Case Report

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Paediatrics Section

# Wilson Disease Presenting as Isolated Dysarthria in a Paediatric Patient: A Rare Case Report

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## ABSTRACT

Wilson disease is an autosomal recessive disorder caused by mutations in the ATP7B gene. It involves the liver, brain, eyes, blood, kidneys, and endocrine glands. The usual manifestations are hepatic (approximately 40%), neurological (approximately 35%), or psychiatric (approximately 10%). Wilson disease without hepatic manifestations is rare. A nine-year-old girl presented to the paediatric outpatient department with two months of difficulty speaking and with occasional purposeless jerky movements for one and a half months. She had tremor while speaking for the past month. The rest of the general and systemic examination was normal. Neurological complaints in an adolescent girl raise suspicion of Wilson disease. Slit-lamp examination showed a Kayser–Fleischer ring. Serum ceruloplasmin was low, and Magnetic Resonance Imaging (MRI) brain showed basal ganglia involvement, which confirms the diagnosis of Wilson disease. The child was managed with copper chelating therapy (D-penicillamine and zinc acetate) and a low-copper-containing diet with supportive care. This case highlights that isolated dysarthria can be a presenting feature of Wilson disease, emphasising the importance of considering this diagnosis in patients with unexplained speech difficulties.

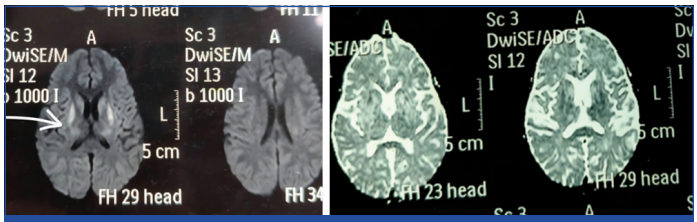
**Keywords:** Autosomal recessive, Ceruloplasmin levels, d-Penicillamine, Hepatolenticular degeneration, Zinc acetate

## CASE REPORT

A nine-year-old girl presented to our paediatric Outpatient Department (OPD) with chief complaints of difficulty in speaking for two months. Parents also noted occasional purposeless jerky movements since one and a half months. The jerky movements occurred randomly. Her speech was slurred and slow, with a low pitch, incomprehensible words, appearing to whisper, nasal twang, and vocal fatigue. The child was not able to open her mouth properly; two-finger mouth opening was present. There was no history of weakness of the upper or lower limbs or deviation of the mouth. There was no history of yellowish discolouration of the body or abdominal distension. There was no history of hospitalisation. There was no family history of any chronic illness. The child was developmentally normal and fully immunised to date. She was born of a non-consanguineous marriage and belonged to lower socioeconomic status per the modified Kuppuswamy classification [1].

On examination, the child was conscious and oriented to time, place, and person. She was afebrile and had pallor. There was no icterus, cyanosis, clubbing, or lymphadenopathy. Vitals were: heart rate 94/min, respiratory rate 18/min, $SpO_2$ 99% on room air, and blood pressure 105/70 mmHg in the left arm. Anthropometry showed weight and height between the 10$^{th}$ and 25$^{th}$ percentiles. Central Nervous System (CNS) examination showed normal higher function with no abnormalities in the cranial nerves. Motor examination showed normal tone and 5/5 power in all limbs with normal deep tendon reflexes. Sensory system, cerebellar signs, and skull and spine examinations were normal. The child had tremor while speaking. MRI brain showed T2/FLAIR hyperintensities involving bilateral basal ganglia and posterolateral thalamus [Table/Fig-1]. Kayser-Fleischer ring was seen on slit-lamp examination and serum ceruloplasmin levels were low (<9.7 mg/dL) [Table/Fig-2].

Differential diagnoses considered were conversion disorder leading to speech issues, neurological illness causing dysarthria, Wilson disease, and vitamin B12 deficiency. However, normal serum B12 levels [Table/Fig-3], normal CNS examination, and the absence of other CNS manifestations ruled out the other possibilities. A final diagnosis of Wilson disease with isolated dysarthria was made.




**[Table/Fig-1]:** MRI brain- T2 FLAIR hyperintensities are seen involving bilateral basal ganglia and posterolateral thalamus.

**[Table/Fig-2]:** Kayser-Fleischer (KF) ring seen on slit lamp examination.

| Investigation | Results | Reference range |
|---|---|---|
| Haemoglobin | 12.6 gm/dL | 12-14 gm/dL |
| White blood cells | 9300/cumm | 4000-11000/cumm |
| Neutrophil/ Lymphocyte | 44%/ 47% | |
| Platelet count | 2.5 lacs | 1.5- 4.5 lacs |
| Urea | 29/0.6 mg/dL | 10-50 mg/dL |
| Creatinine | 0.6 mg/dL | 0.5-1 mg/dL |
| Erythrocyte Sedimentation Rate (ESR) | 08 mm/hour | Less than 10 mm/hour |
| Serum Glutamic-Oxaloacetic Transaminase (SGOT)/Serum Glutamate Pyruvate Transaminase (SGPT) | 47/27 mg/dL | Upto 40 U/L |
| Total bilirubin/direct bilirubin | 0.7/0.2 mg/dL | TB- 0.1-1.2 mg/dL<br>DB- less than 0.3 mg/dL |
| Serum albumin | 3.4 gm/dL | 3.5-5 gm/dL |
| Prothrombin Time (PT)/International Normalised Ratio (INR) | 14.6/0.9 | |
| B12 Levels | 312 | 200-900 pg/mL |
| Ultrasonography (USG) abdomen | Normal study | |
| MRI brain | T2 Flair hyperintensities are seen involving bilateral basal ganglia and posterolateral thalamus. | |
| Slit lamp examination | Kayser Fleischer (KF) ring present | |
| Serum ceruloplasmin | <9.7 mg/dL | <20 is significant |
| 24 hours urine copper | 68 microgram/day | 15-70 mcg/24 hour |

**[Table/Fig-3]:** The table shows investigations done at various OPD visits.

After confirmation of the diagnosis, the patient was started on D-penicillamine 20 mg/kg/day in two divided doses and zinc acetate 25 mg three times daily, with a low-copper diet and supportive care. Pyridoxine supplementation was given. The patient showed significant improvement in speech fluency after two weeks of treatment. The patient was discharged on D-penicillamine 250 mg twice daily and zinc acetate 25 mg three times daily, with calcium supplementation. The patient remains on monthly follow-up in the outpatient department, and monitoring for drug side effects is performed. Complete blood count and Liver Function Tests (LFTs)/ Kidney Function Tests (KFTs) were within the normal range. Fluency and pitch of voice improved. Speech became comprehensible, and purposeless jerky movements disappeared after 3-4 months of treatment. Parental consent was obtained for publication, as the patient is a minor.

| S. No. | Case report | Place of study | Year | Age/gender | Symptoms | Investigation | Treatment |
|---|---|---|---|---|---|---|---|
| 1. | Verma R et al., [9] | King George Medical University, Lucknow, Uttar Pradesh | 2013 | 12 years/female | Anarthria and dystonia for one month | Kayser-Fleischer (KF) ring on slit lamp microscopy, MRI brain, serum ceruloplasmin levels, 24 hour urinary copper | Copper chelating agent and Antichoinergic |
| 2. | El Qadiry R et al., [10] (Case Series) | University Hospital Mohammed VI, Marrakech, Morocco | 2022 | 12 years/male; 11 years/female | First case-Dysarthria, hyper salivation and tremor of upper limb Second case- Gait abnormalities, dysarthria and dystonia | MRI brain, serum ceruloplasmin levels, 24 hour urinary copper, KF ring on slit lamp microscopy | D-Penicillamine, Zinc acetate |
| 3. | Present case report | PGIMS, Rohtak, Haryana | 2024 | 9 years/female | Difficulty in speaking | MRI brain, serum ceruloplasmin levels, 24 hour urinary copper, KF ring on Slit lamp microscopy | D-Penicillamine, Zinc acetate, low copper containing diet |

**[Table/Fig-4]:** Comparative analysis of similar cases [9,10].

## DISCUSSION

Wilson disease, or hepatolenticular degeneration, is a copper deposition disease. The gene involved is ATP7B on chromosome 13q14 [2]. The incidence is 1 in 30,000-50,000 population [3]. It is characterised by excessive copper accumulation in various organs, leading to neurological, hepatic, and psychiatric manifestations. Clinical features range from an asymptomatic state to hepatitis, cirrhosis, fulminant hepatic failure, and neuropsychiatric manifestations [4].

Neurological manifestations are present in 40-50% of cases [5]. CNS presentations include tremor, rigidity, hypokinesia, dystonia, and choreoathetoid movements [6]. The index case presented with dysarthria and purposeless jerky movements. Diagnosis is based on slit-lamp examination (KF ring), low serum ceruloplasmin, elevated 24-hour urinary copper excretion, and ATP7B gene mutation [7]. The diagnosis in the index case was confirmed by MRI of the brain, slit-lamp examination, and low serum ceruloplasmin levels. Management includes dietary copper restriction, D-penicillamine, zinc, trientine, and ammonium tetrathiomolybdate with pyridoxine supplementation [8]. The index case was managed with D-penicillamine 20 mg/kg and zinc acetate with pyridoxine supplementation. The patient's symptoms improved significantly with chelation therapy, underscoring the importance of early diagnosis and treatment. The patient is on monthly follow-up in the outpatient department, and monitoring for drug side effects is performed. Complete Blood Count (CBC) and LFTs/KFTs were within the normal range. Previous similar case reports are described in [Table/Fig-4] [9,10].

## CONCLUSION(S)

Wilson disease is a rare genetic disorder that can present with varied neurological symptoms, including isolated dysarthria, which is a rare and lesser-known initial presentation. Early diagnosis and treatment are crucial to prevent irreversible neurological damage and improve outcomes. This case highlights the importance of considering Wilson disease in the differential diagnosis when patients present with unexplained movement or speech disorders, as prompt treatment can prevent further neurological deterioration and improve outcomes.

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