



Unusual Presentation of a Rare Disease

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Introduction

Wilson disease is a rare inherited disorder that is characterized by the accumulation of copper in the body. Because high levels of copper are toxic to tissues and organs, this build up can lead to damage of the liver, brain and eyes. Signs and symptoms of Wilson disease include chronic liver disease, central nervous system abnormalities, and psychiatric (mental health-related) disturbances.^[1]

^[2] It is caused by a mutation of the *ATP7B* gene and is inherited in an autosomal recessive manner.^{[3][4]} Although there is no cure for Wilson disease, therapies exist that aim to reduce or control the amount of copper that accumulates in the body.^{[1][4]}

Case Report

A 20 -year-old woman presented with history of marriage since 3 years, thereafter she conceived and got spontaneously aborted at 6 months of amenorrhea, conceived again after 1 year and got spontaneous aborted at 3 months of amenorrhea. Patient then shifted herself to parents home and gradually she went mentally retard and developed depressive symptoms, intermittent tremor of the hands. She also reported difficulties with

concentration. Physical examination of the eyes revealed bilateral Kayser–Fleischer rings (Panel A, arrows). Neurologic examination revealed mild dysphagia and dysarthria, and bradykinesia. Laboratory tests revealed elevated serum levels of alanine aminotransferase, aspartate aminotransferase, and γ -glutamyltransferase, as well as low serum levels of ceruloplasmin (0.12 g per liter; reference range, 0.2 to 0.5) and copper (7.1 μ mol per liter; reference range, 11 to 22); the urinary copper excretion was elevated, at 3.9 μ mol per 24 hours (reference range, 0 to 1). A diagnosis of Wilson's disease was made and patient was started on D- penicillamine, zinc and benzodiazepine. Patient is now symptomatically better, no psychological symptoms, conceived for the third time with 7 months of amenorrhea and expecting a healthy baby.

Discussion

Wilson disease can affect many different systems of the body. Affected people often develop signs and symptoms of chronic liver disease in their teenaged years or early twenties. These features may include jaundice; abnormal fluid retention which can lead to swelling of the legs and/or

abdomen; weight loss; nausea and vomiting; and/or fatigue. Unfortunately, some people may not experience any signs until they suddenly develop acute liver failure.^{[5][1][2]}

Affected people often experience a variety of neurologic (central nervous system-related) signs and symptoms, as well. Neurologic features often develop after the liver has retained a significant amount of copper; however, they have been seen in people with little to no liver damage. These symptoms may include tremors; muscle stiffness; and problems with speech, swallowing and/or physical coordination. Almost all people with neurologic symptoms have Kayser-Fleisher rings - a rusty brown ring around the cornea of the eye that can best be viewed using an ophthalmologist's slit lamp.^{[5][1][2]}

About a third of those with Wilson disease will also experience psychiatric (mental health-related) symptoms such as abrupt personality changes, depression accompanied by suicidal thoughts, anxiety, and/or psychosis.^{[5][1][2]}

Other signs and symptoms may include:^{[5][1][2]}

- Menstrual period irregularities, increased risk of miscarriage and infertility in women
- Anemia
- Easy bruising and prolonged bleeding
- Kidney stones
- Early-onset arthritis
- Osteoporosis

Conclusion

The long-term outlook (prognosis) for people with Wilson disease varies and largely depends on timely diagnosis and treatment. If the condition is detected early and treated appropriately, people with Wilson disease can usually enjoy normal health and a normal lifespan. Unfortunately, untreated Wilson disease is associated with severe brain damage, liver failure, and death

References

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