

Atypical Case of Wilson's Disease Without Early GI Symptoms

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Introduction

Wilson disease is Excessive copper deposition in the body organs, particularly in the liver and brain, is a typical feature of the disease which results from a mutation in the copper-transporting gene ATP7B. Wilson's Disease presentation includes hepatic, neurologic, and psychiatric disorders. (1)

Objective

In a healthy body, the liver filters out excess copper and releases it through urine. With Wilson's disease, the liver cannot remove the extra copper properly, thus accumulating it in organs causing bluish discoloration in the nails, kidney stones, premature osteoporosis, lack of bone density, arthritis. The Wilson's Disease occurs in 1 in 30,000 people worldwide. (2)

Method

A 19-year-old woman presented with lower limb pain, weakness, and progressive dystonic movements in her upper limbs, worsening over a month. In the last two weeks, she struggled to stand and became bedridden. Despite a normal spinal MRI, her symptoms progressed to catatonia, personality changes, and behavioral disturbances.

Result

The patient started on D-penicillamine for promoting urinary copper excretion. Zinc supplementation was also used to hinder copper absorption from the gastrointestinal tract. Additionally, antioxidant therapy with vitamin E was given due to its role in scavenging free radicals and its deficiency in Wilson's disease, along with dietary restrictions.

Discussion

The prognosis for Wilson's disease in the 19-year-old patient is influenced by early neurological and psychiatric symptoms alongside Kayser-Fleischer rings without gastrointestinal manifestations. Treatment initiation with chelating agents may improve outcomes, but advanced liver involvement suggests a complex clinical course requiring careful management and monitoring.

Conclusion

A confirmed diagnosis of Wilson's disease relies on clinical manifestations such as cirrhosis, neurological symptoms, and the presence of Kayser-Fleischer rings. Early recognition and initiation of treatment are pivotal in halting disease progression and enhancing patient outcomes.