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**Radiology**

# A Rare Cause of Epilepsy: Hypothalamic Hamartoma

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**Abstract** **Case Report**

Hypothalamic hamartomas (HHs) are rare congenital malformations of the hypothalamus. They are usually responsible for central precocious puberty and developmental delay and remain the leading cause of gelastic seizures. HHs are usually diagnosed in early childhood. This case report presents a rare case of HH in an adult. A 22-year-old patient with a history of drug-resistant epilepsy. The diagnosis of hypothalamic hamartoma (HH) was made on MR imaging. This case highlights the role of MR imaging in determining the etiology of seizures when the clinical history and EEG findings are nonspecific. MRI is also important in differentiating HH subtypes and providing clinicians with detailed anatomic information, especially when surgery is planned.

**Keywords**: Hypothalamic hamartoma, MRI, gelastic seizure.



## INTRODUCTION

Cerebral hamartomas are congenital brain malformations and tumor-like masses. Histologically, they are characterized by a disorganized arrangement of mature neural elements. They are a rare cause of medically intractable focal epilepsy [1]. These tumor-like masses are most commonly located in the temporal and frontal lobes and rarely in the hypothalamus [1]. Hypothalamic hamartoma (HH) is a rare entity often located in the ventral region [2, 3]. Its prevalence varied from 1 in 500,000 to 1 in 100,000 [4]. HH commonly presents with gelastic seizures in childhood and adolescence. However, other forms of partial complex or generalized tonic-clonic seizures may also occur in these patients [5]. Magnetic resonance imaging is the mainstay in the diagnosis of HH and in neurosurgical evaluation when patients diagnosed with HH develop resistance to antiepileptic drugs [5].

## CASE REPORT

A 22-year-old woman with no medical or family history of epilepsy, no history of fever or head trauma presented with generalized tonic-clonic seizures that have developed since she was 9 years old. Recently, these seizures have become more frequent, with a maximum frequency of once per day. Because of the increasing frequency of the seizures, the patient and her parents sought medical advice from a general practitioner and were then referred to the University Hospital Center for treatment. The patient presented a delay in reaching age-appropriate developmental milestones, she had difficulty at school and she was unable to converse normally, could not read or write fluently. The parents reported that since the age of 9 they had noticed persistent laughing behavior, especially during sleep, consistent with gelastic seizures.

Pre-ictally, the patient would have a brief episode of laughter that would then progress to a tonic-clonic seizure lasting a few seconds. Post-ictally, she was usually drowsy for a few minutes. On clinical examination, she had no neurological deficits. Her physical appearance was normal and she had no dysmorphic facial features. Her height and weight were normal. The EEG showed nonspecific intermittent focal epileptic discharges. The patient was treated for generalized tonic-clonic seizures and two types of antiepileptic drugs were prescribed. Brain MRI was performed and showed a mass within the third ventricle. Its base was located at the floor of the third ventricle near the mammillary bodies in projection to the hypothalamic region. The lesion had gray matter-like signal on both T1- and T2-weighted images without enhancement after intravenous gadolinium administration (Figure 1).

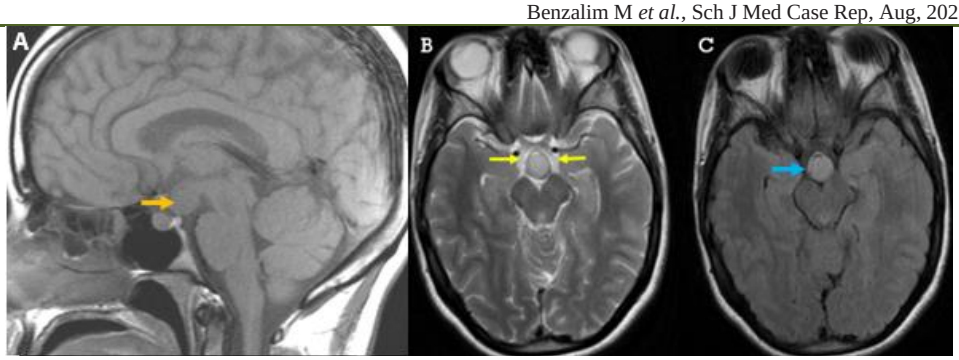


**Figure 1: (A) Sagittal T1-weighted image shows a well-defined mass attached to the floor of the third ventricle extending into the suprasellar cistern (solid yellow arrow) with isosignal to gray matter. (B and C) Axial T2-weighted and FLAIR images show the location of the lesion in the interpeduncular cistern with mass effect on the optic radiations (yellow and blue arrows)**

The lesion extended into the suprasellar cistern. The lesion has had a mass effect on the optic chiasma and the pontine cistern (Figure 2). The diagnosis of hypothalamic hamartoma was made on the basis of clinical findings and specific features on magnetic resonance imaging of the hypothalamic region.

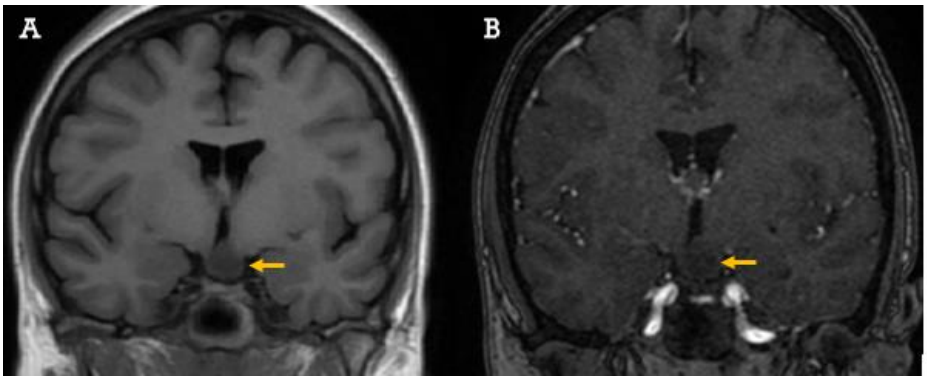


**Figure 2: (A) T1-weighted coronal image showing a pedunculated suprasellar lesion. (B) Post-contrast coronal image shows a non-enhancing hypothalamic hamartoma**

Clinical improvement and a decrease in epileptic seizure frequency were noted. Management consisted of medical treatment and periodic imaging surveillance of the hypothalamic hamartoma. Follow-up of the case for two years showed good clinical improvement, with seizure frequency decreasing to once per week. MR imaging surveillance showed a stable appearance of the hypothalamic mass with no increase in size or compression of adjacent structures (Figure 3). Conservative management based on dual antiepileptic therapy was initiated. Surgical treatment was postponed in view of the patient's clinical and radiological stability.

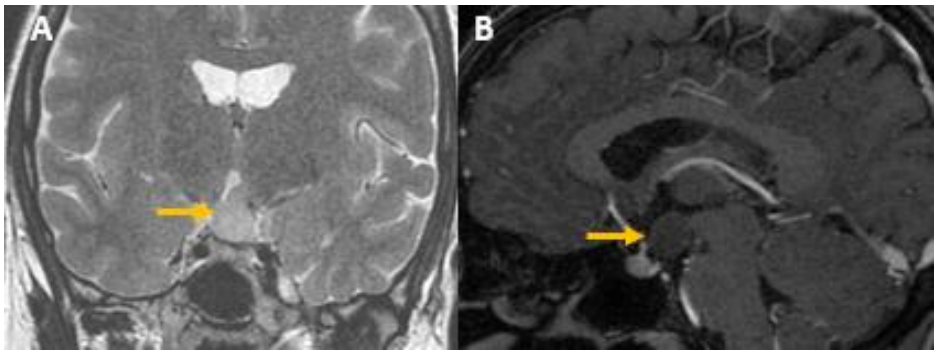


**Figure 3: (A) coronal T2 image and (B) Post-contrast coronal T1-weighetd images through the mass 2 years later, show no size changes of the HH**

## DISCUSSION

Hamartomas are mesenchymal lesions that can be found in any organ system. Hypothalamic hamartomas are gray matter malformations composed of hyperplastic neurons of various sizes. They are located at the base of the brain in the third ventricular floor, between the infundibular recess and the

mammillary bodies. Their prevalence varies from 1 to 2 cases per 100,000 population in children and adolescents. They are often manifested by endocrine and neurological symptoms [5, 6]. Based on the anatomic features, morphologic appearance, and clinical symptoms, two clinical phenotypes have been described. Hypothalamic hamartomas of the posterior hypothalamus and mammillary bodies associated with epilepsy, particularly gelastic and drug-resistant seizures and beginning in infancy, like it was in our case. The second prototypical HH is attached to the tuber cinereum in the anterior hypothalamic region. It is often associated with central precocious puberty [3]. In addition to gelastic seizures, many patients have other comorbidities such as developmental delay, psychiatric symptoms, cognitive problems, and behavioral disturbances [3, 6]. Asymptomatic HH is extremely rare [2]. The age of diagnosis of HH varies from birth to childhood [7]. In our case, the age of diagnosis was late at 20 years.

Epileptic seizures associated with hypothalamic hamartoma may show different types of electroclinical patterns [8]. EEG abnormalities are highly variable [9]. Because of the deep location of HHs and the complex neuronal connections with adjacent brain structures, EEG has limited diagnostic sensitivity and specificity [5]. In the interictal phase, there may be a slowing of the basic rhythm, theta or delta slow wave overload, or images of spikes and spike waves, isolated or in short bursts of variable localization, frontal, temporal, less commonly occipital. These abnormalities increase during hyperpnea and sleep, which may produce continuous peak-wave activity during sleep. However, the EEG may be normal, leading to a delay in diagnosis. Therefore, the diagnosis of HH is based on MRI [9].

MRI plays a critical role in the diagnosis of HH and differentiation of its subtypes [5]. MRI is superior to CT in detecting hypothalamic hamartomas, especially small ones [5]. It shows a well-defined mass whose signal is identical to that of the gray matter [9]. HHs are isosignal on T1-weighted MR images with homogeneous high signal on T2-weighted images [5, 9]. The signal is not enhanced after intravenous administration of gadolinium-based contrast agents [5,9,10]. In the absence of prior surgical therapy, any contrast enhancement must suggest another pathological diagnosis [3]. The main MR imaging features of hypothalamic hamartomas are high T2-weighted signal intensity compared to normal gray matter, absence of contrast enhancement, and no significant increase in lesion size [3, 5]. Cystic portions within hypothalamic hamartomas have been described in 2-3% of cases. They are more common in large lesions. Arachnoid cysts may also be associated with HHS, extending into the prepontine cistern, middle cranial fossa, or even the prechiasmatic space [3]. Midsagittal views are important to differentiate between sessile and pedunculated forms of hypothalamic hamartoma [5, 10]. The sessile form has a base of attachment within the third ventricle, which may be partial or complete. If the hamartoma is attached to the floor of the third ventricle by a pedicle, it is a pedunculated one. The differential diagnosis on imaging includes hypothalamic glioma, optochiasmatic glioma, craniopharyngioma, and germinoma [11, 12]. The presence of calcifications, cysts, positive enhancement, volume changes, and different signal intensity on MR imaging can differentiate these tumors from HH [12].

The majority of patients with hypothalamic hamartoma are resistant to antiepileptic drugs at presentation or during subsequent follow-up. Patients usually require multiple medications to control seizures [3,5]. However, surgical treatment, especially total removal of HHs, remains the modality of choice to achieve good seizure outcomes and behavioral improvement. Several surgical modalities have been described in the literature [13]. The different options for HH treatment are microsurgery through trans-callosal, interfemoral, pterional or orbitozygomatic approaches, stereotactic guided endoscopic surgical resection, gamma knife surgery, stereotactic radiofrequency and thermocoagulation [13, 14]. The indication depends on the size of the tumor, its location in the third ventricle and the age of the patient [14]. In our case, satisfactory seizure control was achieved with two antiepileptic drugs. Surgery should be considered if the patient develops drug resistance, severe cognitive impairment, or behavioral problems.

## CONCLUSION

Hypothalamic hamartomas are rare congenital malformations. Gelastic seizures are the main symptom that these patients develop. However, complex partial seizures, generalized tonic-clonic seizures, drop seizures, and infantile spasms may also be present. Progressive cognitive deficits and significant psychiatric and behavioral comorbidity have been reported. MRI is the modality of choice for the diagnosis and follow-up of HH. It must be performed early to detect and classify HH. MRI findings are highly suggestive for diagnosis, especially similar signal intensity to gray matter, no contrast enhancement, no significant increase in lesion size. Resistance to antiepileptic drugs is common at the onset or during the disease. Therefore, surgery remains the more effective therapeutic modality.

**Conflict of interest**

The authors report no conflicts of interest in this work.

**Patient consent**

Informed consent was obtained from the patient's mother for the publication of this case report.

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