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CASE REPORT

WILSON'S DISEASE PRESENTING WITH NEUROPSYCHIATRIC SYMPTOMS AND KAYSER–FLEISCHER RING COMPLICATED BY URETERIC CALCULUS AND UTI

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ABSTRACT

Background: Wilson's disease is an autosomal recessive disorder of copper metabolism leading to toxic accumulation in multiple organs, most prominently the liver, brain, and cornea. While neuropsychiatric manifestations and Kayser–Fleischer (KF) rings are well-established diagnostic hallmarks, renal complications such as nephrolithiasis and secondary urinary tract infection (UTI) are less frequently recognised, despite their potential to significantly alter clinical outcomes.

Case Presentation: We report the case of a 25-year-old male with known Wilson's disease who presented with progressive memory impairment, behavioural dullness, tremors, and bradykinesia over several months. Slit-lamp examination revealed a distinct KF ring, indicating ongoing systemic copper deposition, corroborated by markedly reduced ceruloplasmin levels. Following a transient neurological exacerbation, the patient later developed acute flank pain, low-grade fever, and anorexia. Ultrasound imaging identified a lower calyceal renal calculus and a distal ureteric calculus causing mild hydroureteronephrosis. Urinalysis demonstrated early features of UTI. He was managed conservatively with intravenous antibiotics, hydration, analgesics, and continuation of chelation and dopaminergic therapy, resulting in rapid symptom resolution and stable renal and neurological status.

Conclusion: This case highlights the multisystemic nature of Wilson's disease, emphasising the need for vigilance regarding renal complications, particularly when new-onset pain or fever emerges in patients presenting primarily with neurological involvement.

Keywords: Chelation Therapy; Kayser–Fleischer Ring; Neuropsychiatric Manifestations; Ureteral Calculi; Urinary Tract Infections; Wilson Disease.

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INTRODUCTION

Wilson's disease (WD) is an autosomal recessive disorder of copper transport caused by pathogenic mutations in the ATP7B gene, resulting in defective biliary copper excretion and toxic accumulation of free copper in hepatic and extrahepatic tissues.[1] Progressive copper deposition leads to oxidative injury, mitochondrial dysfunction, and cellular degeneration across multiple organ systems. The estimated global prevalence of WD is approximately 1 in 30,000 individuals, although genetic screening suggests a higher prevalence of heterozygous carriers and potentially underdiagnosed cases.[2] Clinically, WD presents along a broad spectrum, commonly categorised into hepatic, neuropsychiatric, or mixed forms. Neuropsychiatric manifestations arise from copper accumulation in the basal ganglia, thalami, cerebellum, and cortical regions and frequently include tremors, dystonia, Parkinsonism, cognitive impairment, personality changes, and behavioural disturbances.[3]

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A distinctive and diagnostically significant feature of neuropsychiatric WD is the Kayser–Fleischer (KF) ring, caused by copper deposition in Descemet's membrane of the cornea. KF rings are present in up to 95% of patients with neurological involvement and strongly support the diagnosis when correlated with reduced ceruloplasmin levels and elevated non-ceruloplasmin-bound copper.[4]

Renal involvement in WD is less frequently highlighted but clinically relevant. Copper toxicity can impair proximal tubular function, resulting in aminoaciduria, phosphaturia, distal renal tubular acidosis, nephrolithiasis, and, in some cases, nephrocalcinosis. The mechanism of renal stone formation in WD has been attributed to tubular dysfunction, hypercalciuria, hypocitraturia, and metabolic changes associated with chelators such as penicillamine.[5] Though renal calculi have been documented in WD cohorts, symptomatic ureteric obstruction and secondary urinary tract infection remain relatively uncommon, and their coexistence with active neuropsychiatric deterioration is even more rarely reported. The case we are reporting here is distinctive because it demonstrates a multisystemic trajectory of Wilson's disease: neuropsychiatric decline with a clearly visible KF ring, corroborated biochemical abnormalities, and a concurrent renal complication, namely a distal ureteric calculus causing mild hydroureteronephrosis and superimposed urinary tract infection. The convergence of these manifestations underscores the systemic burden of copper dysregulation and reinforces the need for comprehensive, multidisciplinary evaluation in WD patients presenting with neurological symptoms.

CASE DETAILS

A 25-year-old male patient with a known diagnosis of Wilson's disease presented initially in mid-August 2025 with progressive memory disturbances and subtle neuropsychiatric changes that had been insidiously worsening over several months. His family described increasing forgetfulness, intermittent behavioural dullness, and newly developed tremulousness affecting his daily functioning. On admission, he was thin-built (47 kg), conscious, and oriented, with stable vitals. Neurological examination showed mild symmetric tremors and bradykinesia. During the initial evaluation, a slit-lamp examination was performed due to his background of Wilson's disease, which revealed a faint but clearly visible **Kayser–Fleischer (KF) ring** (as shown in Figure 1), presenting as a copper-brown deposition circumferentially along the corneal periphery. This ocular finding reinforced systemic copper deposition and supported the biochemical evidence of impaired ceruloplasmin synthesis (ceruloplasmin 0.06 g/L) and low-normal serum copper levels. These combined clinical and laboratory features reaffirmed the underlying diagnosis and guided continuation of chelation therapy alongside dopaminergic agents introduced for his extrapyramidal symptoms.

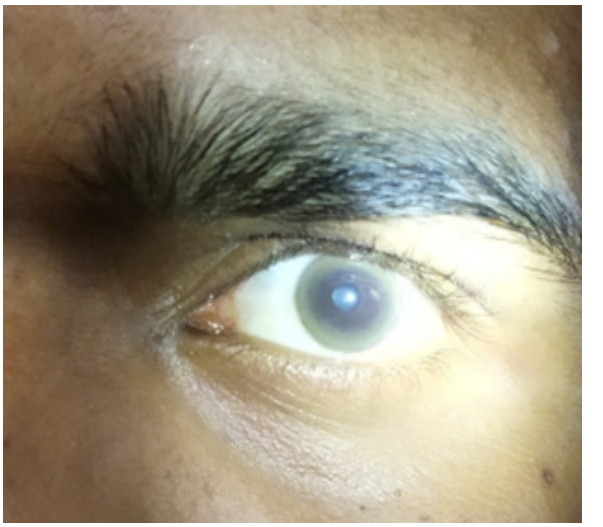

Figure 1 shows a Slit-lamp photograph of the patient's eye showing a circumferential Kayser–Fleischer ring characterised by copper-brown deposition along the peripheral cornea.

Over the subsequent weeks, he experienced a brief neurological exacerbation characterised by increased tremors, rigidity, and fatigue, accompanied by intermittent low-grade fever. A short course of prednisolone stabilised the episode, and he resumed regular activities. Toward the end of September, however, he developed new-onset flank pain that gradually intensified over several days, radiating to the left lower back and accompanied by chills and anorexia. An ultrasound abdomen revealed bilateral Grade-I renal parenchymal changes, a 4.5-mm left lower calyceal calculus, and a 3.5-mm distal ureteric calculus near the vesico-ureteric junction, causing mild hydroureteronephrosis. These findings indicated obstructive uropathy superimposed on metabolic vulnerability associated with Wilson's disease.

He was admitted on 23 October 2025 with suspected UTI secondary to ureteric obstruction. Examination showed right iliac fossa and left hypochondrial tenderness. Urinalysis showed haziness, trace proteins, microscopic hematuria, and later rising pus cells. He was managed medically with intravenous cefoperazone–sulbactam, hydration, parenteral pantoprazole, vitamin supplementation, and antispasmodics. His renal function remained stable throughout hospitalisation, urine output was well-maintained, and there were no signs of systemic sepsis. Within 48 hours, the flank pain resolved, the fever subsided, and the urinary parameters improved steadily. The neurological status remained stable, with no deterioration in tremors or rigidity during this intercurrent illness.

By 25 October, the patient was asymptomatic, tolerating an oral diet, and free from flank pain. The distal

ureteric calculus was expected to pass spontaneously, supported by progressive symptom resolution. He was discharged in stable condition with advice for hydration, continuation of chelation therapy, dopaminergic medications, and scheduled follow-up for renal imaging and neurologic review. Throughout this clinical course, spanning neuropsychiatric deterioration, ophthalmic confirmation of copper deposition, a neurological flare, and later obstructive uropathy with early UTI, timely multidisciplinary intervention ensured a smooth recovery without residual deficits.

DISCUSSION

The patient's neuropsychiatric symptoms, memory disturbances, tremors, bradykinesia, and behavioural dullness align with well-documented neurological presentations of WD caused by copper deposition in the basal ganglia, thalami, and cortical networks.[6] The presence of a KF ring further reinforces the diagnosis, especially in neuropsychiatric disease, where its prevalence reaches 90–95%.[7] In this case, slit-lamp examination played a pivotal role in confirming active systemic copper accumulation and guiding continued chelation therapy.

Renal involvement in WD, however, is frequently under-recognised. Copper-induced proximal tubular dysfunction can lead to hypercalciuria, phosphaturia, aminoaciduria, metabolic acidosis, and nephrolithiasis.[8] A review by Dzieżyc-Jaworska K et al., in 2019, suggests that nephrolithiasis may occur in up to 16–20% of patients with WD, although many remain asymptomatic or undiagnosed.[9] The coexistence of a distal ureteric calculus with hydroureteronephrosis and subsequent UTI, as seen in this patient, is less common and highlights the renal vulnerability imposed both by copper toxicity and metabolic alterations associated with therapy.

Previous reports have also highlighted that renal manifestations in Wilson's disease, though less frequent than hepatic or neuropsychiatric symptoms, may significantly affect morbidity. In a cohort analysis by Zi-Wei Zheng et al. in 2025, [10] nephrolithiasis occurred in approximately 12% of WD patients, with higher rates in those receiving long-term chelation therapy, reinforcing the need for periodic renal surveillance. Consistent with the findings of Maríño et al. in 2023, [11] who showed that neuropsychiatric impairment and systemic complications markedly reduce quality of life in Wilson's disease, our patient's simultaneous neurological deterioration and renal obstruction similarly reflect the substantial multidimensional QoL burden associated with multisystem involvement.

CONCLUSION

This case underscores the complex, multisystemic nature of Wilson's disease, illustrating how concurrent neuropsychiatric deterioration, ophthalmologic stigmata, and renal calculous disease can intersect to produce significant clinical morbidity. The coexistence of a Kayser–Fleischer ring, progressive neurological manifestations, and a symptomatic ureteric calculus complicated by urinary infection highlights the need for a vigilant, multidisciplinary approach to diagnosis and management. Early recognition of renal complications, even when neurologic symptoms dominate the clinical picture, is essential to prevent adverse sequelae such as obstructive nephropathy or sepsis. This report reinforces the importance of comprehensive systemic monitoring, sustained chelation therapy adherence, and coordinated specialist involvement to optimise outcomes and preserve quality of life in individuals living with Wilson's disease.

Conflict of Interest: The authors declare that there are no conflicts of interest related to the publication of this case report.

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Ethical Approval and Consent: This case report was prepared in accordance with institutional ethical standards and the Declaration of Helsinki. The patient provided written informed consent for the publication of anonymised clinical details and relevant imaging findings.

REFERENCES

1. Kaler SG. ATP7A-related copper transport diseases—emerging concepts and future trends. Nature Reviews Neurology [Internet]. 2011 Jan 1;7(1):15–29. Available from: https://www.nature.com/articles/nrneurol.2010.180
2. Członkowska A, Litwin T, Dusek P, Ferenci P, Lutsenko S, Medici V, et al. Wilson disease. Nature Reviews Disease Primers [Internet]. 2018 Sep 6;4(1):21. Available from: https://www.nature.com/articles/s41572-018-0018-3
3. Litwin T, Dzieżyc K, Członkowska A. Wilson disease—treatment perspectives. Annals of Translational Medicine [Internet]. 2019 Apr 1;7(S2):S68. Available from: https://atm.amegroups.org/article/view/23192/23690
4. Ala A, Walker AP, Ashkan K, Dooley JS, Schilsky ML. Wilson's disease. The Lancet [Internet]. 2007 Feb 1;369(9559):397–408. Available from: https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(07)60196-2
5. Kumar N, Prashant L, Goyal V. Wilson's Disease update: An Indian perspective. [Internet]. Document - Gale Academic OneFile. 2021. Available from: https://go.gale.com/ps/i.do?id=GALE%7CA686081563&sid=-googleScholar&v=2.1&it=r&linkaccess=abs&issn=09722327&p=AONE&sw=w&userGroupName=anon%7E28fcc7f8&aty=open-web-entry

6. Bandmann O, Weiss KH, Kaler SG. Wilson's disease and other neurological copper disorders. The Lancet Neurology [Internet]. 2014 Dec 8;14(1):103–13. Available from: https://www.thelancet.com/journals/laneur/article/PIIS1474-4422(14)70190-5
7. Merle U, Schaefer M, Ferenci P, Stremmel W. Clinical presentation, diagnosis and long-term outcome of Wilson's disease: a cohort study. Gut [Internet]. 2006 May 19;56(1):115–20. Available from: https://gut.bmj.com/content/56/1/115
8. Roberts EA, Schilsky ML. A practice guideline on Wilson Disease. Hepatology [Internet]. 2003 May 28;37(6):1475–92. Available from: https://journals.lww.com/hep/citation/2003/06000/a_practice_guideline_on_wilson_disease.35.aspx
9. Dzieżyc-Jaworska K, Litwin T, Członkowska A. Clinical manifestations of Wilson disease in organs other than the liver and brain. Annals of Translational Medicine [Internet]. 2019 Apr 1;7(S2):S62. Available from: https://atm.amegroups.org/article/view/24690/html
10. Zheng ZW, Xu MH, Fan LN, Wang RM, Xu WQ, Yang GM, et al. Renal impairment in Wilson's disease. Kidney International Reports [Internet]. 2025 Apr 22;10(7):2453–6. Available from: https://www.kireports.org/article/S2468-0249(25)00254-2/fulltext
11. Mariño Z, Berenguer M, Peña-Quintana L, Olveira A, Miralpeix A, Sastre I, et al. Health-Related Quality of Life in Patients Living with Wilson Disease in Spain: A Cross-Sectional Observational Study. Journal of Clinical Medicine [Internet]. 2023 Jul 21;12(14):4823. Available from: https://www.mdpi.com/2077-0383/12/14/4823

CONFLICT OF INTEREST
Authors declare no conflict of interest.
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AUTHORS' CONTRIBUTION

The following authors have made substantial contributions to the manuscript as under:

Conception or Design:	HRG, PKY
Acquisition, Analysis or Interpretation of Data:	HRG, PKY, VKN
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All the authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.