

Case Report

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Wilson's disease: a rare rapidly progressive presentation with atypical imaging finding

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ABSTRACT

Wilson's disease is an autosomal recessive inborn error of copper metabolism characterized by inability of the liver to excrete copper into the bile, with excessive deposition of copper primarily in the liver and in the brain. To report a very rare acute presentation with rapidly progressive disease with some atypical imaging finding. Wilson's disease was diagnosed on the basis of typical clinical picture, laboratory biochemical analysis and typical imaging finding. Apart from typical findings there were some atypical and rare findings which is the purpose of reporting these are a) very rare acute presentation and very rapid progression. b) Atypical Cerebral cortical enhancing lesion in both magnetic resonance imaging and computerized tomography scan with hyperintensity in diffusion weighted imaging in the corresponding areas. Aggressive presentation might be due to rapid accumulation of copper in the brain leading to cytotoxic edema in the accumulated area.

Keywords: Brain, Copper, Liver, MRI, Wilson's disease

INTRODUCTION

Wilson's disease is an autosomal recessive inborn error of copper metabolism characterized by inability of the liver to excrete copper into the bile, with excessive deposition of copper primarily in the liver and in the brain. Its incidence varied from 33 to 68 per 100,000 in India and 1 in 30,000-40,000 in worldwide populations.^{1,2} Till now so many case reports are already present in the literature with different pictures of brain involvement either in the grey matter, white matter or both. These types of pattern of involvement were either related to some classic signs of Wilson's disease like "face of giant panda", "miniature panda" or "trident signs" or some time there were extensive involvement of both grey and white matter without any classical sign of Wilson's disease. Brain involvement in our case has full of common and rare presentation reported in earlier literature but the purpose of reporting is the enhancing

cortical lesion which is rare and rather unreported. Other reason of reporting is the rapidly progressive acute onset presentation and probably explained by imaging finding.

CASE REPORT

A 16 years old girl presented with difficulty in walking and abnormal speech developed within a week. Difficulty in walking was due to frequent freezing and festination of gait. She was hypophonic and dysarthric which was spastic type. Hypomimia, Rigidity and bradykinesia especially of right half of body were quite significant. From the last few days she became apathetic. Her disabilities were rapidly progressive and, in a week, or so she became totally dependent on the family members. Family history was negative, and she never had similar symptoms in the past. There was no history of jaundice in the past. Slit lamp examination confirmed bilateral Kayser-Fleischer ring (KF ring). Routine blood

investigation including liver function test were unremarkable. Serum copper was 31.3mcg/dl (N=8-155 mcg/dl), 24 hrs urinary copper was 330 mcg/day (N=2-80mcg/day), serum ceruloplasmin was 6 mg/dl (N=18-35 mg/dl). Magnetic resonance imaging (MRI) brain was done which showed T1 hypointensity, FLAIR and T2 hyperintensity in dorsal midbrain, pons, bilateral putamen, thalamus, caudate and frontal lobe (Left>Right). DWI also showed hyperintensity in midbrain and bilateral frontal lobes. Significant contrast enhancement was noticed in frontal lobes especially in the left side in both MRI and Computerized tomography (CT) head (Figure 1).

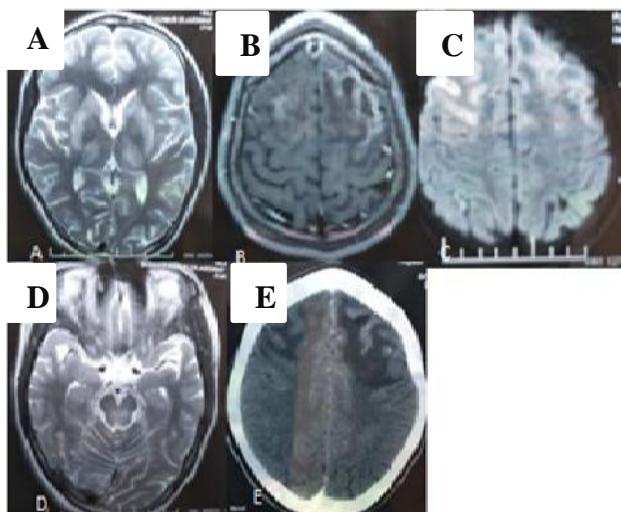


Figure 1: MRI brain (Figure A to Figure D) showing T2 hyperintensity in bilateral putamen, and thalamus. (A): Contrast enhancing asymmetrical lesion in frontal lobe, (B): DWI hyperintensity in bilateral frontal lobe, (C): T2 hyperintense lesion in dorsal mid brain, (D): CECT head, (E): Asymmetrical bilateral contrast enhancing lesion in frontal lobe.

Diagnosis of Wilson's disease was made and oral penicillamine (500mg/day) with zinc acetate (100mg/day) was started. Complete clinical evaluation was done on regular basis and within a week of therapy her clinical status improved remarkably and in 4-6 weeks she became independent for her daily activities. After 6 weeks of therapy 24 hrs urinary copper increased to 567.91 mcg/day. Dose of penicillamine was gradually increased up to 1000mg/day. 6 months after starting therapy she was able to attend her school and even used to help her mother in the household activities. Her urinary copper dropped to 134.6 mcg/day. Dose of penicillamine was gradually tapered to 500 mg/day.

DISCUSSION

Wilson's disease is a rare autosomal recessive disorder of copper metabolism, with a prevalence of about 1 in 30 000 people.³ It is characterized by a decreased biliary copper excretion and a defective incorporation of copper into ceruloplasmin, leading to copper accumulation

especially in the liver, brain, kidney and cornea. It is caused by mutations in the gene encoding P type ATPase (ATP 7B).^{4,5} Wilson's disease may exhibit a variety of clinical symptoms, the most common being liver disease and neuropsychiatric disturbances.⁶⁻⁸ The time lag from the appearance of first symptoms or clinical signs to diagnosis showed a great variation, from 0 to 360 months.⁹ Patients having neurological symptoms present later than having hepatic symptoms after onset of symptoms (44.4 v 14.4 months).⁹

Author's case presented within a week of symptoms onset and progressed very fast. Aggressive presentation might be due to rapid accumulation of copper in the brain leading to cytotoxic edema in the accumulated area. This explanation is supported by findings of brain imaging, like hyperintensities in DWI and contrast enhancement of some of the affected areas.

CONCLUSION

Grey matter and white matter involvement of both cortical and sub cortical area has been reported time to time in the past in Wilson's disease but contrast enhancement of the lesion hasn't been reported earlier unlike author's case.

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