Early White Matter Changes in Wilson Disease

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Abstract
We describe a 12 years old male who presented with one year history of cognitive decline with extrapyramidal features. Wilson disease was diagnosed on basis of biochemical studies and MRI. MRI showed increased signal intensity on $T_2$ weighted images in basal ganglia and supratentorial with infratentorial gray and white matter. Our patient developed white matter changes early in course of disease.

INTRODUCTION
Wilson's classic description of “progressive lenticular degeneration; a familial nervous disease associated with cirrhosis of liver” appeared in 1912. Excessive copper accumulates systemically, particularly in liver and brain. In the brain copper accumulation occurs primarily (but not exclusively) in basal ganglia. Earlier studies have shown white matter changes in the late course of disease. We are reporting a case where such changes has been observed in early stage of disease.

CASE REPORT
A 12 years old male presented with one year history of gradual impairment of intellectual faculties, impulsiveness and six month history of tremor of limbs, generalized slowness of movement and dysarthria. Prior history of liver dysfunction was absent. Neurological examination showed fixity of facial muscles with mouth constantly agape, hypokinetic dysarthria, postural and intention tremor affecting the upper limbs, generalized rigidity and flexed posture.

Wilson disease was diagnosed on the basis of Kayser-Fleischer rings, increased urine copper excretion (300 µgm/day: reference range 25-50 µgm/day), low serum ceruloplasmin (10 mg/dl: reference range 20-42 mg/dl), low serum copper (7 µM/L; reference range 11-24 µM/L). MRI showed increased signal intensity on $T_2$ weighted image in basal ganglia and supratentorial with infratentorial gray and white matters (Figs. 1, 2).

Patient was treated with D-pencillamine for six months and there was slight improvement in the neurological examination.

DISCUSSION
Wilson disease is an autosomal recessive disorder caused by copper adenosine triphosphatase transporter (ATP7B)
Wilson disease is depicted with a wide spectrum of neuroimaging abnormalities. None of the abnormalities is pathognomonic for Wilson disease. Although Wilson disease is often associated with symmetrically high signal in the basal ganglia on T2 weighted images but finding of authors showed that nearly all areas of gray and white matter can be involved. The high signal intensity of white matter on T2 weighted images can be explained by demyelination, softening, spongy formation and cavitary disintegration. The high signal intensity of gray matter nuclei on T2 weighted images can be caused by edema gliosis necrosis and cystic degeneration. Typically white matter lesions occur after prolonged disease or following treatment with D-pencillamine.

Peter Hedera reported white matter changes in early course of disease and now we are reporting new case where the patient developed leukoencephalopathy within one year of disease course. Neurologist needs to recognize the possibility of white matter disease in early stage of Wilson disease; the concurrent presence of abnormal signal on T2 weighted MRI in basal ganglia and appropriate biochemical studies should be obtained to confirm a diagnosis of Wilson disease, as results are rewarding.

**References**


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**Announcement**

**Infectious Disease Certificate Course at Hinduja Hospital in September 2004**

A two week, certificate course in Infectious Diseases from **30th August to 12th September, 2004**, is scheduled to be held at PD Hinduja National Hospital organised by Departments of Medicine, Paediatrics and Microbiology.

Eligibility for Course and who should attend : 0-10 years Post MD Medicine/Paediatrics/Microbiology

Focus : Common challenges in infectious diseases.

Format : Ward rounds, Cases from our archives, Interactive lectures, Microbiology discussions, Visits to various infectious disease hospitals.

Timings : 9 am to 4 pm

Please send a short CV to enable screening for selection as number of seats is limited. Maximum number of registrations : 15

Registration : Rs. 2,500/-

Last Date for Registration : 31.7.2004

Contact : **Dr. Rajeev Soman**, Organising Secretary. Tel. 2444 7704/5.

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