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HEMIMEGALENCEPHALY WITH POLYMICROGYRIA – A CASE REPORT

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Abstract

Hemimegalencephaly on magnetic resonance imaging scan (MRI) consists of cortical gray matter almost uniformly abnormal, areas of increased thickness of the cortical gray matter (GM), abnormal gyral patterns, blurring of the grey-white matter transition, atrophy or hemispheric hypertrophy, demyelination, gliosis. We present a case of ten-year-old boy with a history of infantile spasms and developmental delay who presented to the pediatric neurology room with an episode of disinhibited behavior in family environment. An MRI was performed and isolated hemimegalencephaly with polymicrogyria of the right occipital lobe was diagnosed.

Keywords: electroencephalography, suppression-burst, hemimegalencephaly, magnetic resonance imaging

Introduction

Hemimegalencephaly (HME) is an uncommon anomaly of the brain characterized by one enlarged and dysplastic cerebral hemisphere with different degree of tissue impairment [1,2].

The contra-lateral cerebral hemisphere usually appears normal, except for being compressed [1].

It may be an isolated anomaly or associated with syndromes such as neurofibromatosis type I, tuberous sclerosis, epidermal nevus syndrome, proteus syndrome, unilateral hypomelanosis of Ito and Klippel-Trenaunay syndrome [3].

The etiology of HME remains unknown. It is generally assumed that HME results due to abnormal neuronal and glial proliferation or apoptosis [4].

Macroscopically, one hemisphere is enlarged and there is usually cortical dysgenesis, white-matter hypertrophy, and a dilated and dysmorphic lateral ventricle. The microscopic features include polymicrogyria (PMG), heterotopic grey matter, cortical dyslamination, cystic changes, blurring of the gray-white junction, and an increase in the number of both neurons and astrocytes [1].

The clinical triad of HMEG is typically: (a) intractable partial seizures from the neonatal period or early infancy, (b) hemiparesis and (c) developmental delay. The developmental outcome may be more positive if seizures are well controlled from an early age. Seizures are usually resistant to drugs and control may only be achieved by surgery such as anatomical hemispherectomy [4].

The EEG of HME is characterized by three main patterns of abnormal findings: triphasic complexes of large amplitude; unilateral, rhythmic “alpha-like” activity; and asymmetrical suppression-bursts characterized by “alpha-like” activity. The first pattern is associated with the most severe prognosis and the last one with a good prognosis [2].

MRI gives the best high-resolution structural image of the brain, and has whole brain coverage allowing symmetry and volume comparisons [2].

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Case report

A ten-year-old boy, previously diagnosed with infantile spasms at the age of 6 weeks, presented to the pediatric neurology room with an episode of disinhibited behavior in family environment with coprolalia suggestive for frontal disinhibition for several days. He was born after an uneventful pregnancy, except of a trauma of the mother who fell down the stairs with sacral impact. The birth weight was 3400 g, Apgar score = 9 and there were no signs of perinatal distress.

From the age of 6 weeks he began to have epileptic episodes with infantile spasms, with the appearance in cluster, on awakening or before falling asleep, 10-50 spasms/burst, lasting 10 to 15 minutes. The EEG findings show asymmetrical suppression-bursts characterized by "alpha-like" activity, which is the reason why a neurometabolic disease was suspected, like nonketotic hyperglycemia. Clinical examinations and laboratory tests had ruled out this diagnosis. He started the treatment with phenobarbital at a dose of up to 9 mg/kg/day, with no control of the seizures.

At the age of 4 months, the seizures recurred with focal character: onset with head and eyes deviation to the left, eyelid myoclonus, tonic contracture in extension of the limbs lasting 1-3 minutes and up to 7 seizures/day. On EEG recordings, focal hypsarrhythmia at the level of posterior derivations was present.

He received several antiepileptic associations. However, the seizures persisted until now, at the age of 10, regardless of the combination therapy associated with anxiety, hyperkinetic behavior, consciousness partially preserved, 10-20 seconds duration, up to 20 seizures/day. EEG recordings showed an aspect of electric status for the right occipital derivations, with a tendency to spread to the contra-lateral hemisphere.

In evolution, the neurological status of the patient consists of a left pyramidal syndrome associated with left eye divergent strabismus, horizontal nystagmus on left lateral gaze, central facial palsy of the left side. The psychological status of the patient evolved with moderate motor impairment, especially of the hand with intermittent neglection of the left upper limb. Also he presented autistic-like features: avoiding look in lateral angle, anxiety and hyperventilation episodes in unknown environment and disinhibited behavior in family environment with coprolalia, all of these suggestive for frontal disinhibition.

During his last admission a brain MRI was performed and it showed cortical dysplasia and mild enlargement of the right occipital lobe (Image A,E,I), white matter markedly increased in volume and isointense to cortical gray matter, consistent with graymatter heterotopia (Image B,D,F,G and H). The occipital gyri were presenting overfolding resembling polymicrogyria (ImageA, B and C). Moreover the gray-white junction appeared indistinct. There were no brain tumors and no enhancement of the dysplastic structure (Image D,E and F)

Also the ventricles were asymmetric and dysmorphic due to hypoplasia of the corpus callosum (Image J and K). Because of the enlarged hemisphere the midline can be displaced (Image D,E,F and K).

All these MRI findings supported isolated hemimegalencephaly with polymicrogyria of the right occipital lobe diagnose correlated with patient clinical history and evolution of his neurological and psychological status.

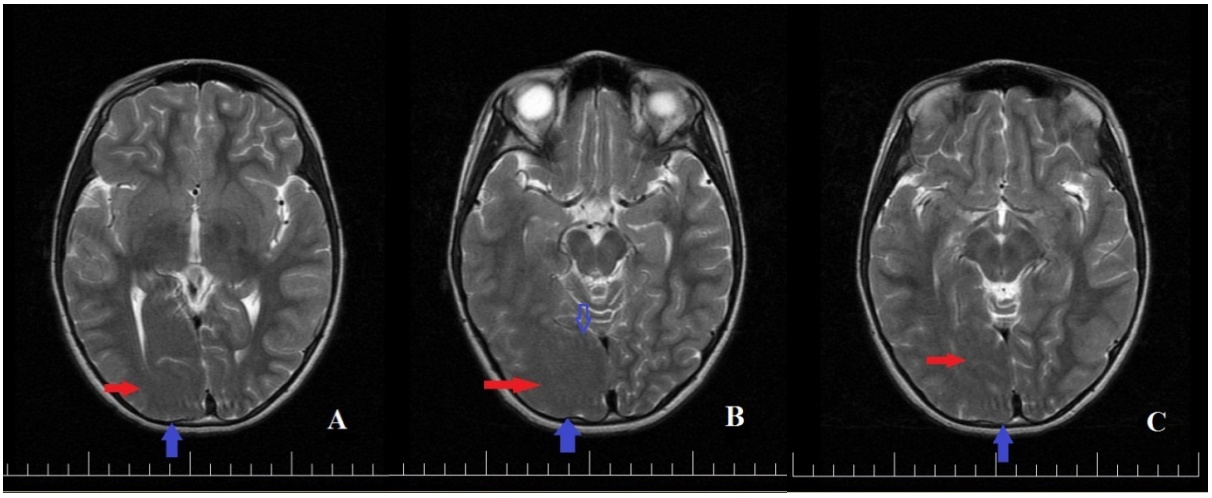

Image A, B and C. T2-weighted axial MRI of brain showing cortical dysplasia and mild enlargement of the right occipital lobe, white matter markedly increased in volume and isointense to cortical gray matter (red arrows) consistent with graymatter heterotopia. The occipital gyri may present polymicrogyria which can be confirmed only microscopically (blue arrows).

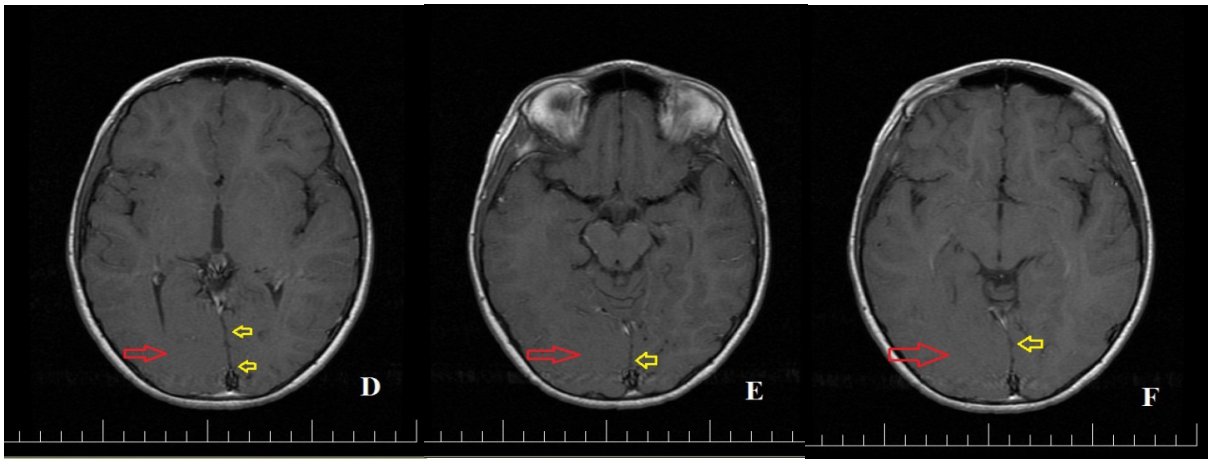

Image D,E and F. T1-weighted+contrast axial MRI of brain showing cortical dysplasia and mild enlargement of the right occipital lobe (red arrows) with mild displacement of the midline of the brain (yellow arrows) and no enhancement of the dysplastic structure.

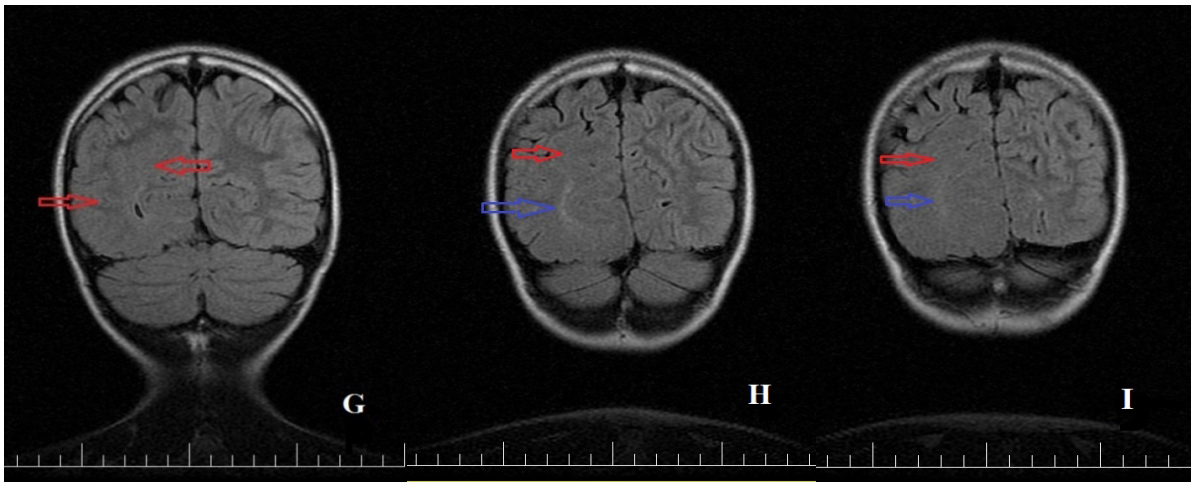

Image G,H and I. T1-weighted coronal FLAIR MRI of brain revealing white matter markedly increased in volume and isointense to cortical gray matter (red arrows) and increase signal intensity in the white matter (WM) consistent with dysmyelination (blue arrows).

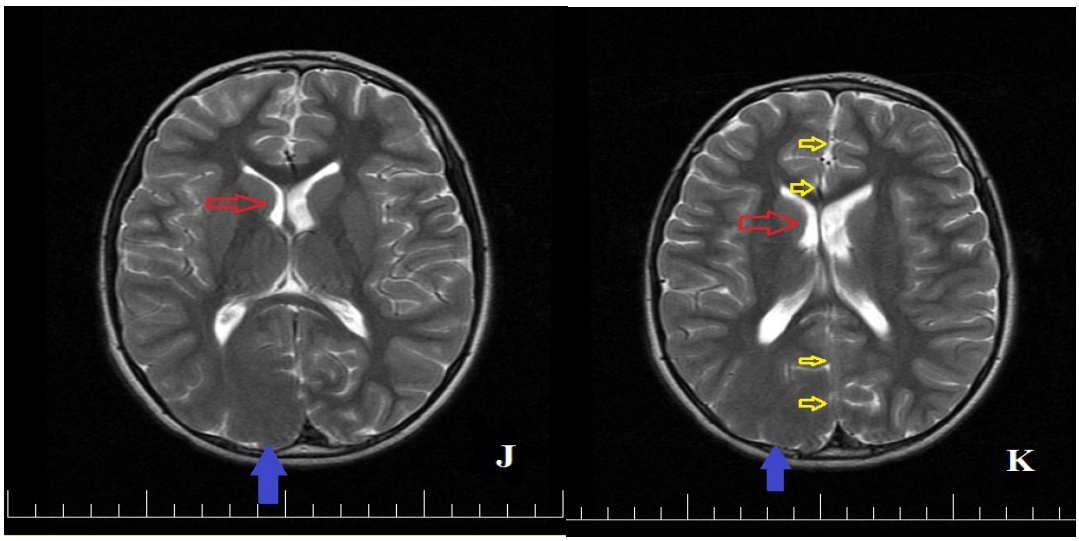

Image J and K. T2-weighted axial MRI of brain showing cortical dysplasia and mild enlargement of the right occipital lobe (blue arrows), asymmetric and dysmorphic right ventricle (red arrows) and mild displacement of the midline of the brain (yellow arrows).

Disscusion

This case shows peculiarities in terms of evolution of EEG: from asymmetrical suppression-bursts characterized by "alpha-like" activity to focal hypssarhythmia and focal electric status, and also the evolution of behavior with frontal disinhibition. Also this patient has been diagnosed in a very late phase of evolution of this disease. During diagnosis it hadn't been included in any syndrome because it presented only few symptoms and the kariotype wasn't performed. The poor financial and educational status of this patient's family did not allow an early diagnosis and unfortunately it has a very unfavourable outcome.

Intractable epilepsy is a common diagnosis in our country because of the lack of experience of radiologists in diagnosing cortical dysplasia with polimicrogirya on MRI.

Hemimegalencephaly on MRI 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 GMWM transition, atrophy or hemispheric hypertrophy, demyelination [4]. During development of the brain, the MRI signal may change. White matter is generally markedly increased in volume, and often contains tissue isointense to cortical gray matter, consistent with graymatter heterotopia. This patient presented almost all of these MRI findings supporting the hemimegalencephaly with polymicrogyria diagnose. The white-matter signal change may be consistent with either dysmyelination 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 enlarged ventricles but asymmetric (Image J and K). There may be enlargement of the ipsilateral cerebellar hemisphere and brain stem, an appearance which was named "total hemimegalencephaly." [1].

This pathology does not affect the whole hemisphere; in general, the posterior regions (parietal, occipital, and posterior aspect of the temporal lobe) are more frequently involved, compared with frontal or anterior aspect of the temporal lobes. About brain size, the characteristic HME might be compromised by atrophy related with a long history of epilepsy which is also the case here, with occipital lobe involvement that seems to have an only mild enlargement compared to the contra-lateral one.

However, any intractable epilepsy should be completely investigated during infancy especially using the MRI features for the correct diagnose of hemimegalencephaly with polymicrogyria and only after that the treatment started which consists of surgical resection of the affected area with fully recovery. In this particular case, it is kind of late for surgical treatment, but is the only way to treat the seizure. Anyhow, the recovery will not be complete because this patient's brain is fully developed and because the affected occipital lobe with all of the neuronal connections after surgical resection should be replaced somehow.

However, it is very important to recognize the abnormal tissue organization to make the correct diagnosis. In children, brain MRI became the gold standard diagnostic tool, with brain enlargement and WM changes. Different degree of changes in MRI T1 and T2 signal intensity reveal the WM abnormalities and are the most important and constant sign in HME. It is a real challenge because those signal abnormalities are related to gliosis and various degrees of destroyed tissue.

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