Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Paediatrics

Wilson's Disease- A Child with An Atypical Presentation

Dr. Shahed Haider Chowdhury[1], Dr. Khondoker Ehsanul Arefin[2*], Dr. Mohsina Akter Lucky[3]

[1]Assistant Professor, Department of Ophthalmology, Bangladesh Medical College Hospital, Dhaka, Bangladesh
[2]Associate Professor, Department of Paediatrics, Bangladesh Medical College Hospital, Dhaka, Bangladesh
[3]Associate Consultant, Department of Paediatrics, MEDIX, United Health Care, Dhaka, Bangladesh

DOI: 10.36347/sjmcr.2024.v12i03.009 | **Received:** 19.11.2023 | **Accepted:** 27.12.2023 | **Published:** 15.03.2024

***Corresponding author:** Dr. Khondoker Ehsanul Arefin
Associate Professor, Department of Paediatrics, Bangladesh Medical College Hospital, Dhaka, Bangladesh

Abstract **Case Report**

Wilson's disease is a hereditary disorder of copper metabolism which leads to copper overload in different tissues of the body. Clinical presentation of Wilson disease can vary widely; therefore, diagnosis is not always straightforward. Here we report a 13-year-old girl presented with diffuse persistent stabbing pain in the abdomen, jaundice & dark urine. She had no history of unconsciousness, convulsion, deterioration of school performance, or alteration of sleep pattern. On examination, she was ill-looking, pale, and icteric. The liver was enlarged. Higher psychic Function was intact, with no neurological deficit. A slit-lamp examination by an ophthalmologist promptly revealed the presence of a "Kayser Fleischer Ring" in both eyes. On investigation, hemoglobin was 5.73g/dl, Total bilirubin-37.82mg/dl, ALT-50 IU, AST-95 IU, PT/INR-3.20, serum ceruloplasmin 21mg/dl. Twenty-four hours of urinary copper excretion after penicillamine challenge was 8784 µg/24 hours. We diagnosed the case as Wilson's disease. This case report aims to share our experience regarding the clinical presentation and diagnosis of Wilson's disease in a child where Kayser–Fleischer rings in eyes appeared before neurological manifestation occurs and normal serum ceruloplasmin which often creates a diagnostic dilemma.

Keywords: Wilson's disease, hereditary disorder, diagnosis, convulsion, penicillamine challenge.

INTRODUCTION

Wilson's disease is an autosomal recessive disorder of copper metabolism which leads to copper overload [1]. Copper accumulates not only in the liver and brain but also in other organs [2]. The prevalence of WD is approximately 0.5 cases per 100000 [3]. Clinical presentation of Wilson disease can vary widely; therefore, diagnosis is not always straightforward. Wilson disease is not just a disease of children and young adults, but may present at any age. The key features of Wilson disease are liver disease and cirrhosis, neuropsychiatric disturbances, Kayser– Fleischer rings (K-F rings), and acute episodes of hemolysis, often in association with acute liver failure [4]. Wilson's disease in children may be obscure and requires extensive investigation to establish the diagnosis [5].

CASE PRESENTATION

A 13-year-old female, 2nd child of 1st-degree consanguineous parents, from Dhaka, presented with diffuse persistent stabbing pain in the abdomen which was more in the right upper quadrant without any radiation, with no aggravating nor relieving factors. Appetite was not reduced except for mild nausea. She was active. On 3rd day, the mother noticed gradually increasing yellowish discoloration of eyes, hands, and feet. Her urine was also very dark. She was drinking adequate water. The stool was normal. She had no itching. She felt feverish but the temperature was not recorded. She had no history of blood transfusion. She has no pets. She could not attend school. Her family lives in a clean well-ventilated house and drinks boiled water. She does not take outdoor food. She has no history of jaundice. She had no history of unconsciousness, convulsion, deterioration of school performance, or alteration of sleep pattern. She has one elder brother and all her family members are healthy.

Though she has not started developing secondary sexual characteristics, her physical and mental growth has followed the developmental milestones until now. On examination, she was ill-looking, pale, and icteric. Skin did not show any spider nevi. Vitals were found normal. The liver was palpable, 3 cm enlarged from the right costal margin along mid clavicular line. The surface was smooth, firm in consistency, tender, and had a rounded lower border. The upper border of liver

dullness was found in the right 5th intercostal space. Higher psychic Function was intact, with no scanning or slurring of speech, and the handwriting was average. Muscle power- 5/5.Tone- normal. Intention and flapping tremors were absent. A slit-lamp examination by an ophthalmologist promptly revealed the presence of K-F Rings in both eyes. On investigation, hemoglobin was 5.73g/dl, Total bilirubin-37.82mg/dl, direct-20.54mg/dl and indirect-17.28mg/dl, ALT-50 IU, AST-95 IU, PT/INR-3.24, Serum ceruloplasmin 21 mg/dl(normal), Hepatotropic viral markers – Negative, urine R/M/E showed 8-10 pus cell/HPF and granular cast 2-3/LPF.Twenty-four hours of urinary copper excretion after penicillamine challenge test was 8784 µg/24 hours.USG of the abdomen showed hepatomegaly with increased echogenicity of hepatic parenchyma. We diagnosed the case as Wilson's disease. She was treated with Penicillamine and zinc acetate, which showed drastically improving PT/INR-1.9 and total Bilirubin - 21mg/dl after 1 week.

We claimed it as an atypical presentation of Wilson's disease since the K-F rings appeared before any neurological manifestations with normal serum ceruloplasmin and acute liver failure without any history of recurrence of jaundice.

DISCUSSION

Diagnosis of Wilson's disease in children is challenging [4]. Our patient had jaundice, hepatomegaly, high PT/INR - 3.24, very high penicillamine challenge test result- 8784 µg/24 hours with normal serum ceruloplasmin and the presence of K-F rings by slit lamp examination. Similar cases were reported by Yüce & colleagues where four patients were diagnosed with Wilson's disease and presented with hepatic manifestations with normal serum ceruloplasmin levels and absence of K-F rings, diagnosis was confirmed by a strong family history and increased urinary and hepatic copper amounts [6]. Approximately 90% of all patients with Wilson disease have low serum ceruloplasmin levels [7]. Our patient had normal serum ceruloplasmin.

Neurological manifestation of Wilson's disease appears in 2nd and 3rd decade [7]. K-F rings are present almost invariably in neurological Wilson's disease [9, 10]. Our patient's age was 13 years. She had K-F ring in both eyes but there was no neurological manifestation. Neurological manifestation without K-F ring is found in 5% of cases [9]. But K-F ring without neurological disorder is rare which is our case.

Mantas & colleagues reported a case with K-F ring without neurological disorder. A 22-year-old woman presented with fatigue, decreased appetite, and jaundice. Her investigations revealed abnormal liver function tests, including low ceruloplasmin and alkaline phosphatase, the presence of K-F rings suggested the diagnosis of Wilson's disease. She had no neurological symptoms [11]. Similar case was reported by Liu & colleagues where an 18-year-old woman was found to have bilateral K-F rings. She had no systemic symptoms. The serum ceruloplasmin level was normal. Liver function tests were also normal. Further evaluations with 24-hour urine copper assay and liver biopsy were positive for Wilson disease [12].

CONCLUSION

Wilson's disease in children may be obscure and requires extensive investigation to establish the diagnosis because history, clinical presentation, and investigations are often misleading, by diagnosis of Kayser-Fleischer rings, ophthalmologists may play a critical role in the early recognition and proper evaluation of such patients.

REFERENCES

1. Bayram, A. K., Gümüş, H., Arslan, D., Özçora, G. K., Kumandaş, S., Karacabey, N., Canpolat, M., & Per, H. (2016). Neurological features and management of Wilson disease in children: an evaluation of 12 cases. *Turk Pediatri Ars*, *51*(1), 15-21. PMID: 27103860; PMCID: PMC4829162. doi: https://doi.org/10.5152/turkpediatriars.2016.3080
2. Esezobor, C. I., Banjoko, N., Rotimi-Samuel, A., & Lesi, F. E. A. (2012). Wilson disease in a Nigerian child: a case report. *Journal of Medical Case Reports*, *6*(1), 1-3. https://doi.org/10.1186/1752-1947-6-200
3. Rodriguez-Castro, K. I., Hevia-Urrutia, F. J., & Sturniolo, G. C. (2015). Wilson's disease: A review of what we have learned. *World J Hepatol*, *7*(29), 2859-70. https://dx.doi.org/10.4254/wjh.v7.i29.2859
4. Ferenci, P. (2017). Diagnosis of Wilson disease. *Handb Clin Neurol*, *142*, 171-180. https://doi.org/10.1016/b978-0-444-63625-6.00014-8
5. Manolaki, N., Nikolopoulou, G., Daikos, G. L., Panagiotakaki, E., Tzetis, M., Roma, E., Kanavakis, E., & Syriopoulou, V. P. (2009). Wilson disease in children: analysis of 57 cases. *J Pediatr Gastroenterol Nutr*, *48*(1), 72-7. doi: 10.1097/MPG.0b013e31817d80b8. PMID: 19172127.
6. Yüce, A., Koçak, N., Ozen, H., & Gürakan, F. (1999). Wilson's disease patients with normal ceruloplasmin levels. *Turk J Pediatr*, *41*(1), 99-102. PMID: 10770682.
7. Richard, K., & Gilroy, R. K. Wilson's disease workup. Medscape 2019; retrieved from https://emedicine.medscape.com/article/183456-workup?form=fpf#c7
8. Yong, L. L., Qun, Z. X., Wei, T. W., Ming, Y. W., Zhen, C. H., & Yu, W. (2019). Acute onset neurological symptoms in Wilson disease after traumatic, surgical or emotional events: A cross-

sectional study. *Medicine, 98*(26), e15917 DOI: 10.1097/MD.0000000000015917

9. Pandey, N., & John, S. Kayser-Fleischer Ring. In: StatPearls [Internet]. Treasure Island (FL): *StatPearls Publishing*; 2023 Jan. Available from: https://www.ncbi.nlm.nih.gov/books/NBK459187/
10. Rodriguez-Castro, K. I., Hevia-Urrutia, F. J., & Sturniolo, G. C. (2015). Wilson's disease: A review of what we have learned. *World J Hepatol, 7*(29), 2859-70. doi: 10.4254/wjh.v7.i29.2859. PMID: 26692151; PMCID: PMC4678372.
11. Mantas, A. M., Wells, J., & Trotter, J. (2013). Kayser-Fleischer rings of acute Wilson's disease. *Proc (Bayl Univ Med Cent), 26*(2), 166-7. doi: 10.1080/08998280.2013.11928948. PMID: 23543978; PMCID: PMC3603737.
12. Liu, M., Cohen, E. J., Brewer, G. J., & Laibson, P. R. (2002). Kayser-Fleischer ring as the presenting sign of Wilson disease. *American Journal of Ophthalmology, 133*(6), 832-834. DOI: https://doi.org/10.1016/S0002-9394(02)01408-3