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CASE IMAGE

Kayser–Fleischer rings: The pathognomonic for Wilson's disease

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Key Clinical Message

Wilson's disease is a genetic disorder of copper metabolism that primarily manifests with hepatic and neurological features. Kayser–Fleischer rings (KF rings) are pathognomonic of Wilson's disease and helps in establishing its diagnosis.

KEYWORDS

copper metabolism, hepatolenticular degeneration, KF rings, Wilson's disease

1 | INTRODUCTION

Wilson's disease is an autosomal recessive disorder of copper metabolism that results in pathological accumulation of copper in various organs and thus has multisystem presentation.

2 | CASE HISTORY AND EXAMINATION

A 25-year-old lady presented to our hospital with a 2-month history of headache and involuntary limb movements. She reported having mild abdominal discomfort that had been on and off for a few months. Physical examination revealed icterus, bilateral pedal edema, splenomegaly, and gait ataxia. Additionally, choreoathetosis, dysarthria, and cognitive impairment were also appreciated and her mother reported behavioral changes. Family history was non-revealing. Greenish brown pigmented rings were seen in the eyes: the Kayser–Fleischer rings (KF rings; Figure 1, arrow), which were further magnified and visualized clearly on indirect ophthalmoscopic examination using 20D lens (right eye). Slit-lamp examination revealed copper deposition in the corneal Descemet's membrane bilaterally.

FIGURE 1 Kayser–Fleischer rings in Wilson disease, grossly visible (arrow) and magnified in the right eye by 20D lens.

3 | METHODS

Laboratory studies showed normal liver and kidney function tests. Abdominal ultrasonography revealed coarse and increased echotexture of liver. Serum ceruloplasmin level was 0.089 g/L (reference range, 0.2–0.6), serum copper level was 50.1 μg/dL (reference range, 80–155), and the 24-h urinary copper excretion was 320 μg (reference range, 10–30).

4 | DISCUSSION AND CONCLUSION

A diagnosis of Wilson disease was made and treatment with copper chelating agent D-penicillamine and zinc was started. At 1-year follow-up, the patient is doing well with improvement in her clinical and laboratory parameters and is on maintenance therapy.

KF rings are seen in 50%–60% of hepatic and 95%–100% cases of neurological Wilson disease.[1] Through pathognomonic for Wilson's, these can also be seen in patients with chronic cholestatic diseases including primary biliary cirrhosis and neonatal cholestasis. These differentials should be considered once Wilson's disease is ruled out or upon strong clinical suspicion of other diagnoses.

AUTHOR CONTRIBUTIONS

Priyanka Singh: Conceptualization; data curation; investigation; writing – original draft. **Bachaspati Subedi:** Data curation; investigation; methodology; supervision; writing – review and editing. **Devraj Mahato:** Conceptualization; data curation; supervision; writing – review and editing. **Mitesh Karn:** Conceptualization; data curation; methodology; writing – original draft; writing – review and editing.

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None.

CONFLICT OF INTEREST STATEMENT

The authors have no conflict of interest to declare.

DATA AVAILABILITY STATEMENT

The datasets used and/or analyzed during the current study are available from the corresponding author upon reasonable request.

CONSENT

Written informed consent was obtained from the patient for publication of this report and any accompanying images.

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