Case 14852

Eurorad ••

White matter lesions in Wilson's

disease

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DOI: 10.1594/EURORAD/CASE.14852 ISSN: 1563-4086 Section: Neuroradiology Area of Interest: Neuroradiology brain Procedure: Diagnostic procedure Imaging Technique: MR Special Focus: Metabolic disorders Case Type: Clinical Cases Authors: Navni Garg, Nimesh Gupta, Deepak Agarwal Patient: 13 years, male

Clinical History:

A 13-year-old boy presented with a history of tremors of bilateral upper limbs and dysarthria progressing over six months to abnormal behaviour and cognitive dysfunction. There was no prior history of liver disease in the child or any family members. Neurological examination revealed abnormal rigidity and posturing with intention tremors affecting the upper limbs.

Imaging Findings:

Laboratory investigations revealed increased urinary copper excretion (280 microgram/day) and low serum copper (8 micromole/litre). Opthalmology review revealed bilateral Kayser Fleischner rings.

Ultrasonography revealed hepatomegaly with coarse liver echotexture.

MRI findings revealed confluent area of T2 / FLAIR hyperintensity involving the subcortical white matter of left superior and middle frontal gyrus and right superior frontal gyrus. Symmetrical T2 / FLAIR hyperintense areas were seen involving the superior cerebellar peduncles, dorsal mesencephalon including red nucleus and lateral aspect of thalami conforming to the dentatorubrothalamic tract. Subtle FLAIR / T2 hyperintensities were seen involving the pons along the pontine tegmentum, reticular nuclei and middle cerebellar peduncles suggesting involvement of the pontocerebellar tract. FLAIR hyperintensities were also seen in bilateral caudate nuclei, putamen and ventrolateral thalami. There was mild cerebral and cerebellar atrophy likely secondary to longstanding copper intoxication. No acute infarct/abnormal parenchymal/leptomeningeal enhancement was seen. **Discussion:**

Wilson's disease, also called hepatolenticular degeneration, is an inborn error of copper metabolism affecting multiple systems. It is inherited in an autosomal recessive manner caused by mutation of copper adenosine triphosphate transporter (ATP 7B) gene which codes for the synthesis of ceruloplasmin, a copper protein transporter [1]. It is characterised by the inability of the liver to excrete copper into the bile with excessive deposition of copper primarily in the liver and in the brain [2]. Clinical presentation is between 5-50 years of age. Symptoms depend upon the area affected.

In the brain, copper accumulation occurs primarily in the basal ganglia, thalami, brainstem and cerebellar peduncles. White matter involvement and atrophic changes are rare and occur late in the course of disease. We are reporting a

case of Wilson's disease with extensive involvement of white matter tracts in an adolescent male with basal ganglia lesions.

Wilson's disease can present with a wide spectrum of neuroimaging abnormalities [3]. Most characteristic findings include hyperintensity involving the lentiform nuclei on T2W sequences with subsequent involvement of caudate nuclei, thalami, pons and midbrain likely secondary to demyelination, softening, spongy formation and cavitatory disintegration [3]. Involvement of the thalami and brainstem structures without significant lentiform nuclei involvement is not uncommon.

Three white matter tracts are predominantly involved: Dentatorubrothalamic, pontocerebellar and corticospinal tracts. The dentatorubral tract originates in the dentate nucleus, passes through the superior cerebellar peduncle, traverses the red nucleus and terminates in the ventrolateral nucleus of thalamus. The pontocerebellar tract originates from pontine nuclei bilaterally and tegmental reticular nucleus and terminates by way of middle cerebellar peduncle, in all locules of cerebellar lobules. These tracts together constitute the cortico-ponto-cerebello-dentato-rubro-thalamo-cortico circuit, which is part of the extrapyramidal system. The abnormalities are thought to be secondary to extrapyramidal grey nuclei involvement. The corticospinal (pyramidal) tract contains projection fibres from the cerebral cortex, which extend into the corona radiate, the posterior limb of internal capsule [4, 5], centre of cerebral peduncle of mesencephalon, rounded foci in base of pons, medulla oblongata and the spinal cord [4, 6].

Subcortical white matter lesions involving frontal, parietal and temporal lobes manifesting as low signal intensity on T1-weighted images and high signal intensity on T2W images have also been reported [7]. Cerebral atrophy with ventricular dilatation, especially of the frontal horns, and cerebellar atrophy may also be seen in Wilson's disease [8]. Correlation between neuroimaging findings and clinical worsening has not been established yet. **Differential Diagnosis List:** Wilson's disease with extensive white matter abnormalities., Hypoxia, Extrapontine myelinolysis

Final Diagnosis: Wilson's disease with extensive white matter abnormalities.

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Description: Axial FLAIR image showing confluent area of hyperintensity involving the subcortical white matter of bilateral superior frontal gyrus. **Origin:** Department of Radiology, Medanta-The Medicty, Gurgaon,India



Description: Axial FLAIR image showing symmetrical hyperintense areas involving the superior cerebellar peduncles and pons. **Origin:** Department of Radiology, Medanta-The Medicty, Gurgaon,India



Description: Axial FLAIR image showing symmetrical hyperintense areas involving the dorsal mesencephalon. There is mild prominence of the cerebellar folia suggestive of cerebellar atrophy. **Origin:** Department of Radiology, Medanta-The Medicty, Gurgaon,India



Description: Axial FLAIR image showing hyperintense areas involving dorsal pons and middle cerebellar peduncles. Fourth ventricle appears prominent. **Origin:** Department of Radiology, Medanta-The Medicty, Gurgaon,India



Description: Axial FLAIR image showing symmetrical hyperintense areas in bilateral caudate nuclei, putamen and ventrolateral thalami. Bilateral sylvian fissures and frontal horns appear prominent.**Origin:** Department of Radiology, Medanta-The Medicty, Gurgaon,India