Leukoencephalopathy with Cystic Changes in Neuro-Wilson

Achyuta Perugu, Balamurugan Nagarajan, Chirag Ahuja[1], Arushi G. Saini

Departments of Pediatrics and [1]Radiodiagnosis, Postgraduate Institute of Medical Education and Research, Chandigarh, India

Clinical

An 11-year-old girl presented with progressive gait impairment, frequent falls, and abnormal twisting movements of the limbs for the past 8 months. There was associated cognitive decline, intermittent focal-onset motor seizures, difficulty in swallowing and speech, and drooling of saliva for the past 5 months. On examination, she was in a minimally conscious state, with persistent generalized secondary dystonia, rigidity, spasticity, and brisk deep tendon reflexes. A clinical diagnosis of childhood-onset, progressive neurodegenerative disorder with involvement of basal ganglia, such as Wilson's disease, juvenile-onset Huntington's disease, and neurodegeneration with brain iron accumulation was considered.

Investigations showed low serum copper (8.6 µg/dL, range 80–160 mcg/dl), low serum ceruloplasmin (5.6 mg/dL, range 22–58 mg/dl), elevated 24 h urinary copper (1053 µg/day, range <60 mcg/24 h), and normal liver function test. She had bilateral Kayser-Fleischer rings. Ultrasonogram showed coarse heterogenous echotexture of the liver with nodularity. Magnetic resonance imaging (MRI) of the brain showed bilaterally symmetrical involvement of thalami, basal ganglia, midbrain, and pons, with cystic leukoencephalopathy [Figures 1 and 2]. A final diagnosis of Neuro-Wilson's disease was concluded, and she was initiated on oral zinc, d-penicillamine, and anti-dystonia measures.

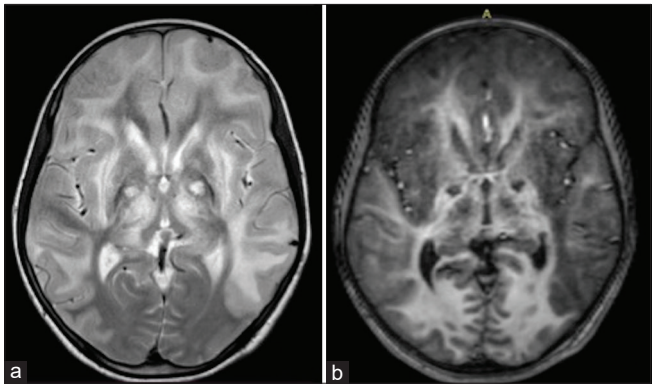

Figure 1: Magnetic resonance imaging of the brain in the patient (a) axial T2-weighted and (b) T1-weighted images show involvement of bilateral basal ganglia, external capsules, thalami, and subcortical white matter

Discussion

Wilson's disease is a distinct neurometabolic disorder of copper metabolism with characteristic involvement of the eye, brain, and liver (hepato-lenticular degeneration). Neurological forms commonly present after 5 years of age with neuropsychiatric problems and movement disorder. The typical neuroimaging features are seen in almost 100% of the cases with neurological dysfunction.[1] These include bilateral striatal involvement, the 'face of giant panda' sign in the midbrain, the 'face of miniature panda' sign in the pons, and the bright claustrum sign.[2,3] Cystic changes are less common, and bilateral cystic leukoencephalopathy mimicking a mitochondrial leukoencephalopathy is an uncommon presentation of Neuro-Wilson's disease. These changes are more commonly seen with rapid neurological deterioration as seen in our case.[4] Clinically, these changes contribute to spasticity and impaired cognition. In children, they have been anecdotally reported with classical basal ganglia changes and indicate poor outcomes despite treatment.[4,5] Our case highlights that Wilson's disease can rarely present with extensive cortical–subcortical lesions, or diffuse leukoencephalopathy, with cystic evolution. Such changes may mimic other disorders such as cystic leukodystrophies and Leigh syndrome. However, such changes indicate a rapidly progressive disease with poor outcomes despite treatment.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information

Address for correspondence: Dr. Arushi G. Saini, Associate Professor, Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh - 160 012, India.
E-mail: doc.arushi@gmail.com

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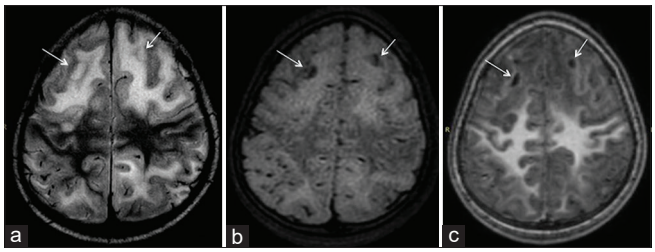

Figure 2: Magnetic resonance imaging of the brain in the patient showing extensive white matter hyperintensities (T2WI and FLAIR) with cystic changes (T1WI) in the frontal lobes (arrows)

to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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