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Article

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Hemimegalencephaly without seizures: report of a case and review of literature

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Abstract: Hemimegalencephaly is a rare malformation of the brain characterized by enlargement of one cerebral hemisphere. The classic clinical triad consists of intractable epilepsy, severe psychomotor delay and hemiparesis. We report a case of a six months old girl, with the radiological features of hemimegalencephaly but with a comparatively benign clinical course. She had mild developmental delay but with no paresis or seizures. Literature revealed only two reported cases of hemimegalencephaly without the presence of seizures. We discuss the clinical and radiological findings of this third reported case.
Key words: Brain malformations hemimegalencephaly, seizures

Introduction

Hemimegalencephaly (HME) is a rare, sporadic type of congenital brain malformation, characterized by enlargement of one cerebral hemisphere. [1] HME is largely thought to be a malformation due to abnormal neuronal and glial proliferation. [2] This is accompanied by dysplasia of the cerebral cortex along with polymicrogyria, agyria/pachygyria, and white matter heterotopia, which results in intractable epilepsy and profound disabilities. [1] The classical clinical triad described is of epilepsy, severe psychomotor retardation and contralateral hemiparesis. [1,4] which are often severe in nature and associated with a significant mortality in infancy. [1,6] Onset of symptoms commonly occurs in the first week of life. [4] Early surgical intervention including hemispherectomy, either anatomical or functional is warranted in these patients to achieve a better outcome in the control of epilepsy and to promote psychomotor development. [3] However, there have been isolated reports of milder clinical presentation [4] including only a single case report of a child having HME without any presentation of seizures. [5] In this report, we describe a child who presented to our department for management of lumber meningomyelocele whose further workup revealed HME.

Case report

A six months old female child was referred to our neurosurgical department for evaluation and management of a lump in lower back that is present since birth along with macrocephaly. She was product of a full-term pregnancy of non-consanguineous parents; the prenatal history was normal and the baby cried immediately after birth. Height at birth was 48 cm and weight was 2500 g along with a head circumference of 41.2 cm with a flat anterior fontanelle along with lump at lower back. She had extra digits on both feet and had mildly delayed neurological development. She had no clinical evidence of paresis of any limbs neither she ever had a seizure or seizure-like episode as per 6 months of age. On examination, a head circumference over the 99th centile was noted in the child but there was no signs or symptoms of raised intracranial pressure. We plotted the head circumference on a growth chart when she was found to have no crossing of centile lines, consistent with normal growth of the child. She was found to have 6X4cm lumber meningomyelocele. Non-contrast CT and MRI of the brain and spine was obtained.

CT and MRI brain of the child demonstrated features typical of HME (Figure 1). Asymmetrical abnormalities were noted, with the left hemisphere larger in volume than the right. This asymmetry was associated with abnormalities both of grey and white matter. The left hemisphere demonstrated cortical malformation and polymicrogyria, as well as generalized loss of white matter bulk with cortical dysplasia. There was associated asymmetry of the ventricular system. Radiological features were consistent with HME. MRI spine was suggestive of lumber lipomenigomyelocele. She underwent sac excision and dural repair and postoperative stay was uneventful. The family was informed that the child did not require neurosurgical intervention for HME currently, but that she was at risk of developing epilepsy and should be closely observed for clinical seizures.

Discussion

HME is a rare anomaly and there is a paucity of large series in the literature. In one of the comprehensive reviews, the authors describe 14 cases, examining clinical, radiological and electrophysiological features. [4] In this series, the authors report that one patient did not have seizures. Beside this isolated case, extensive medline search revealed only one report where occurrence of HME without seizures was described, making the current case only the third reported.

Severe epilepsy is a hallmark of the condition, being present in all 10 patients described in another large series. [1] In fact, HME carries a significant mortality risk due to the severity of the epilepsy [6,7] as well as association with significant psychomotor delay and hemiparesis. The current case is unusual in only having one of the clinical triad (developmental delay) and only in mild form.

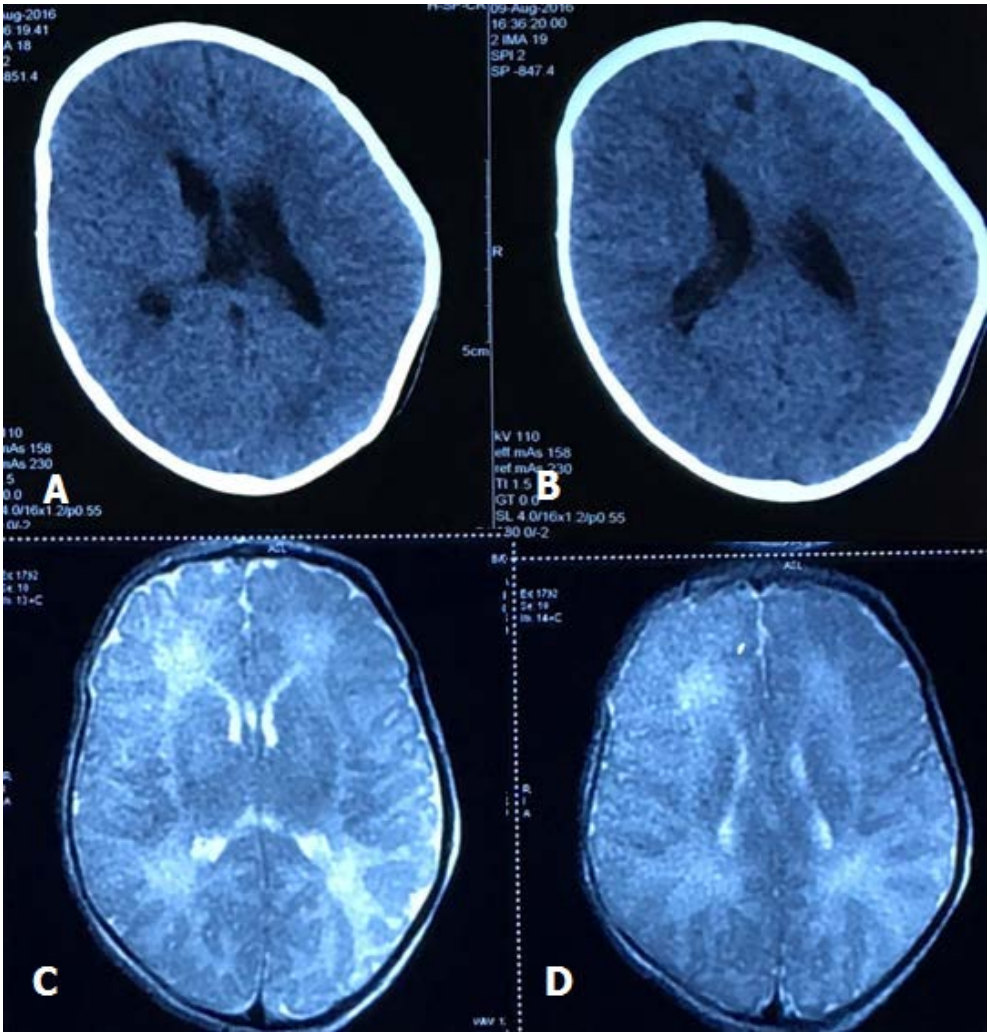

Figure 1 - Noncontrast CT (A & B) and T2W MRI brain (C & D) showing enlarged left cerebral hemisphere with ventricular dysmorphism, white matter changes, cortical dysplasia and polymicrogyria of the left frontal and parietal lobe

HME on brain imaging consists of cortical gray matter almost uniformly abnormal, areas of increased thickness of the cortical gray matter (GM), abnormal gyral patterns, increased signal intensity in the subcortical white matter (WM) on T2-weighted images, blurring of the GM-WM transition, atrophy or hemispheric hypertrophy or demyelination. [8] This patient presented almost all of these MRI findings supporting HME. The WM signal change may be consistent with either demyelination or advanced myelination. The ipsilateral ventricle is usually enlarged and dysmorphic, often with extension of the

posterior horn of the lateral ventricle across the midline. This patient did not have grossly enlarged ventricles but asymmetric.

Vigevano et al., [4] in their series of 14 patients, divided HME into two groups: A and B. Group A was characterized by severe, uncontrollable epilepsy with marked hemiparesis and delay (7/7), whilst group B had more mild clinical features with sporadic seizures and a later onset of epileptic attacks. Radiological features were less severe in group B, with no patient having microgyria on their scan. We consider the current case represents "group B"-type child, who is as yet too young to have had her first seizure. The other possibility is that there is a population of "silent" HME, with absent or mild clinical signs but with radiological features of the malformation and that this was an incidental finding when the child was being investigated for unrelated reasons. As availability of MRI scanning becomes ever more widespread, it may be that further similar cases are detected as children have brain imaging for various other reasons.

Conclusion

It is important to recognize the abnormal tissue organization to make the correct diagnosis. In children, brain MRI is the gold standard diagnostic tool, with brain enlargement and WM changes. Different degree of changes in signal intensity revealing the WM abnormalities is the most important and constant sign in HME. Regular follow up and close observation is required in scarce cases like the present one who do not manifest seizures on presentation for later development of epilepsy so that early surgical intervention may be undertaken.

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