E-Poster

EP-152

The Importance of Early Diagnosis in Wilson Disease

Topic: Diagnosis

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BACKGROUND

Wilson disease (WD) is a rare autosomal recessive disorder of copper metabolism, leading to excessive copper accumulation in the liver, brain, and other organs. Early diagnosis is essential to prevent irreversible neurological and hepatic damage. This case highlights the importance of early detection in reducing disability and improving prognosis.

CASE DESCRIPTION

We present the case of a 23-year-old male, the eldest of four siblings from a consanguineous marriage, diagnosed with Wilson disease at 17 years old. His younger sister was also diagnosed but received early treatment. The patient initially presented with neurological symptoms, including tremor, and mild ataxia, leading to diagnostic confirmation through biochemical tests, brain magnetic resonance imaging, and genetic testing. By age 22, his condition had worsened, presenting with hypersalivation, anarthria, parkinsonism, dystonia and spasticity, severely impacting daily life. Examination revealed rigidity, resting tremor, bradykinesia, dystonic posturing, bilateral knee contractures, and cognitive difficulties. His condition resulted in significant motor disability and dependency for daily activities. The patient received physiotherapy, occupational therapy, and botulinum toxin injections for parotid glands , still his functional prognosis remained poor. In contrast, his younger sister, diagnosed earlier and treated promptly, remained asymptomatic and pursued her studies successfully.

DISCUSSION

Wilson disease exhibits varied hepatic and neurological manifestations. Delayed diagnosis can lead to progressive neurological impairment, as seen in our patient, despite treatment. Early intervention with chelation therapy and appropriate management is critical for preventing severe disability. Family screening in at-risk populations, particularly in consanguineous families, is essential to ensure early diagnosis and improve patient outcomes. The stark difference in disease progression between our patient and his younger sister emphasizes the impact of early intervention. This case reinforces the need for heightened clinical suspicion and timely therapeutic initiation in suspected cases of Wilson disease.

CONCLUSION

Early diagnosis of Wilson disease is crucial for preventing severe neurological impairment. Prompt treatment significantly improves outcomes, as illustrated by the contrast between our patient and his sister. Family screening and early therapeutic intervention should be prioritized for at-risk individuals.

Statement of Disclosures

None

Doi: 10.5606/archisprm.2026.EP-152

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