CASE REPORT

Acute onset anarthria without hepatic manifestation: a rare presentation of Wilson disease

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SUMMARY

Wilson disease (WD) is one of the few inherited but treatable disorder mainly affecting the liver and brain resulting in severe disability or death if left untreated. Hence, it is important to keep a high index of suspicion for diagnosing this clinical entity in appropriate clinical settings. The clinical presentation can be quite variable and they may present solely with neurological features sans hepatic symptoms. Such neurological manifestations usually follow subacute to chronic course. Acute onset anarthria as the heralding and predominant presenting feature has been rarely reported in the literature. We reported a case of a 12-year-old girl who presented with acute onset anarthria and dystonia of 1-month duration. On further evaluation, a diagnosis of WD was made. The patient showed partial improvement after she was started on copper chelating agents and anticholinergics.

BACKGROUND

In 1912, Kinnier Wilson first described Wilson disease (WD) as 'progressive lenticular degeneration'. It is an autosomal recessive disorder resulting from mutations in the ATP7B gene, a membrane-bound, copper-transporting ATPase, mainly affecting the liver and brain. The most common neurological manifestations of WD include movement disorders such as dystonia, incoordination and tremor, emotional liability, dysarthria and seizures which usually follow subacute to chronic course, acute presentations being less common. Acute onset anarthria as the predominant presenting feature has been rarely reported in the literature. With this background, we reported a case of 12-year-old girl who presented with a 1-month history of acute onset anarthria and dystonia. On further evaluation, she was diagnosed as having WD.

CASE PRESENTATION

A 12-year-old girl presented with acute onset inability to speak and abnormal body posturing of 1-month duration. Her parents noticed reduced verbal output. She was unable to speak complete sentences and started communicating with the help of bisyllables, monosyllables and sometimes only gestures. After 3–4 days, she became completely mute. However, she was able to comprehend whatever was being spoken to her. There was no hoarseness or slurring of speech. After 5 days, her parents noticed abnormal twisting movements of the left foot while walking. Within a span of a week, the right foot as well as both the hands developed abnormal posturing. There was no history suggestive of any sensorimotor deficit or abnormal behaviour.

Video 1 The patient demonstrating anarthria.

The patient was born of a non-consanguineous marriage. Perinatal history was uneventful with normal achievement of developmental milestones. Family history was not significant.

On examination, vital parameters including pulse and blood pressure were normal. She was conscious and alert. Anarthria was present (video 1) with preserved comprehension. Ocular examination revealed presence of Kayser-Fleischer (KF) ring visible on naked eye examination, which was confirmed on slit-lamp examination (figure 1). A younger sibling also had KF ring although he was asymptomatic and had normal neurological examination. Hypometric saccades were present. Other cranial nerve examination did not reveal any abnormality. Rigidity was present in all limbs. Power was normal in all limbs. She had a dystonic smile and gaping of the mouth along with dystonic posturing of left foot with extensor posturing. There was no bradykinesia, tremor or any other involuntary

Figure 1 Ocular slit lamp examination showing presence of Kayser-Fleischer ring.

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Figure 2 MRI of the cranium, T2 fluid-attenuated inversion recovery image showing bilateral, symmetrical hyperintensities in basal ganglia and thalamic region.

movement. Deep tendon reflexes were brisk and plantars were extensor bilaterally. Sensory examination and coordination was normal.

INVESTIGATIONS

Routine investigations including a complete blood count, renal and liver function tests, serum electrolytes and chest X-ray were all normal. Slit lamp examination revealed bilateral KF ring. Serum ceruloplasmin was low (0.08 g/L; normal range 0.25–0.46 g/L) and 24 h urinary copper excretion was increased (733.6 µg; normal range 20–60 µg/24 h). The abdominal ultrasound was normal. MRI revealed bilateral and symmetrical hyperintensities in the basal ganglia, thalamus and in the brainstem on T2 and T2 fluid-attenuated inversion recovery (figure 2). Hence, based on these investigations a diagnosis of WD was established.

TREATMENT

The patient was started on D-penicillamine 500 mg/day, zinc acetate 150 mg/day and trihexphenidyl 4 mg/day. She was also advised appropriate dietary copper restriction.

OUTCOME AND FOLLOW-UP

Dystonia and rigidity improved within a week of starting of therapy but anarthria did not show significant improvement even after 4 weeks of therapy.

DISCUSSION

WD usually presents with hepatic, neurological and/or psychiatric manifestations. Commonly, the younger the age of symptom onset, the greater is the degree of liver involvement.¹ The neurological manifestations usually follow subacute to

chronic course, acute presentations albeit rare.² Thus, this case is unusual by virtue of acuteness of neurological presentation and lack of hepatic involvement. The neurological symptoms of WD usually present in the second or third decade.³ However, in India, such patients usually present a decade earlier due to the widespread use of copper utensils for cooking.⁴ The patient presented in the early second decade.

The spectrum of neurological presentation in WD varies widely and may manifest in the form of movement disorders such as dystonia, incoordination, tremors, dysarthria, pseudo-bulbar palsy, seizures, headache and psychiatric manifestations. Dysarthria may sometimes progress to anarthria in untreated and progressive cases. There is a paucity of literature stating anarthria as the presenting feature in WD. We report this case in view of acuteness of onset of anarthria heralding the onset of WD without any preceding dysarthria. The patient developed dystonia after the development of anarthria. As such, anarthria and mutism are a rare occurrence in WD.²

Being a treatable condition, there should be a high index of suspicion for WD in an appropriate clinical setting. This case highlights the importance of suspecting WD in patients who present with recent onset movement disorder and/or extrapyramidal features even in the absence of any clinical or laboratory indicators of hepatic disease. A simple examination for KF ring might give an important clue for further evaluation of WD. Although the absence of KF ring does not rule out WD; fewer than 2% of the cases with predominant neurological manifestations do not have KF ring.⁵ Also, acuteness of neurological presentation, even in the absence of liver disease, should not lead the treating physician from comprehensively excluding the diagnosis of WD, as it is a treatable condition.

Learning points

- ▶ Being a treatable condition, there should be a high index of suspicion for Wilson disease (WD) in appropriate clinical settings.
- ▶ Acute onset of neurological presentation, even in the absence of liver disease, should not prompt treating physician from excluding the diagnosis of WD.
- ▶ Anarthria can be a predominant presenting manifestation of WD.

Contributors RV evolved the hypothesis and AB and NT helped in preparing the manuscript and videos. TSC helped in modifying the manuscript.

Competing interests None.

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