ISSN No:-2456-2165 https://doi.org/10.5281/zenodo.14979294

Silent Copper Storm: A Case of Wilson Disease Presenting as Icterus in an 18-Year-Old Female

[1]Dr. Swetha Sunkara; [2]Dr. Rekha S Patil

[1,2] Institution: Kle Academy of Higher Education and Research, Belagavi

Publication Date: 2025/03/08

Abstract: Wilson disease is a rare autosomal recessive disorder characterized by impaired copper metabolism, leading to copper accumulation in various organs.

This case report describes a 19-year-old female who presented with icterus as the initial manifestation of Wilson disease. Early recognition and treatment are crucial in preventing irreversible complications. This report highlights the diagnostic challenges and management strategies associated with this rare condition.

How to Cite: Dr. Swetha Sunkara; Dr. Rekha S Patil (2025) Silent Copper Storm: A Case of Wilson Disease Presenting as Icterus in an 18-Year-Old Female. *International Journal of Innovative Science and Research Technology*, 10(2), 1703-1704. https://doi.org/10.5281/zenodo.14979294

I. INTRODUCTION

Wilson disease results from mutations in the *ATP7B* gene, leading to defective copper transport and accumulation in the liver, brain, and cornea. Clinical presentations vary from hepatic dysfunction to neuropsychiatric disturbances. Early diagnosis is essential, as prompt treatment can prevent progressive organ damage. Icterus as a primary symptom in young adults warrants consideration of Wilson disease in differential diagnoses.

II. CASE PRESENTATION

A 18 year old female presented with a two-week history of yellowish discoloration of the eyes and skin, dark urine, and generalized fatigue. There was no history of abdominal pain, fever, or neuropsychiatric symptoms. She has no history of alcohol consumption, drug use, or recent travel.

III. EXAMINATION

- blood pressures 120/80 mmhm, pulse rate 90 bpm, saturation 98 % on room air
- No hepatosplenomegaly or ascites, Icterus present in sclera and skin,
- Neurological examination was unremarkable

IV. INVESTIGATIONS

- Liver function tests: Elevated bilirubin (total: 9 mg/dL; direct: 5 mg/dL), AST/ALT ratio > 2
- Serum ceruloplasmin: Decreased (10 mg/dL)
- 24-hour urinary copper excretion: Elevated (>100 µg/24 hours)
- Slit-lamp examination: Kayser-Fleischer rings detected
- Abdominal ultrasound: Mild hepatomegaly with increased echogenicity

V. DIAGNOSIS

The combination of icterus, low serum ceruloplasmin, elevated urinary copper, and Kayser- Fleischer rings confirmed the diagnosis of Wilson disease.

VI. MANAGEMENT

The patient was started on oral chelation therapy with D-penicillamine 250 mg/day and zinc supplementation. A low-copper diet was advised. Regular monitoring of liver function and copper levels was initiated. Genetic counseling was done for family members.

VII. DISCUSSION

Wilson disease often presents with variable clinical manifestations. In young patients presenting with unexplained icterus, it is essential to consider Wilson disease, especially when common causes are excluded. Kayser-Fleischer rings, though not always present, are highly suggestive when observed. Chelation therapy remains the management for preventing disease progression.

ISSN No:-2456-2165 https://doi.org/10.5281/zenodo.14979294

VIII. CONCLUSION

This case emphasizes the need for heightened clinical suspicion of Wilson disease in young patients presenting with icterus. Early detection and appropriate management can significantly improve prognosis and prevent severe complications.

REFERENCES

[1]. Roberts EA, Schilsky ML. Diagnosis and treatment of Wilson disease: An update.
[2]. *Hepatology*. 2008;47(6):2089-2111.
[3]. Ferenci P. Pathophysiology and clinical features of Wilson disease. *Metab Brain Dis.* 2004;19(3-4):229-239.
[4]. Ala A, Walker AP, Ashkan K, Dooley JS, Schilsky ML. Wilson's disease. *Lancet*. 2007;369(9559):397-408.