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WisdomGale

CASE REPORT

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Leigh syndrome: A must-know bilateral symmetrical basal ganglia affection

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ABSTRACT

Leigh syndrome also known as subacute necrotizing encephalomyopathy is a rare progressive neurological disease with a reserved prognosis affecting the brainstem and the basal nuclei, the symptomatology of which is revealed in the majority of cases during the first two years of life. In this article, we report the case of a 4-year-old child referred to our center following a consultation for delayed psychomotor acquisitions with axial hypotonia and elevated lactate levels. Brain MRI showed a characteristic appearance of this syndrome. This clinical case reports the MRI aspect of Leigh syndrome, a rare and serious pathology responsible for bilateral and symmetrical lesions of the basal ganglia.

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Leigh disease; basal ganglia; magnetic resonance imaging

Introduction

Leigh syndrome, or subacute necrotizing encephalomyelopathy, is a rare progressive neurological disease affecting the brainstem and basal ganglia, with symptoms typically appearing in the first two years of life in the majority of cases [1,2]. The prognosis is bleak; however, a positive diagnosis allows for genetic counseling for the parents.

Patient and observation

Patient information and clinical results

This concerns a 4-year-old child who underwent a brain magnetic resonance imaging (MRI) as part of an assessment for delayed psychomotor development. The medical history did not reveal any neonatal distress or other incidents during the neonatal period. Clinical examination revealed axial hypotonia. Laboratory tests showed elevated lactate levels.

Diagnostic evaluation

This prompted a brain MRI (Fig. 1), which revealed bilateral and symmetrical T2, fluid-attenuated inversion recovery (FLAIR), and diffusion hypersignals in the lenticular and caudate nuclei, as well as an isosignal diffusion pattern in the posterior parts of the lenticular nuclei with diffusion restriction. Spectroscopic sequences showed a lactate peak in the central gray nuclei with decreased N-acetylaspartate (NAA) and a slight elevation of choline.

Therapeutic intervention and follow-up

Following the diagnosis, the patient was hospitalized in the pediatric neurology department. Treatment with vitamin supplementation was initiated, and genetic counseling was offered to the parents.

Discussion

The first case of Leigh syndrome was diagnosed based on an autopsy study of a child who died in 1951 by the english physician Denis Leigh [3]. However, this etiological entity encompasses a range of genetic and metabolic abnormalities identified through advances in molecular biology [2]. The pathophysiological mechanism of the lesions is due to a defect in aerobic metabolism (oxidative phosphorylation), leading to severe energy deficits at the cellular level. This involves multifactorial genetic determinants, including mutations in mitochondrial deoxyribonucleic acid (DNA): transmission occurs either in an autosomal recessive mode or in a mitochondrial mode [4–6]. Clinically, the symptoms reflect the involvement of the central

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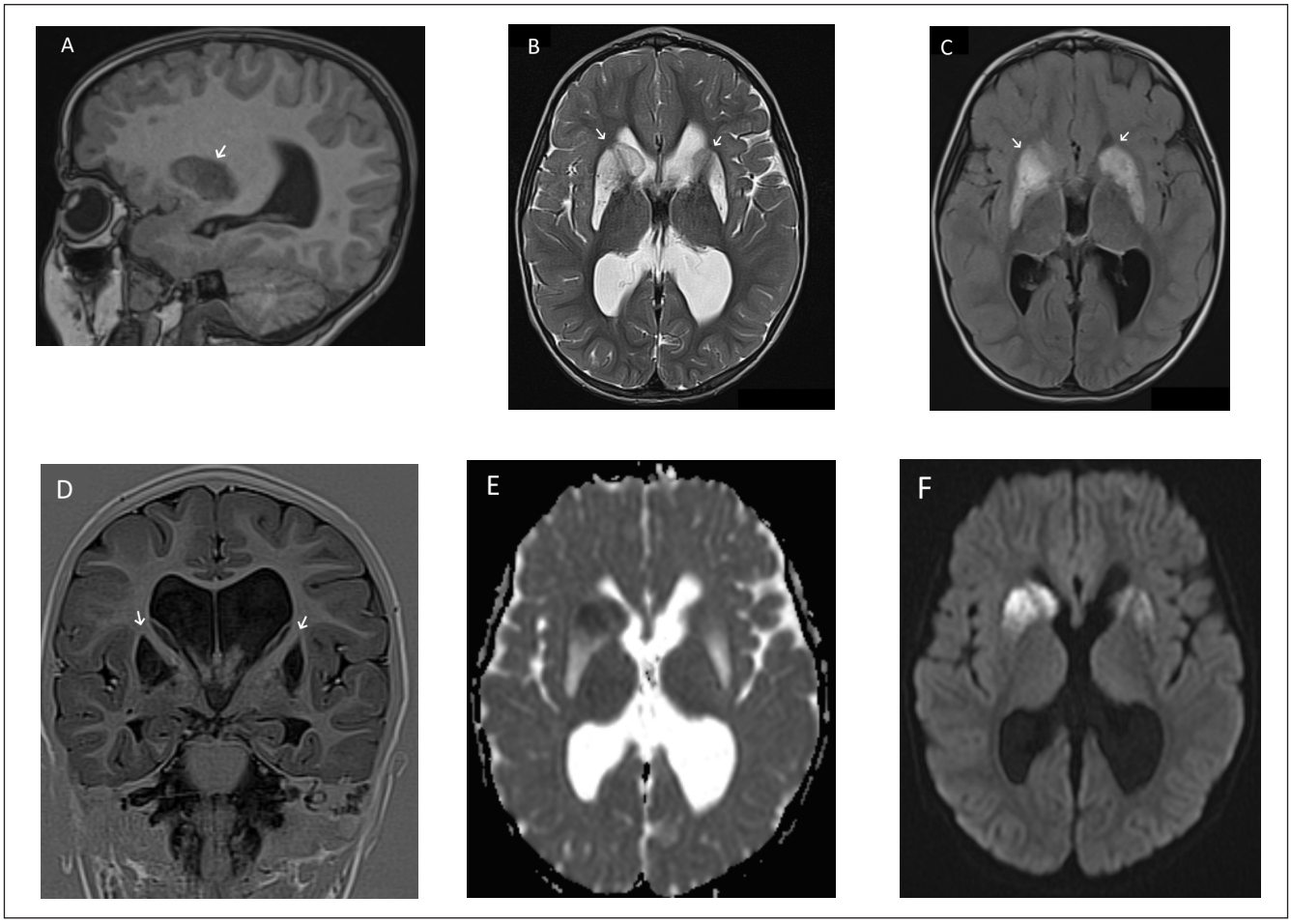


Figure 1. Brain MRI: signal abnormality of the lenticular nuclei: hyposignal on sagittal slice in T1 sequence (A), hypersignal on axial slices in T2 and FLAIR (B) and (C), and hyposignal on coronal slices in TIR (D). Axial slices in ADC (E) and DWI (F) show restricted diffusion in the anterior part of the lenticular nuclei.

gray nuclei and brainstem. They begin in the first two years of life and include neurological signs such as axial hypotonia, nystagmus, ophthalmoplegia, ataxia, and respiratory disturbances [1,7]. The diagnosis may be suspected in the presence of bilateral and symmetrical involvement of the central gray nuclei as T2 hypersignals with restricted diffusion at their level, frequently involving the putamen [4]. However, locations include the thalamus, substantia nigra, cerebellar dentate nucleus, spinal gray matter, and even cerebral white and gray matter with lesions resembling ischemia [1,2,5,7]. Cases of association with mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes syndromes have been reported in the literature [2,8]. The prognosis is bleak, and progressive neurological deterioration often leads to early infant death. However, in cases of certain mutations, treatment with vitamin supplements such as thiamine (vitamin B1) appears to improve symptoms [1,7,9]. Our case, the first diagnosed in our institution, unfortunately, did not undergo genetic sequencing to search for a causative mutation, especially since a founder mutation was discovered in a Moroccan family in 2013 [9].

Conclusion

Magnetic resonance imaging is crucial in the diagnosis of Leigh syndrome and should involve the entire central nervous system to perform a comprehensive lesion assessment and aid in the diagnostic process. The prognosis is severe but nonetheless reflects the underlying etiological heterogeneity due to the variable sensitivity to treatments currently available.

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