

Megalencephalic Leucoencephalopathy [Van Der Knaap Disease] in a Non Agarwal Family

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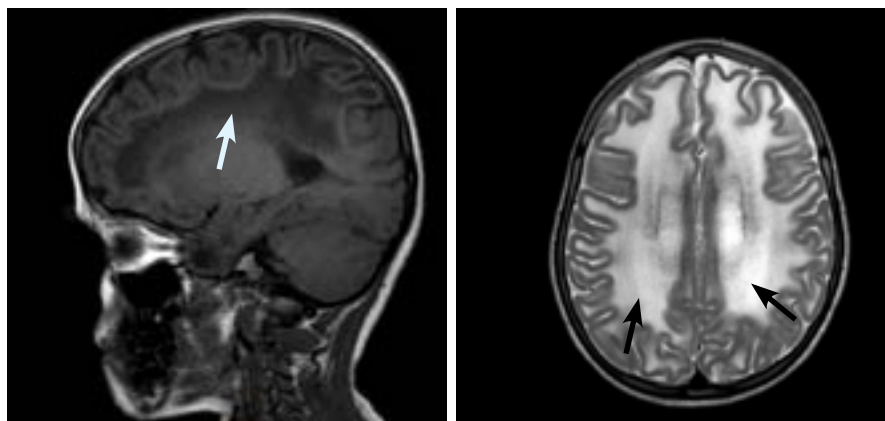


Fig. 1a and 1b: MRI BRAIN shows bilateral symmetrical white matter hypointensity on T1W sequence (Fig. 1a) and hyperintensity on T2 W sequence (Fig. 1b) suggestive of demyelination

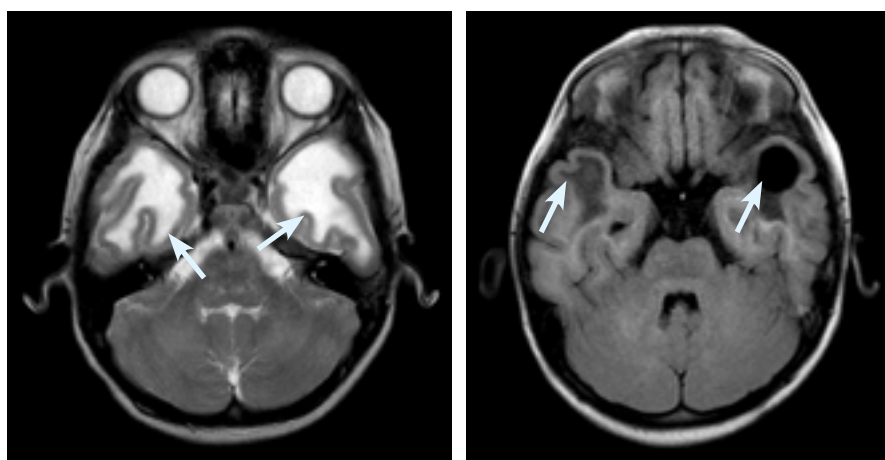


Fig. 2a and 2b: MRI BRAIN shows well defined symmetrical subcortical cysts were noted in anterior temporal lobe which are hyperintense on T2W (Fig. 2a) and suppressed in T2 Flair (Fig. 2b) sequence

A 13 year old girl born of non consanguineous marriage, full term normal delivery in a Hindu family from Tamilnadu. Her parents noticed progressive increasing of head size. She attained social smile by 6 months and head control by 7 months of age. She was not able to walk herself. Her younger brother who is 11 years old has similar complaints in the form of progressive increasing in head size with delayed mile stones.

On examination, there was macrocephaly with head circumference of 57 cms. Spastic weakness of both lower limbs and upper limbs was

present. All DTRS were brisk with extensor plantars. Sensory and cerebellar systems were normal. Abdominal and eye examinations were unremarkable. MRI BRAIN showed bilateral symmetrical white matter hypointensity on T1W sequence (Figure 1A) and hyperintensity on T2 W sequence (Figure 1B) suggestive of demyelination. MRI BRAIN also showed well defined symmetrical

subcortical cysts in the anterior temporal lobe which are hyperintense on T2W (Figure 2A) and suppressed in T2 Flair (Figure 2B) sequence consistent with a diagnosis of megalencephalic leucoencephalopathy. They were treated symptomatically along with physiotherapy.

M e g a l e n c e p h a l i c Leucoencephalopathy [Van der Knaap Disease] is a autosomal recessive disorder described by Van der Knaap in 1995. The new entity neurodegenerative disorder is characterised by infantile onset macrocephaly, cerebral leucoencephalopathy, mild neurological symptoms [pyramidal, extrapyramidal], slow course of functional deterioration, mental decline and seizures. MRI BRAIN shows diffuse symmetrical white matter demyelination with sparing of corpus callosum, internal capsule and brainstem with symmetrical subcortical cysts in the anterior temporal lobe¹. In India, majority of the patients belong to Agarwal community.² Our patient did not belong to this community. Indian patients with megalencephaly and MRI showing extensive demyelination with temporal cysts should raise the suspicion for MLC. This disease is linked to the gene, MLC1 and is localised on chromosome 22qtel.

This case highlights the new entity neurodegenerative disorder with a typical MRI finding in a non-Agarwal community.

References

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