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Megalencephalic Leukoencephalopathy with Subcortical Cysts on Three Tesla Magnetic Resonance Imaging

Üç Tesla Manyetik Rezonans Görüntülemede Subkortikal Kistlerle Birlikte Megalensefalik Lökoensefalopati

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Introduction

A girl aged 4 years presented with progressive macrocephaly and slow deterioration of motor functions with cerebellar ataxia and mild spasticity over the last 3 years. She had recently had one episode of seizure. There was no fever. Her routine blood laboratory results were unremarkable. The patient underwent 3 Tesla magnetic resonance imaging (MRI) of the brain, which revealed symmetrical mildly swollen diffusely abnormal T2 hyperintense cerebral white matter on both sides along with T1 hypointense and T2 hyperintense cysts in the bilateral anterior temporal lobes and superior frontal lobes (Figure 1, 2). The differential diagnosis of macrocephaly and diffuse leukoencephalopathy includes Canavan disease, Alexander disease, infantile-onset gangliosidosis, glutaric aciduria, and megalencephalic leukoencephalopathy with subcortical cysts (MLC) (1). However, none of these share the characteristic combination of macrocephaly, diffuse cerebral white matter abnormalities and subcortical cysts. Hence, the combination of the above MRI features was diagnostic of MLC. The patient was started on antiepileptic drugs along with physiotherapy sessions.

MLC was identified in 1995 by van der Knapp et al. (1). Mutations in a gene called MLC1 seem to be the cause of this autosomal recessive disorder (2). MLC is the most common leukodystrophy with megalencephaly observed in India, and the majority of the patients have been from the Aggarwal community (3). MLC characteristically shows early onset and slow progression, whereas differentials like Canavan and Alexander disease show rapid progression. There is no curative option available, as such, supportive therapy includes antiepileptic drugs in case of seizures; physical therapy to improve motor function; and special education and speech therapy if necessary (2). Prenatal diagnosis is possible by analysis of DNA extracted from fetal cells obtained through

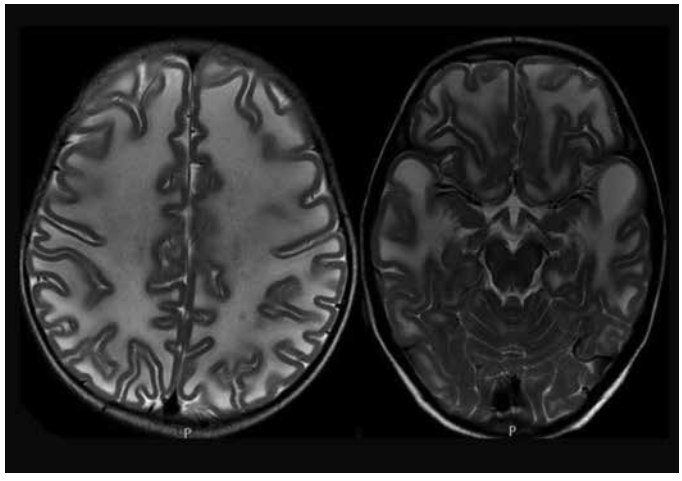

Figure 1. Axial T2-weighted image shows diffuse symmetrical T2 hyper-intense signal change in bilateral cerebral white matter and T2 hyper-intense subcortical cysts in the bilateral anterior temporal lobes

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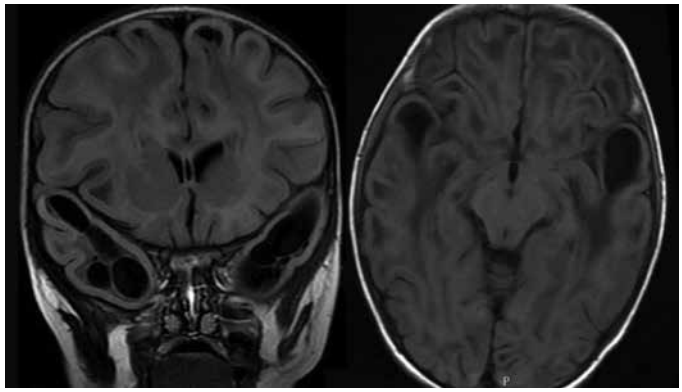

Figure 2. Coronal and axial T1-weighted image showing bilateral symmetrical T1 hypo-intense subcortical cysts in anterior temporal lobes and superior frontal lobes

amniocentesis or chorionic villus sampling (3). MLC must be included in the differential diagnosis for any patient who presents with infantile-onset macrocephaly with signs and symptoms of white matter because early physical rehabilitation may prolong ambulatory life. Though 3 Tesla MRI provides superior image quality of the affected brain, routine sequences do not reveal any new findings.

Ethic

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