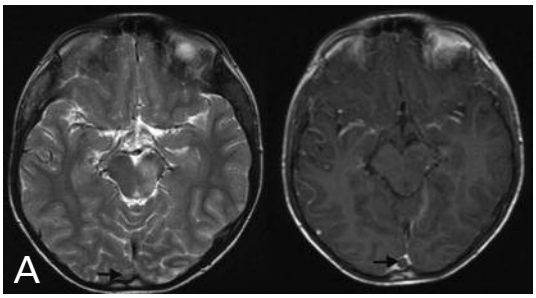
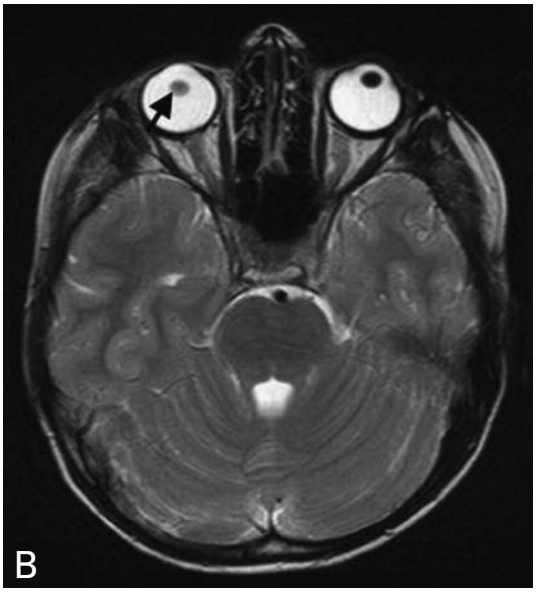
JBR–BTR, 2010, 93: 322.

IMAGES IN CLINICAL RADIOLOGY





Homocystinuria, typical brain MRI findings

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An 8-year-old male presented with recent history of headache, several episodes of convulsion and visual field deficits. Brain MRI revealed multiple T_2 hypersignal intensity foci within the left cerebral peduncle, basal ganglia, thalami and left parietal cortex, believed to represent ischemia. There was also iso–slightly hyperintense signal at the site of superior sagittal sinus, replacing the normal flow void pattern, suggestive for dural sinus thrombosis. After gadolinium injection thrombosis of superior sagittal sinus was confirmed (Fig A). Evaluation of orbits disclosed posterior dislocation of the right lens (Fig B). These findings were compatible with the diagnosis of homocystinuria which was later confirmed by laboratory data.

Comment

Homocystinuria is an inborn error of methionine metabolism caused mainly by low activity or a deficiency of the enzyme cystathionine- -synthase. This defect results in intimal irregularities that cause arteriosclerosis, arterial thromboembolism ,and venous thrombosis in children and young adults. This appears to be the result of excess homocysteine, which changes coagulation factor levels, prevents small arteries from dilating, and causes proliferation of smooth muscle cells in arterial walls. Homocystinuric infants appear healthy at birth, and their early development is unremarkable until seizures, developmental slowing, or cerebrovascular accidents occur between 5 and 9 months of age. Ectopia lentis is seen in more than 90% of affected individuals. Lenticular dislocation has been recognized as early as 18 months of age, but it generally occurs between 3 and 10 years.

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