

Visual Vignette

Submitted by

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Case Presentation: A 5-year-old boy was brought to us because his parents had noticed a rapid increase in his height during the previous 2 years, in conjunction with deepening of his voice during the last 12 months and premature maturation of his genitalia during the last 6 months. On examination, he was 137 cm tall, which was considerably above the 97th percentile for his age with a height SD score of +2.5 SD. The patient had well-developed muscles and weighed 30 kg. On genital examination, he had bilateral testicular volumes of 10 mL, pubic hair of Tanner stage 3, and an adult-sized penis. No skin lesions or skeletal abnormalities were detected. Results of the fundus and neurologic examinations were normal. Laboratory studies showed serum total testosterone and serum gonadotropin levels in the normal adult range. Blood levels of 17-hydroxyprogesterone, dehydroepiandrosterone sulfate, and cortisol were within the normal limits. The bone age was estimated to be approximately 12.5 years. Magnetic resonance imaging of the pituitary region revealed a normal sella and pituitary gland. A well-defined homogeneous lesion (14 by 14 by 11 mm) in the basal cistern below the mammillary body was isointense relative to the gray matter on T1-weighted images and did not enhance after administration of gadolinium (Fig. 1). **What is the diagnosis?**



Fig. 1

Answer: Tuber cinereum hamartoma. The well-defined nonenhancing homogeneous lesion in this region, which is isointense with the surrounding gray matter, is consistent with a hamartoma. Hypothalamic and tuber cinereum hamartomas are nonneoplastic nodules, consisting of mature neurons and glial cells. The hamartomas are either embedded in the hypothalamus (intra-hypothalamic) or attached to the ventral hypothalamus by a stalk anywhere from the tuber cinereum to the mammillary bodies (parahypothalamic). The degree of hypothalamic displacement correlates with the findings on clinical presentation. The parahypothalamic hamartomas are usually associated with only central precocious puberty and rarely with acromegaly, and they are generally nondisabling. The intra-hypothalamic variety is associated with more severe symptoms including intractable seizures, which may be focal or gelastic (“laughing”) seizures with secondary generalization. Two thirds of patients with intra-hypothalamic hamartomas have developmental delays and cognitive impairment (1).

As in our current patient, central precocious puberty is easily treated medically with gonadotropin-releasing hormone agonist therapy. Surgical therapy is rarely indicated. Antiepileptic drugs are used as a first-line intervention for control of seizures. In patients in whom the seizures are not adequately controlled with drugs, however, early neurosurgical referral to a medical center with expertise in the surgical management of hypothalamic hamartomas is important.

REFERENCE

1. Arita K, Kurisu K, Kiura Y, Iida K, Otsubo H. Hypothalamic hamartoma. *Neurol Med Chir (Tokyo)*. 2005;45:221-231.

(Editor’s Note: Submissions to “Visual Vignettes” are welcomed. Please send to
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