

Case Report

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Hypothalamic hamartoma with gelastic seizures: a case report

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ABSTRACT

Hypothalamic Hamartoma (HH) may have diverse clinical manifestations. Its hallmark association is with gelastic seizures. Gelastic epilepsy is characterized by episodes of loud, hollow, mirthless, stereo-typed, forced laughter. The patient may stare and giggle briefly without any other motor manifestations. Hypothalamic hamartoma is most often the cause of gelastic seizures. Here, authors report a case of gelastic seizure with hypothalamic hamartoma in a 14-month-old boy with an associated tonic clonic seizure. This case highlights the possibility of underdiagnosed hypothalamic hamartoma in younger age groups among pediatric population.

Keywords: Forced laughter, GABAergic, Gelastic seizures, Pituitary hamartoma, Precocious puberty

INTRODUCTION

Hypothalamic hamartomas are rare with an estimated prevalence rate of 1 in 50,000 to 1 in 100,000. Chances are that pediatrician, or a pediatric neurologist may not encounter one in his career. It may have a varied presentation, ranging from a catastrophic epilepsy in early childhood with severe learning difficulties and behavior problems, precocious puberty, to a milder form of gelastic epilepsy with little or no associated cognitive difficulties. The striking characteristic feature is the occurrence of the episodes of forced laughing, or gelastic seizure.^{1,2} Many cases may be unrecognized and may go undiagnosed, particularly if they are associated with other more severe seizure types.^{2,4} The aim of this report is to increase awareness of laughter as an ictal epileptic manifestation and highlight a case with an unusually benign course of hypothalamic gelastic seizure.

CASE REPORT

A 14-month-old boy of mongoloid race with apparently normal features reported to our centre with repeated

episodes of inappropriate laugh, blank stares and tonic-clonic seizures. He was born at term to a primi gravida with normal vaginal delivery and cried immediately at birth. He developed pathologic jaundice on the first day of life for which exchange transfusion was done on the second day of life. There was no obvious congenital anomaly, clinically. He was soon started on direct breast feeding.

Few days after the baby was born, he started having infrequent episodes of loud, forced laughter, which normally is not expected of his age. Each episode would last for few seconds with an associated blank stare, after which the baby normalizes and continued his expected activity for his age. At about 3 months of age, he often wakes up at night with the abnormal forced loud laugh of about 15 to 20 seconds duration, followed either by episodes of tonic clonic seizures which often lasts for a minute or blank stare with automatisms. CT brain showed no abnormality. EEG was done on two occasions and both were not suggestive of epilepsy. MRI brain done in centre showed hypothalamic hamartoma. The morphology was consistent with hamartoma and the

location was in the region of hypothalamus, particularly the mammillary bodies and the diagnosis of gelastic seizures with pituitary hamartoma was made.

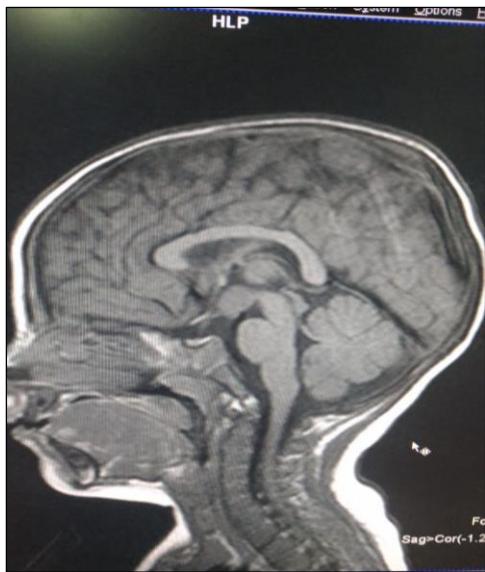


Figure 1: Sagittal showing the hamartoma.

Treatment was initiated with phenobarbitone at 6 mg/kg/day following which the tonic clonic episodes of seizure were controlled but the laughing episodes continue to remain refractory. His developmental quotient is normal for his age. Clinical neurological examinations were normal for his age. Hearing and vision assessments were normal.

DISCUSSION

Hypothalamic gelastic epilepsy is often a progressive epileptic seizure disorder. In the large majority of cases the laughter starts in infancy, becoming longer and more frequent over time with associated impairment of consciousness.^{1,2} Subsequently, patients develop seizures of various types: generalized tonic-clonic seizures, tonic seizures, drop attacks, and complex partial seizures. Additionally, progressive cognitive and behavioral impairment develops for most patients. More than half will suffer from precocious puberty.^{2,3} Hypothalamic hamartoma can be associated with a wide spectrum of epileptic conditions, ranging from a mild form with seizures characterized by urge to laugh and no cognitive involvement up to a catastrophic encephalopathy with early onset gelastic seizures, precocious puberty and mental retardation.¹⁻³ Moreover, a refractory, either focal or generalized, epilepsy develops during the clinical course in nearly all the cases. Neurophysiologic and neuroimaging studies have demonstrated that HH itself generates GS and starts a process of secondary epileptogenesis responsible for refractory focal or

generalized epilepsy.⁴ The intrinsic epileptogenicity of hypothalamic hamartomas may be explained by the neurophysiological properties of small GABAergic, spontaneously firing HH neurons. Surgical ablation of HH can reverse epilepsy and encephalopathy.⁵

CONCLUSION

Hypothalamic hamartomas are rare tumors associated with the triad of gelastic epilepsy, precocious puberty, and developmental delay. Gelastic seizure is often the first type of seizure that these patients develop, although other type of seizures is frequently present such as complex partial seizures and generalized tonic-clonic seizures. Progressive cognitive deficits, precocious puberty and the presence of important psychiatric comorbidity are widely described. Since these subsets of seizures are often refractory, invasive approaches are often required. The surgical procedure that has shown the best seizure outcome is the transcallosal approach but other nonsurgical procedures such as high frequency thermal coagulation are gaining popularity because of good results.

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