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Case Report

Wilson Disease Diagnosis and Management

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Abstract

A 16-year-old girl, born to non-consanguineous parents, presented with 8 months of speech disturbance, 7 months of upper limb tremors, and 5 months of abnormal limb posturing with gait difficulty. She sought evaluation at a nearby facility for these symptoms, where she was diagnosed with Wilson's disease. One month into treatment, she began experiencing continuous posturing of both upper limbs, characterized by backward extension of both hand, striatal toe, slowness of gait, and started having falls. Laboratory investigations revealed decreased levels of serum ceruloplasmin and copper.

Over time, a wide range of presentations has been documented, including skeletal and hematological manifestations, along with varied combinations of symptoms.

Treatment should be started promptly and carefully monitored, especially for individuals who are presymptomatic at the time of screening. She was initiated on Penicillamine at a dose of 250 mg once daily for 2 weeks, which was then increased to 250 mg twice daily for the subsequent 2 weeks.

Screening first-degree relatives of patient for detecting individuals who may be asymptomatic. Screening primarily involves gathering a medical history focused on previous jaundice, liver issues, and neurological or psychiatric symptoms.

Keywords: *Wilson Disease; Screening; Chelating Agent; Ceruloplasmin; Penicillamine*

Abbreviation

WD: Wilson Disease

Introduction

The article explores recent advancements in both diagnostic techniques and therapeutic options for Wilson's disease.

If a sibling is found to carry the mutated gene, they can begin treatment early, even before symptoms appear.

Materials and Methods

Screening first-degree relatives of patients with Wilson's disease, advanced genetic testing, measuring serum ceruloplasmin, urinary copper levels, and imaging studies, symptomatic care and traditional chelation therapy.

Results and Discussion

- Recent advances in the genetic understanding of Wilson's disease have shifted the focus toward gene therapy, innovative treatments targeting the ATP7B gene, and approaches aimed at correcting mutant proteins to prevent copper buildup.
- Slit-lamp microscope allows the ophthalmologist to detect Kayser-Fleischer rings.
- Hepatic diagnosis shows that during a biopsy, a thin needle through the skin directly into the liver.
- Penicillamine (Cuprimine, Depen), which acts as a copper-chelating agent to help remove excess copper from the body.
- Surgical treatment with deep brain stimulation DBS has been attempted in select patients with refractory tremors and dystonia.

Conclusion

Wilson's disease is a treatable genetic disorder affecting copper metabolism.

Lately, a range of unusual presentations of Wilson's disease has been reported. With advances in genetic testing and improved knowledge of Wilson's disease, numerous phenotypes and genotype associations have been identified.

Several therapeutic agents have been explored to enhance Wilson's disease management, but many newer treatments are still under investigation.

Overall, timely diagnosis and early treatment are crucial for halting the progression of Wilson's disease [1-4].

Conflict of Interest

There is no conflict of interest.

Financial Interests

Pharmaceutical company funding: If a researcher receives funding or compensation from a company developing treatments for Wilson disease (like zinc therapies or chelators such as trientine or penicillamine), they might be biased-intentionally or not-in designing trials, interpreting results, or publishing findings.

Stock Ownership

Owning shares in a biotech company involved in treating Wilson disease could influence the researcher's decisions or interpretation of data.

Patent Ownership

Holding a patent for a diagnostic method or treatment for Wilson disease can also be a significant COI.

Clinical or Professional Bias

1. Dual roles: A physician-researcher who treats Wilson disease patients may have a vested interest in proving the effectiveness of a treatment they personally use.
2. Academic pressure: If a researcher's reputation or promotion depends on producing positive results, they may feel pressured to highlight successes or downplay negative findings.

Personal or Ideological Bias

1. Advocacy affiliations: A strong affiliation with patient advocacy groups could lead a researcher to emphasize patient-centered outcomes while neglecting critical safety data.
2. Family connection: Having a family member with Wilson disease could unconsciously influence the researcher's objectivity.

Bibliography

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