Megalencephalic Leukoencephalopathy with Subcortical Cysts

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INTRODUCTION

Megalencephalic Leukoencephalopathy is a rare entity which was first described by Van Der Knaap et al in 1995.[1] It is a progressive neurodegenerative disorder characterized by slow progression of mental deterioration. Megalencephaly sets in early in the disease usually in first year and can be more than 4 standard deviation above the mean. Seizures are present in almost all the patients. [2] We present typical MRI findings in a case of megalencephalic leukodystrophy with sub-cortical cysts.

Megalencephalic leukoencephalopathy with sub-cortical cysts is a rare entity that presents with macrocephaly and seizures. Mental deterioration is often mild with slow progression. Macrocephaly may be present at birth or develops in first year. Typical magnetic resonance imaging (MRI) findings help to clinch the diagnosis.

PRESENTATION OF CASE

A 12-year-old male patient presented with long history of generalised tonic clonic seizures, frequent falls and decreased school performance. On physical examination his head size was enlarged. MRI (Figure 1A-C) revealed abnormal signal in bilateral subcortical white matter appearing hyperintense on T2W and hypointense on T1W images. Cysts were seen in bilateral anterior temporal subcortical white matter. The boy belonged to Aggarwal ethnic community. Based on the characteristic clinical background and MR findings, diagnosis of Megalencephalic leukoencephalopathy with subcortical cysts was made.

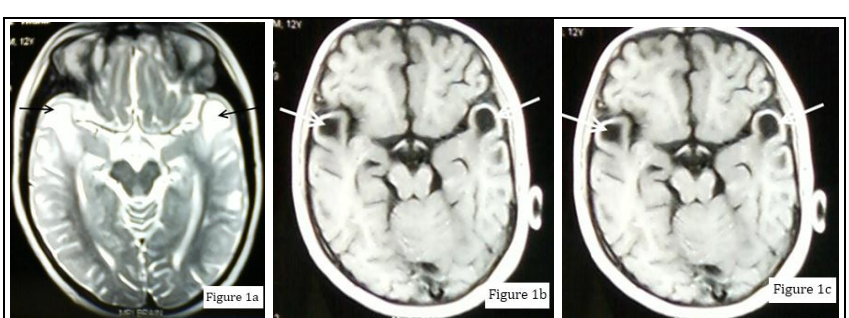

Figure 1. (A-C). Axial T2W (a) and T1W (b) Images Showing Subcortical Cysts in Bilateral Anterior Temporal Lobes (Black Arrows in 1a and White Arrows in 1b). Abnormal T1 Hypointensity in Subcortical White Matter in Bilateral Frontal Lobes (White Arrows in 1c).

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DISCUSSION

Initially it presents with macrocephaly in the first year of life which may normalise later on. Initially development is deceptively normal. However, with time, there is slow motor and mental deterioration. Seizures and extrapyramidal symptoms such as dystonia and athetosis may also be seen. There is only mild cognitive impairment. [2-4] On MR imaging, initially there is swelling of the subcortical white matter which progresses to characteristic subcortical cysts which are more prevalent in temporal lobes. There is relative sparing of central grey matter structures and brainstem. Characteristic MRI findings are sufficient for the diagnosis.[5,6] Over time, there is progressive enlargement of the size of the cysts and loss of white matter volume. Since its first description in 1995, many cases have been reported in the literature. Largest of them belong to Aggarwal community in India.[6,7] It is an autosomal recessive disorder. Clinical course may be static or slowly progressive.[4] There is no effective treatment. Antiepileptic's and physical therapy are the mainstay of the treatment.

CONCLUSIONS

Megalencephalic leukoencephalopathy with subcortical cysts is a rare condition. It should be suspected in any child with progressive neurological deficit and macrocephaly, especially those belonging to Aggarwal community. Awareness of characteristic MRI findings helps to clinch the diagnosis.

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