

Answer to this month's Radiological Conference

Answer:

b) Wilson's disease

Radiological findings

Axial non-enhanced CT brain shows bilateral symmetrical hypodensities in the lentiform nuclei. Calcification is also observed incidentally at the basal ganglia on both sides (Figure 4). Axial T2-weighted MR images show focal areas of hyperintense signal in the

putamina (more marked in the peripheral zones) and dorsal mid brain (Figures 5 & 6).

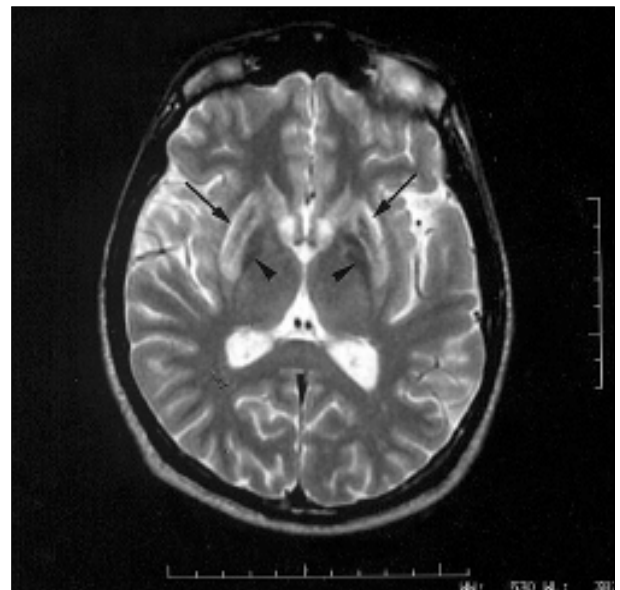
Figure 4: Axial non-enhanced CT brain identical to Figure 1 with addition of arrows. Symmetrical hypodense areas are present in the lentiform nuclei (arrow). Note the calcification in the both basal ganglia (arrowhead)



Figure 5: Axial T2-weighted MR image identical to Figure 2 with addition of an arrow. High signal lesion is seen at the dorsal mid brain (arrow)



Figure 6: Axial T2-weighted MR image identical to Figure 3 with addition of arrows. Areas of hyperintensity are seen at the lentiform nuclei bilaterally (arrow). Note the peripheral zones are more affected. Decreased signal intensity in the basal ganglia is probably due to calcification (arrowhead)



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Discussion

Wilson's disease

Wilson's disease is an autosomal recessive disorder of copper metabolism caused by a deficiency of ceruloplasmin and the defect has been identified in chromosome 13. It has an approximate incidence of 1:1,000,000 with a higher incidence among European Jews, southern Italians, Taiwanese and Japanese.

Abnormal deposition of copper occurs in various tissues but primarily in the liver, lentiform nucleus of the brain, cornea, bones and kidneys. Wilson's disease is characterised by cirrhosis of the liver and degeneration in the basal ganglia (hepatolenticular degeneration). Although the disease can occur at any age, most of the affected patients become symptomatic between the first and second decades. Pathological changes in the brain consist of a loss of neurons and gliosis in the basal ganglia, thalami, brainstem, and dentate nuclei. These changes are most likely a result of copper deposition in the brain or as a complication of liver disease.

Chelating agents, such as penicillamine or trientine, which increases the urinary excretion of copper, have been used in Wilson's disease for reversing the positive copper balance. Recently, zinc sulfate, which blocks the absorption of copper and increases copper excretion in the stool, has been used in the long-term treatment of Wilson's disease with promising results.

The clinical features of Wilson's disease include liver failure, tremor, dysarthria, bradykinesia, dystonia and dementia.

The typical CT findings are generalised cerebral atrophy with hypodensity in the basal ganglia bilaterally. Hypodense lesions in the brainstem, dentate nucleus and cerebellar white matters are also frequently present. Contrast enhancement is generally absent. Since MR imaging is more sensitive than CT in detecting the pathological changes, it should be the modality of choice in evaluation of the disease. On T2-weighted MR images, high signal lesions may be present in the lentiform nuclei, caudate nuclei, thalami, brainstem and dentate nuclei. The peripheral zones of the lentiform nuclei are most severely affected. The abnormalities are typically hypointense on T1-weighted MR images. Rarely, increased signal intensity in the basal ganglia on T1-weighted MR images may be seen in patients with portal-

systemic encephalopathy. The possible explanations include the paramagnetic effects of copper or iron.

Carbon monoxide poisoning

Bilateral high signal lesions on T2-weighted MR images at the basal ganglia is characteristic for carbon monoxide poisoning. Carbon monoxide poisoning leads to cytotoxic oedema and necrosis of the globus pallidus, which accounts for the high signal intensity on the MR images. Other locations, such as hippocampus, white matter, cortical gray matter, can also be affected. The diagnosis is confirmed by the presence of carboxyhemoglobin in the blood.

Leigh's Disease (subacute necrotizing encephalopathy)

It is a rare metabolic disorder caused by enzyme deficiencies necessary for oxidative metabolism within mitochondria (eg. pyruvate carboxylase, pyruvate dehydrogenase, cytochrome c oxidase). The enzyme deficiencies lead to accumulation of lactate and pyruvate in serum and cerebrospinal fluid.

Affected individuals usually present in first 2 years of life and early childhood with ataxia, dystonia, motor weakness and behaviour disorders. Death from respiratory failure within the first year is typical. The clinical features and pathological changes are similar to Wernicke's encephalopathy in adults. Pathological changes are present in the basal ganglia, thalami, dorsal midbrain, pons and spinal cord.

On MR imaging, the lesions are hypointense on T1-weighted image, hyperintense on T2-weighted image and typically symmetrical. The MR features may be indistinguishable from Wilson's disease but the diagnosis can be established by the age of onset and biochemistry tests.

Cerebral anoxia

Cortical and ganglionic injuries may be caused by cerebral anoxia. Symmetrical high signal lesions on T2-weighted MR images may be present in the basal ganglia,

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caudate nuclei, thalami and cerebral cortex. The high signal intensity on MR images is due to the presence of oedema. Anoxic changes in the basal ganglia can be very subtle in infants during the acute phase. CT is helpful to demonstrate the low-density lesions in patients whose MR images are equivocal.

Osmotic myelinolysis

Osmotic myelinolysis is a toxic demyelinating disease that associated with chronic alcoholism, rapid correction of hyponatraemia and hypernatremia. The central pons is the most common location but extrapontine myelinolysis may be found in the basal ganglia, midbrain and thalami. The lesions are hyperintense on T2-weighted MR images and the central pons is typically hyperintense with relative sparing of the descending corticospinal

tracts. The diagnosis can be established by using imaging findings and clinical history. ■

Further readings

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