International Journal of Pharmaceutical and Bio-Medical Science

ISSN(print): 2767-827X, ISSN(online): 2767-830X

Volume 04 Issue 06 June 2024

Page No: 573 -575

DOI: https://doi.org/10.47191/ijpbms/v4-i6-14, Impact Factor: 7.792

Hypothalamic Hamartoma in an 11-Year-Old Female: Clinical Presentation, Diagnostic Evaluation, and Management Strategies – A Case Report and Literature Review

Kliti Pilika[1], Xhesika Xhetani[2], Anita Pilika[1], Aldo Shpuza[3]

[1]University Hospital Center "Mother Teresa", Tirane, Albania

[2]Korca Region Hospital, Albania

[3]Department of Public Health, University of Medicine, Tirane, Albania

ABSTRACT

Background: Hypothalamic hamartomas (HH) are rare, benign brain tumors that can cause severe neurological symptoms, including epilepsy. This report presents a case of an 11-year-old female with HH, highlighting the clinical presentation, diagnostic workup, and management strategies.

Case Presentation: An 11-year-old female with no prior medical history presented with generalized tonic-clonic seizures during sleep, characterized by convulsions, cyanosis, tongue biting, frothing at the mouth, and loss of consciousness lasting 1-2 minutes. Subsequent episodes included focal Jacksonian seizures involving the face and right eye myoclonus, progressing to generalized convulsions. Neurological examination and development were normal, with no family history of epilepsy. MRI revealed a sessile lesion in the left hypothalamus with T2/FLAIR hyperintensity and T1 hypointensity. Laboratory tests, including thyroid function, gynecological ultrasound, and various blood tests, were normal. A 48-hour video EEG indicated sensorimotor cortex involvement in both hemispheres, likely linked to the hypothalamic lesion.

Management: Initial management involved observation without antiepileptic medications. Due to persistent seizures, the treatment plan included oxcarbazepine, titrated to 1200 mg/day, and Keppra (levetiracetam) 1000 mg/day. Efforts were made to discontinue Keppra and maintain monotherapy with oxcarbazepine. Given the small size of the lesion and effective seizure control with medication, surgical intervention was not pursued.

Conclusion: This case underscores the importance of comprehensive diagnostic evaluation and individualized treatment plans for patients with HH. While medical management can be effective for small lesions, surgical options reported in the literature provide alternative strategies for larger or treatment-resistant cases. A multidisciplinary approach is crucial in managing the complex presentation of HH to optimize patient outcomes and improve quality of life.

KEYWORDS: Hypothalamic hamartoma, Pediatric epilepsy, Diagnostic evaluation, Medical management

ARTICLE DETAILS

Published On:
22 June 2024

Available on:
https://ijpbms.com/

INTRODUCTION

Hypothalamic hamartomas are congenital, non-progressive lesions in the hypothalamus that typically lead to multiple seizure types, cognitive decline, and psychiatric symptoms, and this activity reviews their evaluation, treatment, and management, highlighting the roles of interprofessional teams in patient care (1). One estimate, which may be conservative due to the difficulty in detecting hypothalamic hamartomas (HH), suggests that about 30,000 people worldwide have HH, with approximately 5% of these cases associated with Pallister-Hall syndrome (2). Rare diseases that affect the nervous system, especially in children, have started to be identified and reported in Albania, even though it is a small country in terms of population, and their absolute numbers are very small (3-5). To select the most appropriate treatment for each patient, unique clinical circumstances and lesion anatomy must be analyzed, with minor gelastic seizures often managed by observation, while more severe

cases may require antiepileptic medications, GnRH agonists, or various surgical options such as open resection, Gamma Knife radiosurgery, or thermoablation techniques to control seizures and associated symptoms (6–8). Hypothalamic hamartoma often requires a multidisciplinary team including pediatricians, neurosurgeons, neurologists, pediatric endocrinologists, nurses, pharmacists, and neuroradiologists for accurate diagnosis and comprehensive care.

The aim of this case report is to provide an in-depth review of the clinical presentation, diagnostic evaluation, and management strategies for hypothalamic hamartomas (HH), with a focus on a case study of an 11-year-old female. It also seeks to highlight the importance of a multidisciplinary approach in managing HH to optimize patient outcomes and improve quality of life.

CASE PRESENTATION

An 11-year-old female with no prior medical history presented with generalized tonic-clonic seizures during sleep, characterized by convulsions, cyanosis, tongue biting, frothing at the mouth, and loss of consciousness lasting 1-2 minutes, followed by post-ictal confusion. Initially, she was seen by a neurologist, who opted for observation without medication. Shortly thereafter, she developed another episode starting with focal Jacksonian seizures affecting the face and right eye myoclonus, progressing to generalized convulsions. Her neurological examination and developmental milestones were normal, and she performed excellently in school. She had a normal full-term pregnancy and delivery, with no perinatal events. Her parents were phenotypically healthy, and her 6-year-old brother was also healthy. There was no family history of epilepsy or other neurological disorders. MRI revealed a sessile lesion in the left hypothalamus, showing T2/FLAIR hyperintensity and T1 hypointensity compared to the periventricular white matter, with no significant enhancement post-contrast. (Figure 1)

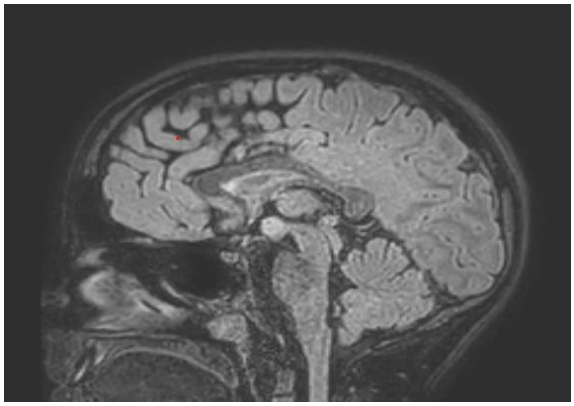

Figure 1: Sessile lesion on the left side of the hypothalamus with a hyperintense signal on T2/FLAIR and a hypointense signal on T1 compared to theperiventricular white matter, without significant enhancement after contrast agent injection.

Laboratory tests, including thyroid function, gynecological ultrasound, hand X-ray, and various blood tests, were normal.

(Figure 2)

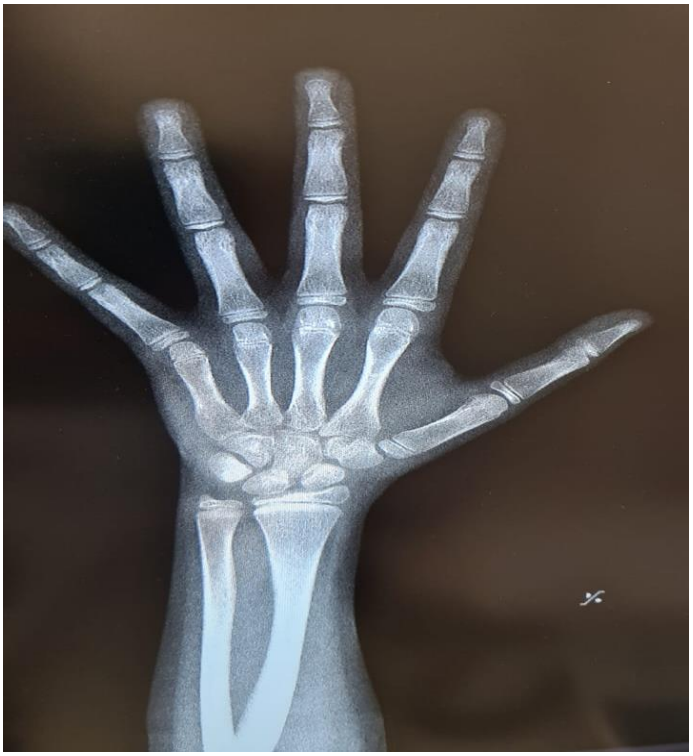

Figure 2: Hand X-ray shows normal bone maturity for the age.

A 48-hour video EEG suggested involvement of the sensorimotor cortex in both hemispheres, likely related to the hypothalamic lesion. The initial management strategy involved observation without antiepileptic medications. However, due to persistent seizures, the treatment plan included oxcarbazepine, titrated to 1200 mg/day, and Keppra (levetiracetam) 1000 mg/day. Efforts were made to discontinue Keppra and maintain monotherapy with oxcarbazepine. Given the small size of the lesion and effective seizure control with medication, surgical intervention was not pursued. Literature reports various surgical options, including Gamma Knife Radiosurgery, Stereotactic Thermoablation, Transcallosal Interforniceal Resection, Transventricular Endoscopic Resection, and Pterional Resection, which may be considered for larger or refractory cases.

DISCUSSION

The case of an 11-year-old female with hypothalamic hamartoma (HH) presented here provides significant insights into the clinical presentation, diagnostic evaluation, and management strategies for this rare condition. She presented with generalized tonic-clonic and focal Jacksonian seizures, highlighting the complexity and diagnostic challenges of this rare condition. Literature has long reported the varied

presentations of hypothalamic hamartomas, as evidenced by cases with precocious puberty, epileptic laughter, and abnormal behavior, and other presented with cerebral seizures, as in our case (9).

Diagnostic workup included MRI, which revealed a sessile lesion in the left hypothalamus, and a 48-hour video EEG showing sensorimotor cortex involvement, essential for accurate diagnosis. Another study using EEG-fMRI revealed activation and/or deactivation in subcortical structures and neocortices in all patients, with 6 out of 8 patients showing activation in or around the hypothalamus with the HH interface before spike onset (10). Initial management involved observation without medication, but due to persistent seizures, the patient was treated with oxcarbazepine and levetiracetam, demonstrating the necessity of individualized treatment plans. In different cases literature reports various surgical options, including Gamma Knife Radiosurgery, Stereotactic Thermoablation, Transcallosal Interforniceal Resection, Transventricular Endoscopic Resection, and Pterional Resection, which may be considered for larger or refractory cases (11). The rarity and complexity of HH require a careful, tailored approach to treatment, as demonstrated by the successful management of seizures in this patient without the need for immediate surgical intervention. This case underscores the importance of a comprehensive diagnostic evaluation and highlights the potential for effective seizure control with medical management in select HH cases.

CONCLUSION

This case underscores the importance of comprehensive diagnostic evaluation and individualized treatment plans for hypothalamic hamartomas (HH). Effective seizure control was achieved with medical management in this 11-year-old female, avoiding the need for immediate surgical intervention. However, for larger or refractory cases, various surgical options exist, emphasizing the need for a multidisciplinary approach to optimize patient outcomes and improve quality of life. Future research should focus on refining treatment strategies and exploring new modalities for managing HH.

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