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A Rare Case of Encephalopathy in a 9 Year Old Child

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Abstract: ***Megalencephalic leukoencephalopathy with subcortical cysts also known as Van Der Knapp disease which is an inherited autosomal recessive disorder with a specific MRI feature and a variable clinical course. On MRI, Frontal and temporal subcortical cysts are the diagnostic hallmark. It usually presents with cerebellar, pyramidal signs and seizures. Megalencephaly can be detected early. It has been seen that it has been more prevalent in north Indian population, however it has been seen globally. We encountered a patient who is a 9 year old male child came with history of developmental delay, seizures with psychotic features. four patients and describe the clinical and radiological features of these patients.***

Keywords: megalencephalic leukoencephalopathy, subcortical cysts, Van der Knaap disease, rare encephalopathy, pediatric neurology

1.Case Report

A 9 year old male child referred to neurology department with history of developmental delay since 1 year of age, enlarged head size, multiple episodes of seizures with history of psychotic symptoms for last 4 years. Plain MRI with spectroscopy was ordered by the neurophysician and was referred to the radiological department for the same. It showed macrocephaly with diffuse T2W white matter hyperintensities in bilateral cerebral hemispheres with large subcortical cysts in bilateral anterior temporal lobes. On multivoxel MRI spectroscopy, there is increased choline peak with reduction in NAA (N-Acetyl aspartate). So, with these findings, the diagnosis of megalencephalic leukoencephalopathy with subcortical cysts was made.

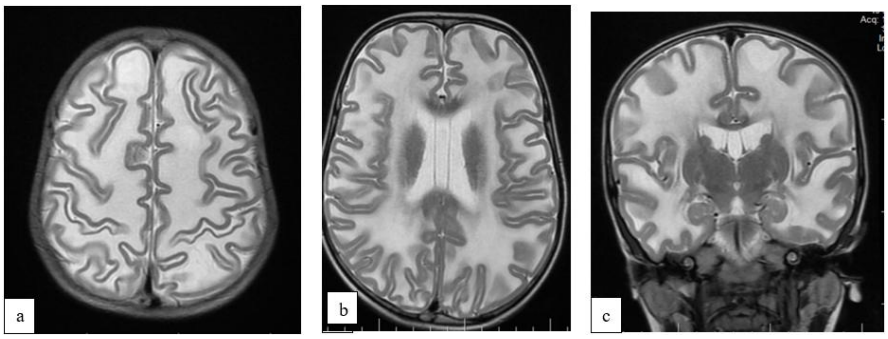

Figure 1: a), b) and c) T2W axial images and coronal showing diffuse white matter hyperintensity in bilateral cerebral hemispheres

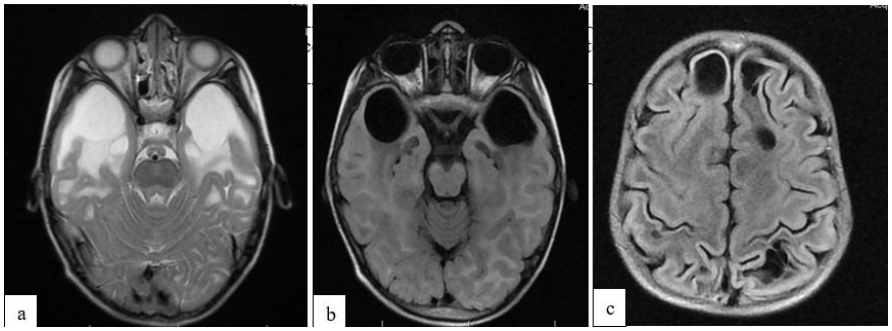

Figure 2: a), b) and c) T2W, FLAIR axial images and coronal showing subcortical cysts in bilateral anterior temporal lobes and frontal lobes

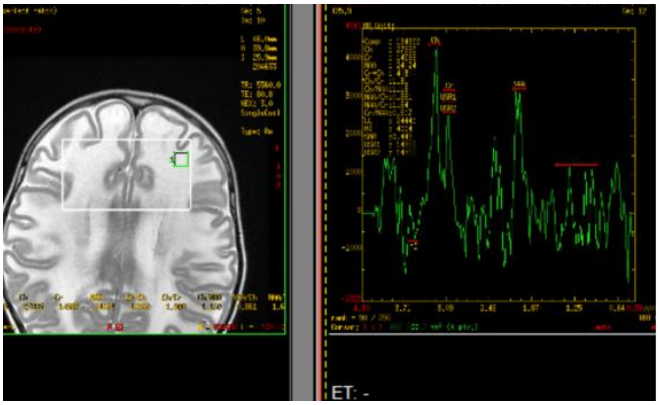

Figure 3: Single voxel MR spectroscopy shows elevated choline peak with decrease in NAA/ Cr and Choline/Cr ratios

2.Discussion

Description of Megalencephalic leukoencephalopathy (MLC) with subcortical cysts was first given by van der Knaap, et al. around 1995. It is a is an autosomal recessive disorder due to mutations in MLCI gene which has its locus in chr22qter with a low carrier rate. It has more occurrence in communities where consanguinity marriages are common. It is probably an integral membrane protein [1, 2]. The age at onset of symptoms ranges from birth to 25 years, with a median age at onset of 6 months.[3] MLC is known for its mild clinical course in the setting of abnormal MR findings. In MLC, macrocephaly may be present at birth or develop within the first year of life in all patients. Early development of the child can be normal or delayed.

Motor functions are detoriated slowly, whereas cerebellar ataxia and mild spasticity manifests in early childhood. As clinical course progresses, few patients may manifests with late extrapyramidal movement abnormalities with dystonia and athetosis. Mental decline occurs later as the age progresses. Mostly the patients present with epileptic seizures. [1, 4]

In typical cases, MR shows 'swollen white matter' along with diffuse symmetrical supratentorial white matter changes in the cerebral hemispheres with sparing of central white matter structures. Subcortical cysts are almost always present in the anterior aspect of temporal region and may be seen in frontoparietal region. There is relative sparing of the white matter. There may be increase in size and number of subcortical cysts. On MR spectroscopy, there is moderate decrease in NAA/ Cr and Choline/Cr ratios. [1, 4]

The differential diagnosis includes **Alexander disease, Canavan's disease, infantile-onset GM2 and GM1 gangliosidosis.** These conditions have progressive infantile onset leukoencephalopathies that are fatal within the first decade of life, however, MLC has a remarkably slow course in neurologic function. [1]

3.Conclusion

Large head size, ethnicity, pyramidal and cerebellar features, with a mild clinical course along with characteristic MRI features, are the key to diagnosing this disease in present clinical practice.

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