White matter changes on magnetic resonance imaging in a patient with neurodegenerative disease

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A 14-year-old Sri Lankan male had progressive dysarthria, dysmetria, gait ataxia, tremor, bradykinesia for 6 years with cognitive decline ensuing 1 year prior to admission. His sister was suffering from a similar illness. His parents were non-consanguineous. On examination, the patient had a Glasgow coma score of 8. He had quadrihypermic reflexes with extensor plantar response. Cerebellar assessment was difficult. Cogwheel rigidity was noted. There was no flapping tremor. Hepatosplenomegaly was elicited on abdominal examination. Kayser-Fleischer (KF) rings were confirmed by slit lamp examination.

Magnetic resonance imaging (MRI) showed increased signal intensity on T2-weighted image in basal ganglia and supratentorial with infratentorial gray and white matter [Figure 1].

Biochemical analysis is as follows: Serum ceruloplasmin level - 1 mg/dl (18-45), urinary copper excretion - 4.5 µmol/24 h, Aspartate transaminase (AST) (SGOT) - 48 U/l (10-35), Alanine transaminase (ALT) (SGPT) - 57 U/l (10-40), alkaline phosphatase - 226 U/l (100-360), serum bilirubin - 9.2 µmol/l (5-21), serum protein - 70 g/l, albumin - 39 g/l, globulin - 31 g/l, prothrombin time - 12.8 s International normalized ratio (INR 1.3), APTT - 20 s, serum ammonia - 25 µmol/l (<35 µmol/l), Na⁺ - 140 mmol/l, K⁺ - 4.5 mmol/l, serum creatinine - 62 µmol/l. Cerebrospinal fluid analysis was normal.

Commentary

Extrapyramidal features including bradykinesia and cogwheel rigidity, hepato-splenomegaly, the presence of KF rings, very low serum ceruloplasmin levels and increased urinary copper excretion seen in the above patient confirmed Wilson disease (WD). In the majority of patients with symptomatic WD neuroimaging studies are abnormal. WD has a wide spectrum of neuroimaging abnormalities.[1] The most conspicuous observations are atrophy of the brain and signal abnormalities in the basal ganglia. However, nearly all areas of gray and white matter can have T2 high signal changes.[2] The above case highlights the striking white matter changes on MRI.

The white matter changes in the above patient are diffuse. When a child presents with progressive cognitive decline over a long period of time with such MRI changes a heritable leukoencephalopathy needs to be considered. When the changes are diffuse and the patient does not have megalencephaly the differentials to be considered are: Vanishing white matter disease, Pelizaeus-Merzbacher disease and mitochondrial disorders.[3] It is unusual for WD to present with diffuse white matter changes as the changes are generally found in the posterior part of the brain. However, clinical and laboratory findings in the above patient confirmed WD and diffuse changes have been reported previously.[2] The high signal intensity of white matter on T2-weighted images in the above patient can be due to demyelination, softening, spongy formation and cavitary disintegration.[3]
MRI images in the above patient also demonstrate grey matter 
T2 high signal intensity changes, which are due to edema gliosis 
necrosis and cystic degeneration.\cite{1}

Although white matter changes are known to occur with long-term 
disease or following penicillamine therapy there are cases of white 
matter changes in the early stages of the disease as well. The 
incidence of white matter changes have been reported by several 
studies. Van Wassenaer-van Hall \textit{et al.} reported an incidence of 41% 
in 1995.\cite{4} Two Indian studies revealed white matter changes in 
10\% and 25\% of WD.\cite{5,6} There is a paucity of data from Sri Lanka.

This case highlights the importance of recognizing white matter 
changes, which occur in WD especially when evaluating a 
neuropsychiatric disorder.

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