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IMAGES IN PEDIATRIC SCIENCES

Megalencephalic leukoencephalopathy with temporal cysts

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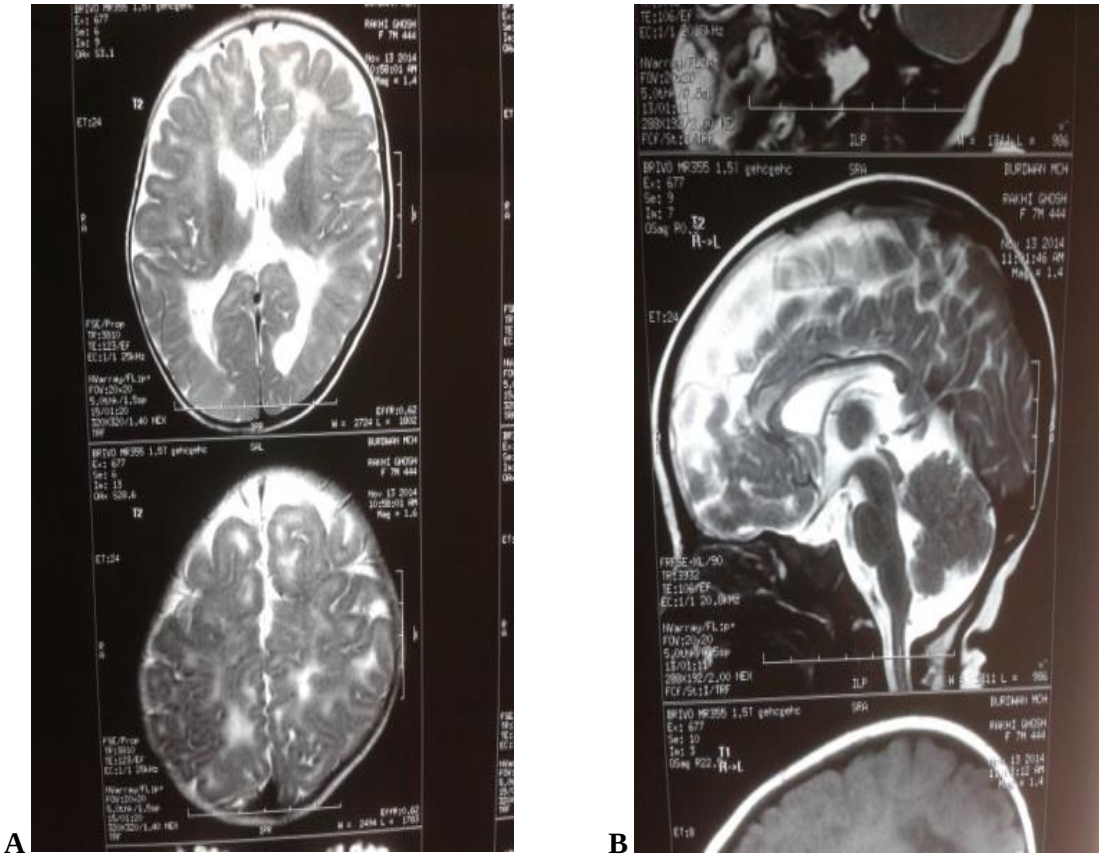

Figure 1. T2W Brain MRI images. A: bilateral hyperintense posterior periventricular white matter and centrum semiovale with bilateral temporal cystic changes. B: extensive demyelination.

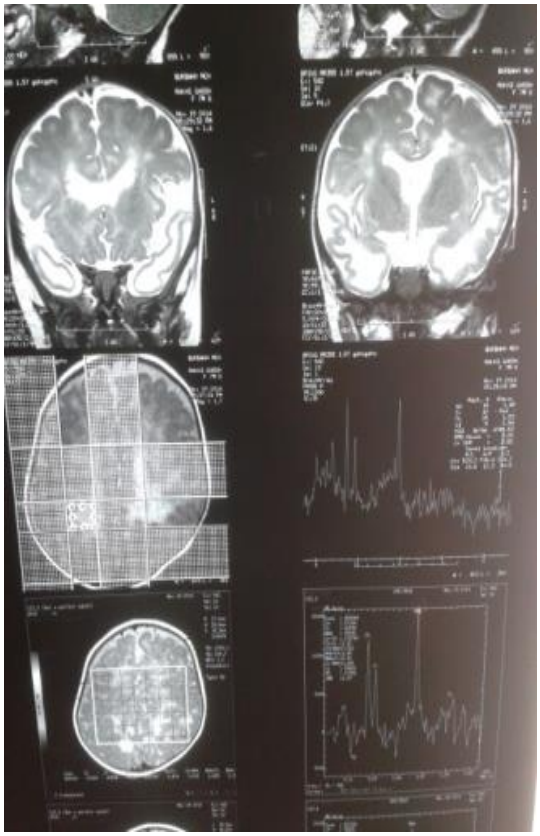

Figure 2. MR spectroscopy over the white matter lesion revealed increased choline peak, mildly decreased Cr peak with normal N-acetylaspartate peak.

A seven months old female child from parents of non consanguineous marriage presented to us with progressively enlarging head size and delayed developmental milestone like absent head holding and inability to sit with support. She had no history of convulsion. On examination, she had a head circumference of 51 cm (> 3rd SD, WHO), tone was normal in all limbs with normal reflexes and no organomegaly was noted. Electromyography and nerve conductance velocity were normal. MRI of brain showed altered intensity hypo in T1 hyper in T2/ FLAIR along the deep white matters of both posterior perventricular white matter and centrum semiovale with cystic changes in both temporal lobes. MR spectroscopy over the white matter lesion revealed increased choline peak, mildly decreased creatinine peak with normal N-acetylaspartate (NAA) peak. Choline: creatinine = < 2, choline: NAA=<1 and NAA: creatinine = 1.5 in single voxel and 2.8 in multi voxel.

Megalencephalic leukoencephalopathy (MLC) is a rare autosomal recessive disease (1). The degree of macrocephaly is variable ranging from 4-6 SD above the mean (2). Almost all patients have epilepsy since infancy (3). Slow deterioration of motor functions, ataxia and spasticity appears in early childhood period. Dystonia, athetosis and other extrapyramidal lesions are also mentioned in some patients in later age group. Most of the patients succumb to death in their first or second decades, few survive till fourth decade as reported by Subhra Aditya et al. (4). Genetic basis shows that the disease is caused due to mutation in MLC1 gene, which is located in chromosome 22qtel (5,6). It is found to be more common in the 'Agarwal' community in India (7). The typical MRI brain findings are 'swollen white matter' and bilaterally symmetrical diffuse supratentorial changes in the cerebral hemispheres relatively sparing the central white matter structures like corpus callosum, internal capsule and brain stem. Grey matter is usually spared. With time the 'swollen white matter' decreases and cerebral atrophy ensues (4). The subcortical cysts may increase in size and number. Moderate decrease in NAA/ Cr and Choline/Cr ratios have been reported in patients with MLC on MR spectroscopy (8).

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