Case Report

Wilson disease masquerading as a primary movement disorder: When an unexplained tremor demands a metabolic search

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ABSTRACT

Wilson disease is an important but often overlooked cause of progressive movement disorders in young adults. A 23-year-old woman presented with progressive involuntary shaking movements involving all four limbs, with dysarthria and dystonic features. She had a history of seven consecutive first-trimester abortions. Routine liver, thyroid, renal, and hematological investigations were normal, while evaluation for antiphospholipid disease did not reveal an alternative explanation. Slit-lamp examination showed bilateral Kayser–Fleischer rings; serum ceruloplasmin was 8 mg/dL, serum copper 56 µg/L, and 24-h urinary copper 110 µg/24 h. Magnetic resonance imaging showed bilateral thalamic and dorsal midbrain T2 hyperintensities. The documented Leipzig score was 7, supporting the diagnosis of Wilson disease. She was treated with D-penicillamine and pyridoxine. This case highlights that Wilson disease should be considered in young patients with progressive unexplained movement disorders even in the absence of overt hepatic disease.

Key words: Ceruloplasmin, Dystonia, Kayser–Fleischer ring, Movement disorder, Tremor, Wilson disease

Wilson disease is an autosomal recessive inborn error of copper metabolism caused by mutations in the *ATP7B* gene, with a highly variable clinical phenotype. Although hepatic involvement is common, neurological manifestations may dominate the presentation and can occur without clinically obvious liver disease. Wilson disease can manifest as tremor, dystonia, dysarthria, ataxia, chorea, or a Parkinsonism-like syndrome. These phenotypes can closely resemble primary movement disorders and may delay recognition of the underlying metabolic disease. The diagnostic challenge is particularly important in young adults. When a progressive movement disorder remains unexplained despite appropriate neurological evaluation, potentially reversible metabolic disorders should be reconsidered [1,2].

We report a young woman whose presentation strongly suggested a movement disorder but whose eventual evaluation revealed Wilson disease. The diagnostic journey, rather than the mere presence of neurological Wilson disease, is the central feature of this case.

CASE REPORT

A 23-year-old woman presented with progressively worsening involuntary shaking movements. The illness had initially begun with abnormal shaking involving the upper limbs, but over time the movements progressed to involve all four limbs. The progressive nature of the symptoms was accompanied by speech difficulty and slurring of speech, resulting in increasing functional impairment. Her obstetric history was significant for seven consecutive spontaneous abortions, all occurring before 10 weeks of gestation. There was no history of examination of the fetal products. She had no history of unprovoked arterial or venous thrombosis.

At presentation, the clinical picture was predominantly neurological. Neurological examination revealed normal higher mental functions (except for dysarthric speech with difficulty in articulation). A prominent asymmetric action tremor, particularly a wing-beating type tremor, involving all four limbs but more pronounced in the left upper limb was noted. Dystonic posturing and increased muscle tone were also noted. Facial examination revealed increased rigidity, particularly involving the muscles around the mouth and jaw. Passive movement of the limbs demonstrated increased muscle tone. Power in all four limbs was normal. Cerebellar examination revealed ataxia with impaired coordination. There were no psychiatric manifestations. There were no clinically apparent signs of hepatic decompensation; there was no jaundice, ascites, hepatomegaly, or splenomegaly.

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The patient's age, progressive involuntary movements, tremor, dystonic component, and dysarthria initially directed the diagnostic approach toward a movement disorder spectrum. The main differential diagnoses included early-onset Parkinsonism, essential tremor, dystonic tremor or primary dystonia, and other secondary movement-disorder syndromes. In view of the recurrent pregnancy loss, antiphospholipid syndrome and autoimmune disease were also evaluated.

Laboratory investigations are listed in Table 1. Thus, despite the striking neurological phenotype, the work-up did not provide a satisfactory diagnosis. Rather than labeling the patient as having an unexplained primary movement disorder, the possibility of a treatable inherited metabolic disorder was reconsidered. Because of her young age, progressive neurological manifestations, recurrent pregnancy loss with no evidence of antiphospholipid syndrome, and the absence of a convincing alternative diagnosis, Wilson disease was specifically evaluated. Slit-lamp examination revealed bilateral Kayser–Fleischer rings. The combination of a young patient with a progressive movement disorder, Kayser–Fleischer rings, markedly reduced ceruloplasmin, increased urinary copper excretion, and supportive magnetic resonance imaging (MRI) findings indicated the possibility of Wilson disease with predominant neurological presentation (Table 2). Liver biopsy and genetic mutation analysis could not be performed. A Leipzig score of ≥4 is diagnostic; the patient's score of 7 supported the diagnosis of Wilson disease (Table 3).

The patient was treated with D-penicillamine 250 mg 4 times daily to increase copper excretion, along with pyridoxine supplementation to prevent pyridoxine deficiency associated with D-penicillamine therapy.

DISCUSSION

The phenotype in this patient is comparable with previously reported neurological presentations. Elendu *et al*. described a young adult with prominent tremor, dystonia, and dysarthria, emphasizing that neurological Wilson disease may initially be approached as a movement disorder [3]. Parkinsonism, dystonia, tremor, and cerebellar manifestations have also been described in neurological Wilson disease, supporting the mixed extrapyramidal and cerebellar phenotype observed in our patient [1,2]. Thus, tremor, dystonia, dysarthria, and ataxia in a young adult with an unexplained progressive movement disorder should prompt consideration of Wilson disease.

The MRI findings in our patient-bilateral thalamic and dorsal midbrain T2 hyperintensities were within the recognized neuroimaging spectrum of neurological Wilson disease. In a cohort of 364 patients with neurological Wilson disease, abnormal MRI signals involved the putamen most frequently, followed by the pons, thalamus, and midbrain [4]. Reviews similarly describe T2/Fluid-attenuated inversion recovery abnormalities of the basal ganglia, thalamus, midbrain, and pons, while the "face of the giant panda" sign occurs only in a subset of patients [5]. Therefore, absence of a classic MRI sign does not exclude the diagnosis.

The history of seven consecutive first-trimester abortions is notable, as pregnancy loss has been reported in women with untreated Wilson disease [6]. Copper toxicity and placental copper accumulation may contribute to adverse pregnancy outcomes, but causality cannot be established, and an independent obstetric cause cannot be excluded because further evaluation was not performed.

Table 1: Laboratory investigations of the patient

Parameters	Investigation	Patient value/ finding
Laboratory investigations	Hemoglobin	11.1 g/dL
	Alanine aminotransferase (ALT)	32 U/L
	Aspartate aminotransferase (AST)	28 U/L
	Urea	20 mg/dL
	Creatinine	0.8 mg/dL
	Lupus anticoagulant	Negative
	β2-glycoprotein I antibody	12 U/mL (negative, <20)
	Anticardiolipin antibody	8 U/mL (negative, <20)
	Antinuclear Antibody (ANA) by HEp-2	Low titre, 1:20

Table 2: Imaging findings and Wilson disease-specific investigations of the patient

Radiological findings	MRI brain	Bilateral thalamic and dorsal midbrain T2 hyperintensities
	Ultrasonography of abdomen	Grade 1 fatty liver
	FibroScan	5 kPa — no significant fibrosis
Wilson disease-specific investigations	Serum ceruloplasmin	8 mg/dL
	Serum copper	56 µg/L
	24-hour urinary copper	110 µg/24 h

Table 3: Leipzig score assessment

Component	Patient finding	Points
Kayser–Fleischer rings	Present	2
Neurological symptoms	Severe/progressive tremor, dystonia, and dysarthria	2
Serum ceruloplasmin	8 mg/dL (<10 mg/dL)	2
24-hour urinary copper	110 µg/24 h; 1–2×ULN (laboratory ULN=100 µg/24 h)	1
Coombs-negative hemolytic anemia	Negative	0
Hepatic copper/ ATP7B mutation	Not performed	0
Total	2+2 + 2+1	7

Treatment was initiated with D-penicillamine and pyridoxine supplementation to prevent pyridoxine deficiency associated with D-penicillamine therapy. In neurological Wilson disease, chelation should be introduced cautiously because paradoxical neurological worsening can occur; clinically significant worsening may require dose reduction or consideration of trientine. Tetrathiomolybdate is an emerging copper-lowering option where clinically available. In this patient, zinc was planned as maintenance therapy after initial decoppering and clinical stabilization [6]. Zinc induces metallothionein in intestinal enterocytes, which preferentially binds dietary copper and prevents its absorption; the copper-bound enterocytes are subsequently shed into the gastrointestinal lumen, increasing fecal copper excretion. Zinc therefore maintains a negative copper balance and helps prevent reaccumulation. Zinc is also an appropriate therapeutic option in asymptomatic/presymptomatic Wilson disease, particularly in patients without significant hepatic disease, and may be used during pregnancy and breastfeeding [6]. Liver transplantation remains an option for acute liver failure or decompensated liver disease, and may also be considered in severe neurological disease [7].

CONCLUSION

Wilson disease can hide in plain sight as a movement disorder. In a young patient with progressive tremor, dystonia, and dysarthria, the absence of jaundice or abnormal routine liver tests should not prematurely exclude Wilson disease. The key diagnostic step is to reconsider a metabolic cause when common movement disorder differentials do not adequately explain the phenotype.

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REFERENCES

1. Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J, editors. Harrison's Principles of Internal Medicine. 22nd ed., Vol. 2. New York: McGraw Hill; 2025.
2. Dooley JS, Lok AS, Garcia-Tsao G, Pinzani M, editors. Sherlock's Diseases of the Liver and Biliary System. 13th ed. Hoboken, NJ: Wiley-Blackwell; 2018.
3. Elendu C, Amaechi DC, Jingwa KA, Elendu TC. A rare presentation of Wilson disease with neurological symptoms: Case report and genetic analysis. Medicine (Baltimore) 2024;103:e37099.
4. Yu XE, Gao S, Yang RM, Han YZ. MR imaging of the brain in neurologic Wilson disease. AJNR Am J Neuroradiol 2019;40:178-83.
5. Rędzia-Ogrodnik B. Brain magnetic resonance imaging in Wilson's disease-significance and practical aspects-a narrative review. J Clin Med 2024;13:4081.
6. Socha P, Jańczyk W, Zanetto A, Burra P, Czlonkowska A, Debray D, *et al*. EASL-ERN clinical practice guidelines on Wilson's disease. J Hepatol 2025;82:690-728.
7. Litwin T, Bembenek J, Antos A, Kurkowska-Jastrzębska I, Przybyłkowski A, Skowrońska M, *et al*. The maternal and fetal outcomes of pregnancy in Wilson's disease: A systematic literature review and meta-analysis. Biomedicines 2022;10:2072.

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