Large Hypothalamic Hamartoma With Calcification and Cystic Components in an Adult—Case Report—

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Abstract

A 24-year-old female presented with an unusual case of hypothalamic hamartoma manifesting as seizure. Neuroimaging findings were atypical, showing the large tumor (maximum diameter, 50 mm) with a cystic component and calcification. Surgery was performed and histological examination demonstrated heterotopia. Hamartoma should be considered in the differential diagnosis of a suprasellar, non-enhanced mass attached to the hypothalamus. Excessive unnecessary surgery should be avoided, and intraoperative pathological examination may lead to enhanced assessment and better outcomes.

Key words: adult-type hypothalamic hamartoma, seizure, heterotopia, calcification, cystic component

Introduction

Hypothalamic hamartoma is a non-neoplastic, congenital mass consisting of heterotopic neural tissue that commonly manifests as gelastic seizures and/or precocious puberty in infants. Patients typically present with developmental disturbances because of uncontrollable seizures. Computed tomography (CT) and magnetic resonance (MR) imaging usually show hypothalamic hamartomas as small (diameter, 10–30 mm) round masses that are connected to the hypothalamus, with almost identical signal intensity to that of brain parenchyma, with clear demarcation, and no contrast enhancement. Hypothalamic hamartomas do not usually develop cysts or calcifications. Morphologically, pedunculated types of hypothalamic hamartomas are typically associated with precocious puberty, whereas sessile types of hypothalamic hamartomas are associated with seizures. Precocious puberty in patients with hypothalamic hamartoma is treated using a luteinizing hormone-releasing hormone analogue, and hypothalamic hamartoma is considered to be epileptogenic and is likely to cause gelastic seizures. Other secondary lesions may also assume epileptogenic properties that cause various types of seizure patterns. Seizures may be controlled by medication, but surgical treatment must be considered for uncontrollable seizures. Endoscopic disconnection between the hypothalamic hamartoma and the hypothalamus is based on the hypothesis that epileptic waves are initiated from the hypothalamic hamartoma and diffuse to the hypothalamus. Other types of surgeries have varying outcomes, but morbidity is considered high because of the position of the tumor.

Here, we report a case of a large hypothalamic hamartoma in an adult with atypical neuroimaging and physical features.

Case Report

A 24-year-old female was admitted to a hospital because of sudden loss of consciousness and a tonic-clonic seizure. She showed no signs of developmental disturbances, led a normal life, worked as a clerk in a bookstore, and was engaged to be married. She had been treated for depression 5 years prior to the current episode but had been stabilized with medication. Neuroimaging examination detected a suprasellar mass, and the patient was subsequently referred to our hospital.

On admission, her state of consciousness was clear. She showed bitemporal light sensitivity depression on automatic static perimetry. Neither pituitary endocrine dysfunction nor elevation in luteinizing hormone or follicle-stimulating hormone levels was observed. The patient and her father had polycystic kidneys, but her current kidney function was normal. We administered valproic acid (400 mg/day) as an anticonvulsant.

Radiography demonstrated expansion of the sella turcica (Fig. 1). CT revealed a large suprasellar mass with a cystic component and a high density lesion (Fig. 2A), but without enhancement by contrast material (Fig. 2B). MR
imaging showed the tumor as an isointense mass compared with gray matter on T1-weighted imaging and as an iso- to hyperintense mass on T2-weighted imaging, which was not enhanced by gadolinium. The boundary between the hypothalamus and the tumor appeared obscure (Fig. 3A). The pituitary stalk and gland were compressed caudally, and a cystic component was present within the tumor (Fig. 3B). Angiography showed the bilateral arterial segments C2 through A1 were elongated by tumor compression. No tumor stain was visible, but an aneurysm was detected at the bifurcation of the left posterior communicating artery.

Surgery was performed via an interhemispheric approach. The tumor appeared to consist of cerebral cortical tissue (Fig. 4A). Both the optic nerves were strongly compressed (Fig. 4B). The border between the tumor and the optic nerves was clear. Intraoperative pathological examination indicated non-neoplastic neural tissue. We could not confirm involvement of the chiasma and optic tract because of the size of the tumor. On the basis of the condition of the optic nerves, we thought that the optic chiasma and tract were cranio-dorsally compressed by the tumor. Exercising the greatest care not to worsen the visual disturbances, we achieved the maximum possible decrease in the tumor mass under neuronavigational guidance (Figs. 3C and 4C). The final histological diagnosis was neural heterotopia (Fig. 5).

The patient tolerated general anesthesia, recuperated...
Hypothalamic hamartoma on CT. Furthermore, histologically, a tissue, suspected of being calcification, was observed in the area, being present on CT and MR imaging, and a high density was present on CT and MR imaging, and a high density enhancement by gadolinium. A possible type craniopharyngioma. Solid-type craniopharyngioma must be differentiated from low-grade glioma and solid-type hypothalamic hamartoma.6) Hypothalamic hamartoma shows an enhanced mass with absence of calcification and cyst formation. MR imaging shows hypothalamic hamartomas as isodense, non-enhanced masses on T1-weighted imaging, without enhancement by gadolinium. CT shows hypothalamic hamartomas as isodense, non-enhanced masses with absence of calcification and cyst formation. MR imaging shows hypothalamic hamartomas as isointense masses on T1-weighted imaging and iso-enhanced masses on T2-weighted imaging, without enhancement by gadolinium. In our case, the hypothalamic hamartoma was relatively large, with a maximum diameter of 50 mm.

Neuroimaging examination of hypothalamic hamartoma typically reflects the findings of neural heterotopia. CT shows hypothalamic hamartomas as isodense, non-enhanced masses with absence of calcification and cyst formation. MR imaging shows hypothalamic hamartomas as isointense masses on T1-weighted imaging and iso-enhanced masses on T2-weighted imaging, without enhancement by gadolinium. A possible reason is that the blood vessels of the hypothalamic hamartoma retain the blood-brain barrier. The pituitary gland and stalk are relatively easy to identify on MR imaging with contrast medium, even if compressed by a large hypothalamic hamartoma. Hypothalamic hamartoma must be differentiated from low-grade glioma and solid-type craniopharyngioma. Solid-type craniopharyngioma appears as a relatively hypointense mass on T1-weighted imaging compared with hypothalamic hamartoma. The presence of calcifications and cyst formation commonly contradicts the diagnosis of hypothalamic hamartoma. However, in the present case, a cystic component was present on CT and MR imaging, and a high density area, suspected of being calcification, was observed in the hypothalamic hamartoma on CT. Furthermore, histological examination confirmed the presence of calcification.

Discussion

Hypothalamic hamartoma is commonly observed as a small round mass with clear demarcation. The diameter of hypothalamic hamartomas ranges between 5 and 50 mm but is generally approximately 10–30 mm. Eight hypothalamic hamartomas treated by gamma knife surgery ranged from 8 to 22 mm, and 14 hypothalamic hamartomas treated by surgery because of uncontrollable seizure measured 10–42 mm. Some childhood cases of relatively large hypothalamic hamartoma have also been reported. 14,19,20 In our case, the hypothalamic hamartoma was relatively large, with a maximum diameter of 50 mm.

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Calcification in hypothalamic hamartoma was previously unknown. Calcification is basically a nonspecific degenerative change in old lesions that may have occurred due to various causes. The minerals in the plasma adhere to the tissue via osteopontin or osteonectin. Calcifications are observed in old infarcts or in other degenerative changes in the brain. Macrophages secrete osteopontin, and infiltrate into lesions where ischemic or other inflammatory changes occur and initiate the calcification process. In the present case, calcification was seen to be complete, and the underlying mechanisms are difficult to elucidate. However, we think that a similar mechanism, via osteopontin or osteonectin, may be responsible in this case. A case of thalamic hamartoma with calcification has been reported. Moreover, certain cases of cyst formation in hypothalamic hamartomas are known. The mechanism of cyst formation is considered to involve ischemic change followed by liquefactive necrosis. A large hypothalamic hamartoma that could cause circulation disturbances may show the presence of calcification and cyst formation. Hypothalamic hamartoma should not be excluded based on the neuroimaging findings of calcification or cystic components.

Treatment of the present case needed to consider the diagnosis, seizure activity, and visual disturbances. Glioma and solid-type craniopharyngioma were also considered as differential diagnoses because of the neuroimaging findings. Non-neoplastic neuronal tissue was confirmed by intraoperative pathological diagnosis which lead to the minimum invasiveness of the treatment. The seizure for which the patient was admitted was the first attack. Hypothalamic hamartomas have better prognoses in adults than in children. Assessment of 14 adult-type hypothalamic hamartomas manifesting as seizures in adulthood found that patients with adult-type hypothalamic hamartomas showed milder abnormal behaviors and fewer learning disabilities than those showed by pediatric patients. Many of the patients were able to work, marry, and lead normal lives. Their seizures were easy to control with medication and because of their high level of social and cognitive function, such patients can treated by minimally invasive surgery or drug therapy. Our patient manifested with seizure in adulthood, so we expected satisfactory seizure control with medication.

Removal of a tumor only for seizure control is considered to have high morbidity. Since visual disturbance was also present, we performed partial removal of the tumor, with excision of as much tumor tissue as possible. The optic nerves were strongly compressed by the tumor. We could not confirm the condition of the optic chiasma and tract because of the tumor size. We predicted that these components were compressed crano-dorsally in the direction of the optic nerve. We performed partial removal of the tumor under neuronavigation guidance to reduce the compression on the optic nerve, chiasma, and tract. The patient achieved full recovery from her visual disturbance after surgery. Intraoperative electroencephalography would have been ideal for understanding the mechanism of the seizure in this case, but the partial destruction of the tumor may enable this patient to live.
normally with satisfactory control of the tumor effects. The present adult case of large hypothalamic hamartoma manifesting as seizure showed atypical neuroimaging findings, including calcification and a cystic component. Hamartoma should be considered in the differential diagnosis of a suprasellar, non-enhanced mass attached to the hypothalamus. Excessive unnecessary surgery should be avoided, and intraoperative pathological examination may lead to enhanced assessment and better outcomes.

References


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