

Leigh's Disease (Subacute Necrotising Encephalomyelopathy) - A Case Report

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INTRODUCTION

Leigh's disease or Sub-acute necrotising encephalomyelopathy (SNE) is a rare neurodegenerative disorder of childhood that was first described by Leigh in 1951 [1].

It is possible to come to a diagnosis of probable SNE during life on the basis of clinical signs and symptoms, mode of inheritance, metabolic abnormalities, and neuroimaging findings [2]. We report a rare case of Leigh's disease diagnosed on CT and MRI and discuss the role of imaging in its diagnosis.

CASE REPORT

A three year old girl presented with regression of the achieved milestones, generalized weakness and involuntary movements for the past three months. Clinical examination revealed the child had concomitant and convergent squint. CNS examination showed increased tone in the lower limbs, bilateral extensor plantar reflexes and intentional tremors. Laboratory analysis showed metabolic acidosis.

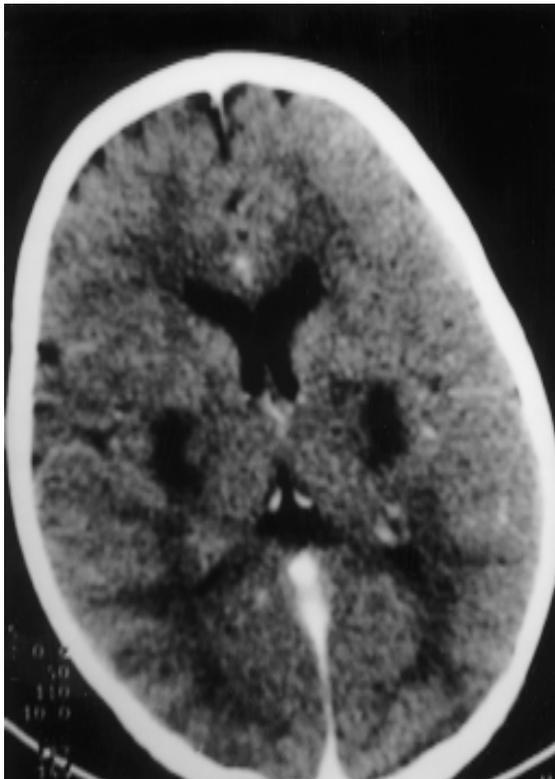


Fig 1. CECT showing bilaterally symmetrical non-enhancing hypodensities in the putamina.



Fig 2. Axial T1 weighted MR image showing symmetrical hypointense lesions in the putamina.

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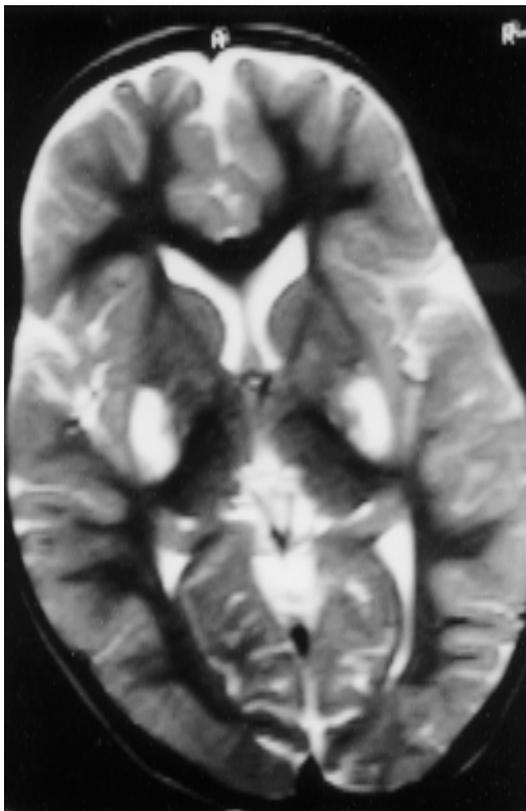


Fig 3. Axial T2 weighted MR image at the same level showing the lesions which are hyperintense and almost similar to CSF signal intensity.

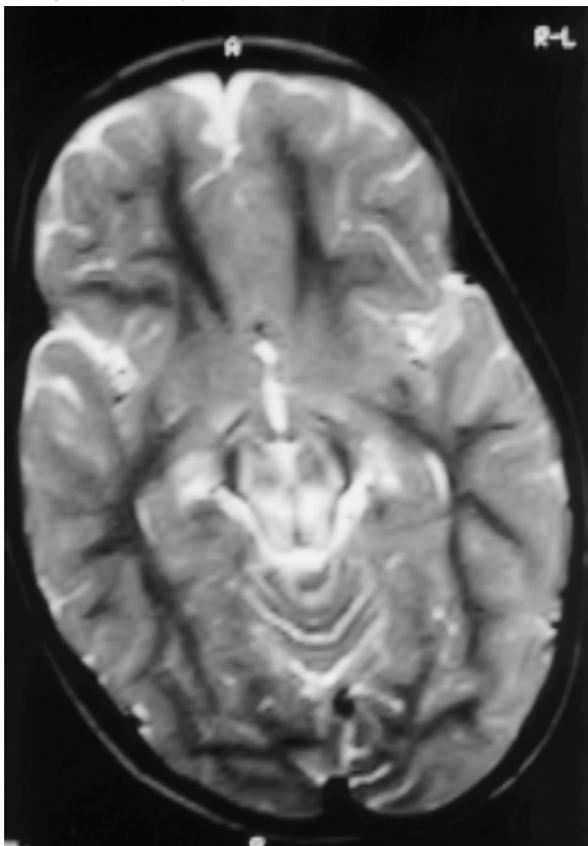


Fig 4. Axial T2 weighted MR image at the level of midbrain showing hyperintense signal in the peri-aqueductal grey matter.



Fig 5. Axial T2 weighted MR image at the level of cerebellum shows symmetrical hyperintense lesions in the dentate nuclei of the cerebellum.

The CECT scan showed bilaterally symmetrical non-enhancing low attenuating lesions in the putamina (Fig. 1.). MRI studies showed bilaterally, symmetrical lesions in the putamina, which were hypointense on T1 weighted and hyperintense T2 weighted images. (Fig.2,3). The peri-aqueductal grey matter and dentate nuclei of the cerebellum were also involved and showed the similar signal characteristics (Fig.4,5). Based on the clinical features and imaging findings, a diagnosis of Leigh's disease or SNE was made.

DISCUSSION

Leigh's disease or SNE is a rare progressive neurological disorder of the childhood. It is usually inherited in an autosomal recessive fashion, and the underlying defect can be at any of a number of sites in the enzyme pathway for respiratory metabolism. Associated mitochondrial enzyme deficiencies are pyruvate carboxylase, pyruvate dehydrogenase, cytochrome C oxidase, and Complex 1 (NAD-Coenzyme Q Reductase) deficiencies [3,4]. The pathology of SNE is characterized by capillary proliferation with bilaterally symmetric gray and white matter necrosis, spongiform degeneration or vacuolization and

demyelination [3,5].

It presents early in life with psychomotor regression, abnormal muscle tone, weakness, dystonia, brainstem and cerebellar dysfunction (ataxia), visual loss, missed milestones or regression of the achieved milestones, tachypnea, and seizures [1,3,6]. Age of onset of symptoms is usually less than 2 years (infantile form), but others may present in childhood (juvenile form) and unusually in adulthood. Death usually occurs within a few years after onset of symptoms, typically from progressive respiratory failure [4,5]. Laboratory analysis shows metabolic acidosis with elevated blood, CSF lactate, and pyruvate concentrations [3].

The lesions are seen as non-enhancing hypodensities on CT. Low attenuation in the putamina on CT is considered to be characteristic of the disease [5,7]. MR imaging shows bilateral areas of abnormal high signal intensity on T2 weighted images in the basal ganglia, periventricular white matter, corpus callosum, periaqueductal grey matter and brainstem [3,5,6]. In the basal ganglia, the putamen is particularly involved. In one series, 100% of the patients with proven SNE had putaminal involvement [3].

Proton spectroscopy has demonstrated elevated brain lactate levels in the basal ganglia, occipital cortex, and brainstem. This abnormality was greatest at sites of abnormal signal intensity on T2-weighted images [8].

The diagnosis of Leigh's disease should be considered in appropriate clinical and laboratory settings whenever

symmetrical hypodensities are encountered in the putamina and midbrain on CT and further investigated with MRI, which is more sensitive than CT in establishing an antemortem diagnosis of Leigh's disease.

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