuts (like

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almonds or walnuts) or granola for added texture and flavor.

4. Enjoy your Greek yogurt with honey as a healthy and delicious breakfast!

## **What is Wilson disease, and how does it affect the body?**

**Michael Schilsky, M.D.:** Wilson disease is a disorder that is inherited in an autosomal recessive fashion due to mutations in the gene ATP7B that causes a failure in copper homeostasis with pathological retention of copper first in the liver, and later in other organs, mainly in the central nervous system. The natural history of untreated Wilson disease is one of progressive liver injury from asymptomatic to full-blown liver failure due to copper accumulation over time, and later in the brain, leading to neurological disability or to psychiatric symptoms.

## **Why is Wilson disease often misdiagnosed, and what should patients do if they suspect they have it?**

**MS:** The phenotype for Wilson disease is very variable and its symptoms may mimic other disorders of the liver or neurological or psychiatric disease. This leads to a delay in diagnosis and treatment and more symptomatic diseases, in particular with those who have neurological or psychiatric symptoms as their presenting signs of disease. There are specific tests and examinations that can be ordered to help establish Wilson disease as the cause of the symptoms. The Leipzig scoring system uses these to help with diagnosis, and scores of higher than 3 are suggestive of the diagnosis. The elements of this score include serum ceruloplasmin, a lower hemoglobin due to hemolysis (nonimmune), an

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hour urine copper excretion, liver biopsy for histology and for copper quantitation, and mutation analysis of DNA for ATP7B pathogenic mutations. Each of these has weighted scores that can be tabulated to determine if more testing is needed, the diagnosis is established or alternative diagnosis should be sought.

### **What are the main treatment options for Wilson disease?**

**MS:** The main treatments include medical therapy with chelating agents, D-penicillamine and trientine or zinc salts. Additional treatment suggestions include adherence to a low copper diet, and for those with cirrhosis, standard measure such as screening/surveillance for esophageal or gastric varices and for hepatocellular carcinoma (HCC). Medical therapy is lifelong. For those where medical therapy is not effective or who present as too ill for medical treatment, liver transplant is lifesaving and cures the disorder.

### **What challenges do patients with Wilson disease face, and how can they best advocate for their health?**

**MS:** This is a rare disorder, and there are often few local experts for the care of this disorder. There are centers of excellence for the care of these patients as designated by the patient-run organization [Wilson Disease Association](#), and patients may seek expert multidisciplinary care at these sites. I would suggest patients join the organization and learn more about the disease and become advocates for affordable and accessible care for their disease.

### **What advancements in research and treatment give**

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**MS:** There are new clinical trials ongoing for gene replacement therapy, and future trials planned for gene repair and for new medical therapies with novel mechanisms of action that advance current therapy. Additionally, new testing is being developed to help with diagnosis that may be useful for newborn screening programs (ATP7B peptide analysis in dried blood spots), and assays to assess copper status in the patient that may be useful for monitoring treatment.

**What advice do you have for someone newly diagnosed with Wilson disease?**

**MS:** Obtain a careful evaluation to confirm your diagnosis and determine the best therapy for your disorder. Do not neglect to treat other symptoms such as neurological or mental health issues that can arise. Most importantly, take your medications as prescribed and avoid anything that might be injurious to the liver. Lastly, make sure family members are appropriately screened for this disorder.

*In addition to liver-healthy recipes, the ALF offers a simple liver health quiz, available in [English](#) and [Spanish](#). To learn more about liver health, including education, awareness and screening, visit [Think Liver Think Life](#), and to help researchers find better treatments and cures for liver diseases, visit [ALF's Patient Registry](#).*

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