Alexander’s Disease Presenting as Status Epilepticus in a Child

Sir,

Leukodystrophies in children can present with almost any neurological deficit.\(^1\) Though seizures are not uncommon in Alexander’s disease (AD), status epilepticus has not been reported.\(^2\) AD had been documented only once in a neurologically normal child.\(^3\)

A three-year old boy was evaluated following an attack of status epilepticus. He was born at term to non-consanguineous parents. He had attained developmental milestones normally. He did not have any learning disability. There was no family history of seizures or neurological impairment. At the age of three, he developed generalised tonic-clonic seizure lasting for 45 minutes, without fever. His head circumference was 54 cm (97\(^{th}\) percentile = 51.4 cm). He did not have any neurological deficits. Blood counts, serum electrolytes, renal and liver functions, blood glucose, serum aryl sulfatase and serum very long chain fatty acids were normal. Serum and cerebrospinal fluid lactate were also normal. Profile of aminoacids and organic acids in urine and serum was normal. Urine did not show metachromatic granules. EEG and evoked potentials (visual and auditory) were normal. He had normal nerve conduction in limbs. MR imaging of brain showed diffuse symmetrical signal intensity change in the white matter (sub-cortical and periventricular), which was hypointense in T\(_1\) and hyperintense in T\(_2\) weighted sequences. Changes were accentuated in frontal regions [Figure 1]. There were mild signal intensity changes in the basal ganglia, thalamus and brainstem. The cortex was spared. There was no enhancement after intravenous gadolinium. Two weeks later, he had two more brief complex partial seizures. Treatment was started with carbamazepine. He did not develop seizures till the last follow-up (2 years). There was no evidence of cognitive decline or neurological deficits.

Diagnosis of AD in our child was based on the presence of large head and typical MRI brain picture. He had three of the five MRI features (extensive white matter abnormalities which had frontal predominance, signal changes in basal ganglia, thalamus and brainstem) suggested by van der Knaap.\(^4\) Glial fibrillary acid protein (GFAP) gene is considered a reliable molecular marker for the diagnosis of infantile AD.\(^5\) But this genetic test was not available. The first neurological symptom in our child was status epilepticus at the age of three. Hence, he was in between the infantile and the juvenile types. Seizures are well described in the infantile and juvenile forms of AD.\(^1,2,5\) However, this is the first report of a child with AD, whose only neurological problem was seizure. First presentation as status epilepticus has not been reported before. There is a single reported case in the literature describing a three-year old boy who had growth failure and macrocephaly. He did not have any neurological symptom or sign. MR imaging of the brain showed features of AD, which was confirmed by biopsy.\(^3\) AD in adults can be asymptomatic.\(^6\) Our case and the literature suggest that AD can present with minimal or no neurological impairment. Any child with unexplained macrocephaly needs an MR imaging of brain, even in the absence of neurological signs, to detect unusual pathologies.

![Image](image.png)

**Figure 1**: MRI of brain (T2 W sequence) shows frontal dominance diffuse hyperintensity in the white matter involving also the U fibres. Caudate nucleus (right > left) shows hyperintense signals. Both thalami show hypointense signals. Brainstem is not shown in this cut.

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**References**


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