Glutaric aciduria type1: CT diagnosis

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Sir,

A 4-year old boy presented with delayed mental growth, speech disturbances and abnormal size of head. Physical examination revealed macrocephaly. There was incomplete achievement of mental milestones. Computed tomographic (CT) scan of brain revealed bilateral frontotemporal atrophy [Figure 1], bilateral enlarged sylvian fissures and few hypodensities in the lentiform nuclei. History did not reveal any evidence of accidental or non-accidental head injury. Biochemical investigations clinched the diagnosis.

Glutaric aciduria type 1 (GA-1) is an autosomal recessive inborn error of lysine, hydroxylysine and tryptophan metabolism that results from a deficiency of glutaryl-CoA dehydrogenase.

Common features on neuroimaging include increased spaces anterior to the frontotemporal lobes [Figure 2] (vs. frontotemporal atrophy) wide sylvian fissures, (giving a "bat-wing" formation) and prominent interhemispheric fissures[1]. There may be diffuse hypodensity of the basal ganglia. Widening of the sylvian fissure, mesencephalic cistern and expansion of CSF spaces anterior to the temporal lobes are cardinal signs of GA-1. If combined with abnormalities of the basal ganglia and white matter, GA-1 should be strongly suspected[2].

A prominent clinical feature of infants and children with glutaric aciduria type 1 is macrocephaly[3]. The finding of very widely open opercula suggests glutaric acidemia type I, and if combined with basal ganglia lesions is almost pathognomonic, especially in a child with macrocephaly[1].

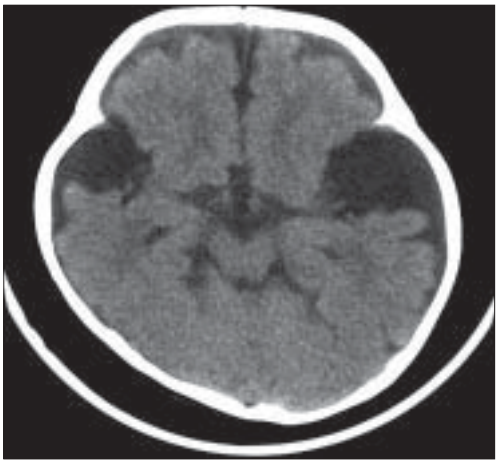

Figure 1: Frontoemporal atrophy and widened CSF spaces

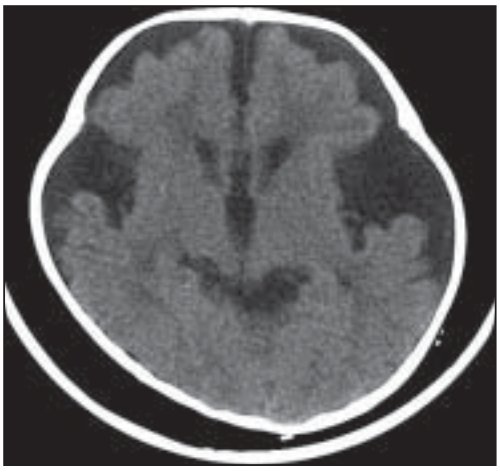

Figure 2: "Bat-wing" dilatation of the sylvian fissures and prominent interhemispheric fissure

Conventional T2-weighted and fluid-attenuated inversion recovery magnetic resonance images of the brain showed hyperintensity in the caudates and putamina bilaterally with subtle involvement of the medial frontal lobes. Diffusion-weighted magnetic resonance images showed striking restricted diffusion in the caudates and putamina consistent with acute necrosis.[4]

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