JBR–BTR, 2013, 96: 362-364.

HYPOTHALAMIC HAMARTOMA

L. Topff[1], G. Gelin[1]

Hypothalamic hamartomas are rare congenital malformations presenting with central precocious puberty, gelastic epilepsy and developmental retardation. We report a case of an 11-month-old girl with recurring bloody vaginal discharge. Diagnosis of hypothalamic hamartoma was made on the basis of clinical findings and specific features on magnetic resonance imaging of the hypothalamic region.

Key-word: Hamartoma.

Hypothalamic hamartomas are rare congenital malformations presenting with central precocious puberty, gelastic epilepsy and developmental retardation. The disease manifests early in life and may occur in neonates. The prevalence is estimated as high as 1 in 50.000-100.000 (1).

The diagnosis of hypothalamic hamartomas is based on clinical and radiologic features. The use of MR imaging has increased the incidence of these lesions. In this article, we present a case of hypothalamic hamartoma and review its radiologic characteristics through published cases, with an emphasis on MR appearance.

Case report

An 11-month-old girl presented with a recurring bloody vaginal discharge for 3 months. Physical examination revealed pubic hair growth and breast enlargement. The infant had normal psychomotor development. Her weight and height corresponded to the 50th percentile for age. However, her skeletal age was 2.5 years according to the Greulich-Pyle method.

MR imaging of the hypothalamic and pituitary region was performed in order to establish the cause for precocious puberty. The protocol included axial and sagittal TSE T2-weighted images (Fig. 1), coronal T1 pre- and post-gadolinium images (Fig. 2), and sagittal T1 post-gadolinium images (Fig. 3). The study showed a well-circumscribed round lesion, 8 mm in diameter, in the interpeduncular cistern. The lesion was located posterior of the pituitary stalk, caudal of the mammillary bodies and against the pons and near the basilar artery. It was homogeneous and isointense to adjacent brain parenchyma on T1- and T2-weighted images. Gadolinium-enhanced images revealed no contrast enhancement of the nodule. These findings were consistent with a hypothalamic hamartoma.

As surgical resection was too risky due to the anatomic location of the lesion, the infant received medical treatment with gonadotropin-releasing hormone (GnRH) agonist triptorelin.

Discussion

Hypothalamic hamartomas are ectopic, non-neoplastic masses of normal neuronal tissue. The pathogenesis of precocious puberty in patients with hypothalamic hamartomas is considered to be the result of ectopic secretion of gonadotropin-releasing hormone (GnRH) or glial factors (2).

The radiological presentation of hypothalamic hamartoma is a well-circumscribed solid lesion attached to or embedded in the hypothalamus. The diameter of the lesion can be 5 to 50 mm and is usually between 10 to 30 mm (3). MR imaging is superior to computed tomography in displaying the exact size and anatomic location. The signal intensity of the hypothalamic hamartoma is generally isointense to that of normal grey matter on T1-weighted images and slightly hyperintense or isointense on T2-weighted images (3-9). The lesion does not enhance after the administration of gadolinium. The size of the lesion remains stable over time (3).

Differential diagnosis of hypothalamic hamartoma in children with precocious puberty includes optic glioma, astrocytoma, craniopharyngioma, lymphoma, and germ-cell tumor (3, 10). The detection of cysts, calcifications, contrast enhancement and tumor growth suggests neoplastic lesions.

Boyko et al. (7) noted that pedunculated hypothalamic hamartomas are correlated with precocious puberty while sessile hamartomas are correlated with seizures. Arita et al. (5) further investigated the relationship between MR findings and clinical manifestations of hypothalamic hamartomas in a retrospective study. They classified hypothalamic hamartomas into two categories based on imaging findings. The parahypothalamic type hamartoma is located in the suprasellar cistern and is only attached to the floor of the third ventricle or suspended from the floor of the third ventricle by a peduncle. Most patients in this category exhibited precocious puberty and had no seizures or developmental retardation. The intrahypothalamic type hamartoma is involved or enveloped by the hypothalamus and distorts the third ventricle. The patients in the latter category presented primarily with epilepsy, but developmental retardation and precocious puberty could also be associated. Furthermore, Freeman et al. (8) found that epileptogenic hamartomas are malformations of the mammillary region and are attached to one or both mammillary bodies. These findings suggest a role for these structures in epileptogenesis.

The treatment options of hypothalamic hamartoma associated with precocious puberty are long-acting GnRH agonists and surgery. GnRH therapy is the first choice to safely and effectively stop pubertal maturation (3). The treatment of seizures is more complex and includes antiepileptic drugs and surgical intervention.

We conclude that MR imaging is important in establishing the diagnosis of hypothalamic hamartoma in patients with central precocious

From: 1. Department of Medical Imaging, Ziekenhuis Oost-Limburg, Genk, Belgium.
Address for correspondence: Dr L. Topff, Department of Medical Imaging, Ziekenhuis Oost-Limburg, Schiepse Bos 6, 3600 Genk, Belgium. E-mail: laurens@topff.be

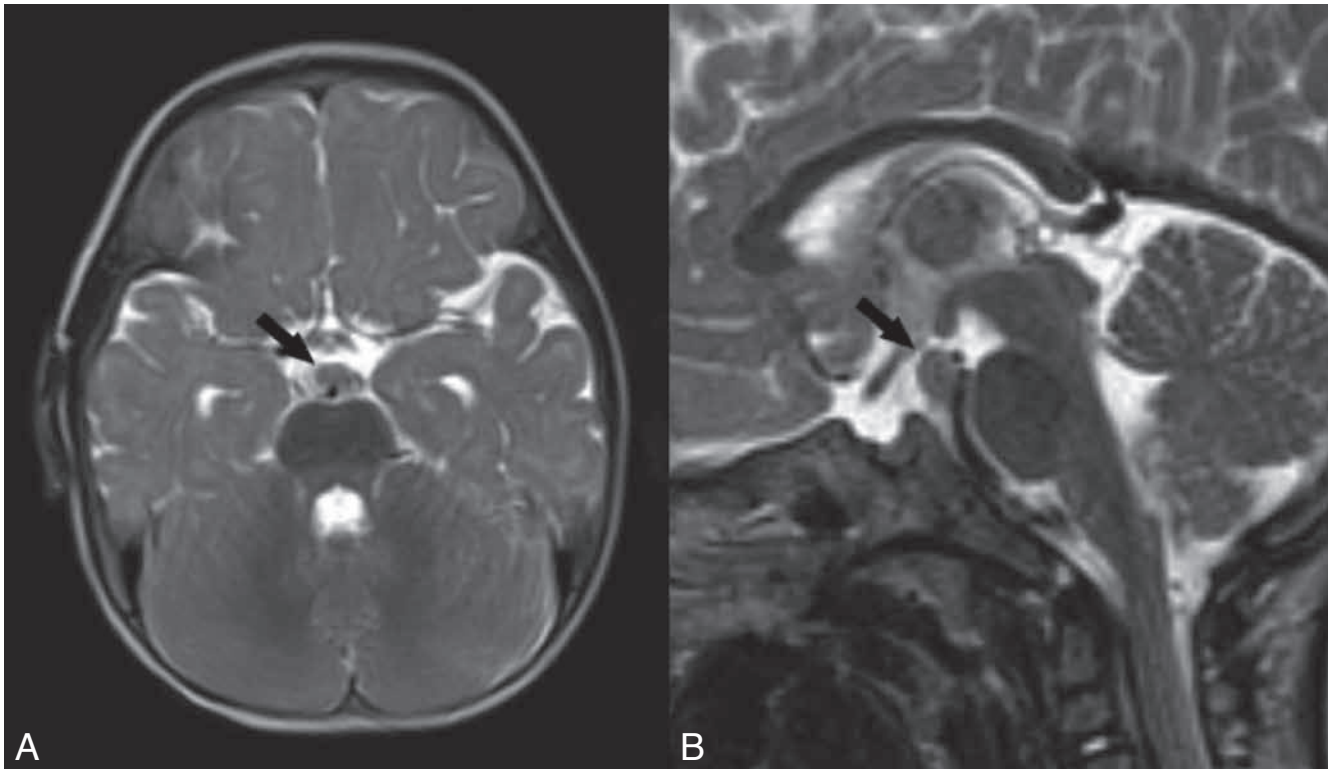

Fig. 1. — Axial (A) and sagittal (B) TSE T2-weighted MR images demonstrating a well-circumscribed lesion in the interpeduncular cistern (arrows). It is located caudal of the mammillary bodies. The lesion was isointense to adjacent brain parenchyma.

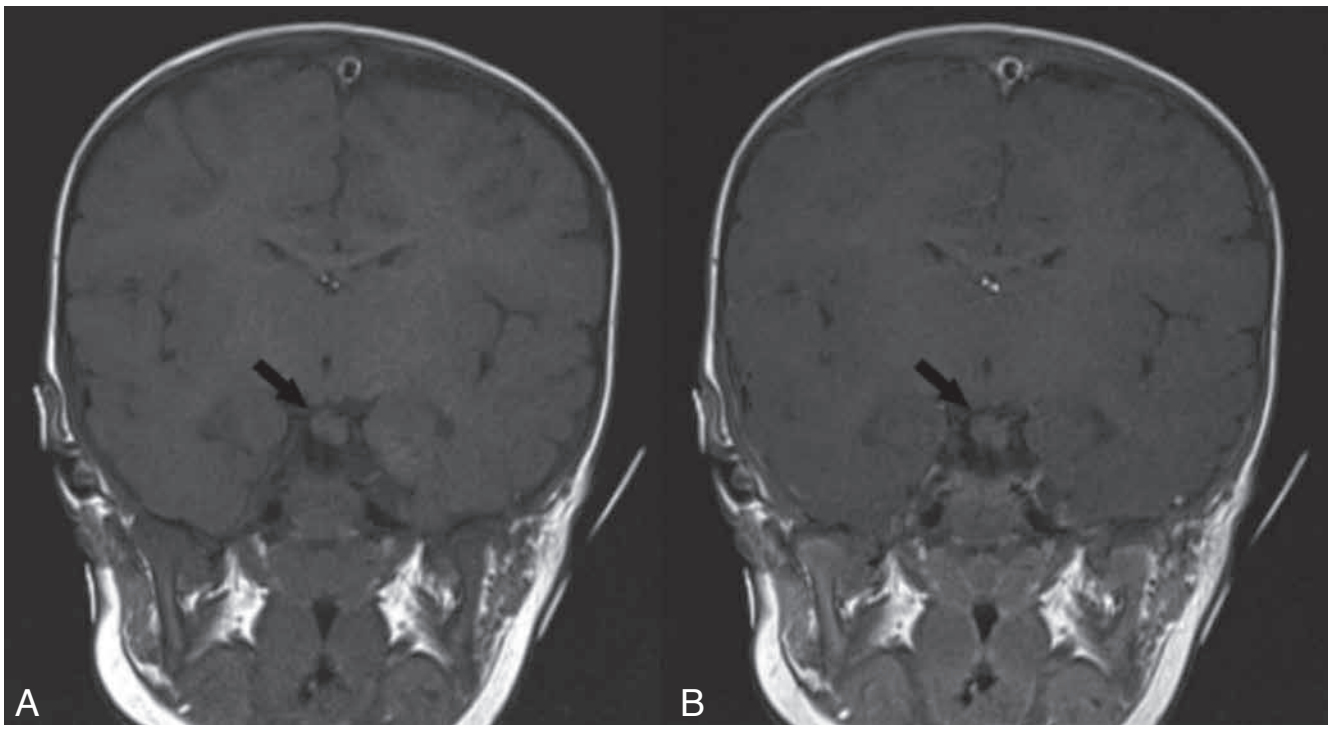

Fig. 2. — Coronal T1 (A) pre- and (B) post-gadolinium MR images demonstrating a lesion in the interpeduncular cistern without contrast enhancement and isointense to adjacent brain tissue (arrows).

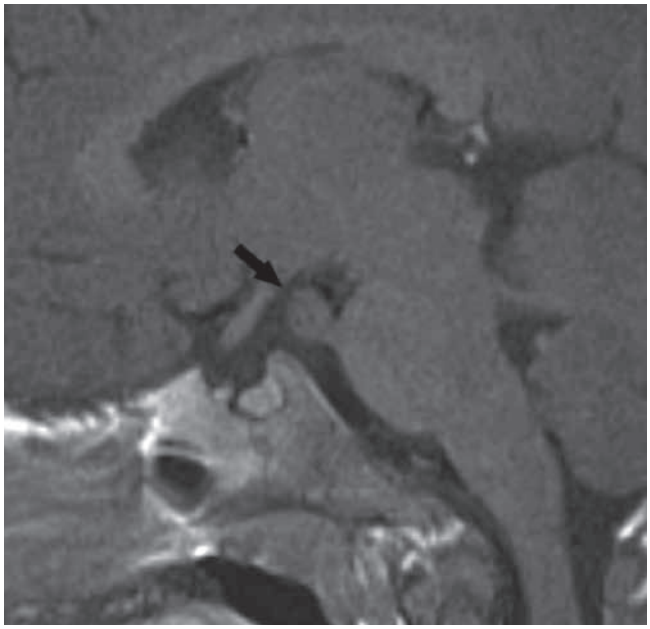

Fig. 3. — Sagittal T1 post-gadolinium MR image demonstrating the location of the lesion adjacent to the pons (arrows). There was no contrast enhancement of the lesion.

puberty. The lesion is generally isointense relative to normal grey matter on T1-weighted images and does not enhance after contrast medium administration. Understanding the anatomic location of the lesion in relation to the adjacent structures is useful in the preoperative assessment and prognosis.

References

1. Weissenberger A.A., Dell M.L., Liow K., et al.: Aggression and psychiatric comorbidity in children with hypothalamic hamartomas and their unaffected siblings. *J Am Acad Child Adolesc Psychiatry*, 2001, 40: 696-703.
2. Jung H., Ojeda S.R.: Pathogenesis of precocious puberty in hypothalamic hamartoma. *Horm Res,* 2002, 57: 31-34.
3. Arita K., Kurisu K., Kiura Y., Iida K., Otsubo H.: Hypothalamic hamartoma. *Neurol Med Chir* (Tokyo), 2005, 45: 221-231.
4. Barral V., Brunelle F., Brauner R., Rappaport R., Lallemand D.: MRI of hypothalamic hamartomas in children. *Pediatr Radiol*, 1988, 18: 449-452.
5. Arita K., Ikawa F., Kurisu K., et al.: The relationship between magnetic resonance imaging findings and clinical manifestations of hypothalamic hamartoma. *J Neurosurg*, 1999, 91: 212-220.
6. Lona Soto A., Takahashi M., Yamashita Y., Sakamoto Y., Shinzato J., Yoshizumi K.: MRI findings of hypothalamic hamartoma: report of five cases and review of the literature. *Comput Med Imaging Graph*, 1991, 15: 415-421.
7. Boyko O.B., Curnes J.T., Oakes W.J., Burger P.C.: Hamartomas of the tuber cinereum: CT, MR, and pathologic findings. *AJNR Am J Neuroradiol*, 1991, 12: 309-314.
8. Freeman J.L., Coleman L.T., Wellard R.M., et al.: MR imaging and spectroscopic study of epileptogenic hypothalamic hamartomas: analysis of 72 cases. *AJNR Am J Neuroradiol*, 2004, 25: 450-462.
9. Bruninx G., Widelec J., Delcour C.: Gelastic epilepsy and precocious puberty due to hypothalamic hamartoma. *JBR-BTR*, 2003, 86: 146-147.
10. Valdueza J.M., Cristante L., Dammann O., et al.: Hypothalamic hamartomas: with special reference to gelastic epilepsy and surgery. *Neurosurgery*, 1994, 34: 949-958.