Case report: Wilson’s Disease MR Imaging

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Abstract: The spectrum of Wilson’s extends from acute hepatitis to fulminant hepatic failure and associated various neurological manifestations. Here, we report a case of Wilson’s disease, and discuss its MRI findings.

Introduction

An autosomal recessive hereditary inborn error of metabolism, Wilson’s disease or hepatolenticular degeneration is an uncommon disease, caused by mutations in the ATP7B gene on chromosome 13. This leads to a deficiency of ceruloplasmin. The disease involves the liver and the lentiform nucleus of brain which cause the symptoms. The hepatic forms such as acute hepatitis, chronic hepatitis, cirrhosis of liver and acute fulminant hepatic failure occurs in childhood. Neurological manifestations classically seen in young adults and early symptoms are in-coordination, dysarthria, dystonia, rigidity, tremor and difficulty with fine motor tasks. Copper deposition in Golgi complexes and the mitochondria results in oxidative damage primarily to the liver, brain, kidney, skeletal system, and eye. Selective vulnerability of the corpus striatum to mitochondrial dysfunction accounts for the predominant basal ganglia volume loss seen in Wilson’s. Typical sites of cerebral involvement are deep grey matter and deep central white matter and dentate nucleus of cerebellum. Grey matter nuclei involvement is more common, usually bilateral symmetric in the putamen, caudate, thalamus, globus pallidus, dentate nucleus, pons and mesencephalon.

1. Case Report

A 17 year old girl, came with complains of abdominal pain, distension and difficulty in speaking. She had history of exhibiting involuntary movement of the limbs episodic abnormal posturing and rigidity of limbs. No h/o jaundice, hematemesis, melena, hemoptysis, fever, rash, joint pains, chest pain, history of drug intake, bleeding disorders or consanguinity in the family. Her developmental milestones were normal. On examination her vital were stable. Liver and spleen were not palpable.

Neurological examination showed dystonia, choreiform movements of the limbs, gait ataxia and Babinski’s sign was positive. Liver function test were normal. Serum Ceruloplasmin level was 18mg/dl, serum copper was 312mcg/dl. 24 hours urinary copper was 1144mcg/24hrs. Serum calcium level was 7 mg/dl. On MRI, T2-weighted images revealed high signal intensities in the bilateral basal ganglia and putamen region, same region is hypointense on T1-weighted images. Ophthalmoscopic examination by slit lamp showed Kayser-Fleischer rings in both eyes. Patients was diagnosed as wilson’s disease and started on penicillamine therapy and she responded well to therapy. The patients dystonia and abnormal movements were reduced.

2. Discussion

In Wilson’s is characterized by an inability of the liver to excrete copper into the bile and there is abnormal deposition of copper in the basal ganglia, eyes, liver and other tissues. The clinical presentations of WD are hepatic and neuropsychiatric problems. Chronic active hepatitis, culminating in cirrhosis is the most common hepatic presentation, but some patients present with fulminant liver failure. Typical neurological sign include tremor, rigidity, drooling, speech changes, incoordination, tremor, difficulty with fine motor tasks, and gait difficulties. Psychiatric manifestations include compulsive behavior, aggression, depression and impulse behavior. Clinical presentation of Wilson’s is between 5 to 50 years. However, early childhood Wilson’s usually presents with liver disease and neurological manifestations are rare before the age of ten years. Diagnosis is based on clinical evaluation along with biochemical and neuroimaging confirmation. Biochemical studies reveal a low serum ceruloplasmin level (<20 mg/dl) and increased urinary copper excretion (more than >100 ig copper per 24 hours). Hepatic copper estimation, of more than 250 g/g of dry tissue (Normal 15-55 ig/g) is the most definitive method of diagnosis.
Figure 1 & 2 MRI T2 & T1-weighted image reveals hyper & hypointensity in bilateral Basal Ganglia & Thalamus

Figure 3 & 4 show Isointense to hypointense lesions on T1-Weighted Image
Figure 5 & 6 show hyperintensities on T2 Flair images

Figure 7 & 8 show diffusion restriction on DWI
In patients with Wilson’s disease, neuroimaging abnormalities occur in gray matter of lentiform, caudate and thalamic nuclei. Cerebral atrophy with ventricular dilatation especially of the frontal horns and cerebellar atrophy are also frequently observed in Wilson’s disease. Our patient did not have significant ventricular dilatation or cerebellar atrophy. Ventricular dilatation, brainstem atrophy, and posterior fossa atrophy are other possible findings.

MRI provides a more elaborate anatomical information than CT scan of brain on the structure of basal ganglia and brain stem. On MRI there is bilateral symmetrically altered signal intensity in the caudate, lentiform, external capsule, internal capsule, ventrolateral thalami and they are hypointense on T1-weighed images and hyperintense on T2-weighed sequences. There is diffusion restriction on DWI. The original description of the ‘face of the giant panda’ sign by Hitoshi et al consisted of high signal intensity in the tegmentum, preservation of signal intensity of the lateral portion of the pars reticulata of the substantia nigra and hypointensity of the superior colliculus.

In Wilson’s patients, neuroimaging abnormalities occur in gray matter of lentiform, caudate and thalamic nuclei. The disease is treated with lifelong use of chelating agents such as D-penicillamine or trientine hydrochloride, drugs that help remove copper from tissue. Zinc is used for maintenance therapy and for treatment of asymptomatic siblings.

4. Conclusion

There is a rapid rise in the number of Wilson’s disease diagnosed especially in developing and new economies. MRI has gained an important part in arriving at the diagnosis in Wilson’s disease.

4. References


