

Hamartoma of the Hypothalamus

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Hypothalamic hamartoma is a rare, tumor-like malformation associated with gelastic epilepsy, precocious puberty and behavioral disorders.

A twenty-year-old Bahraini male presented with poorly controlled epilepsy. MRI brain revealed a hypothalamic lesion inferior to the third ventricle, suggestive of hypothalamic hamartoma.

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Hypothalamic hamartoma is a congenital malformation, which causes complex partial seizures refractory to anticonvulsants in adulthood. It has been considered as the most common neurological cause of precocious puberty¹.

MRI plays a major role in the diagnosis of hypothalamic hamartoma. The hamartomas are usually seen as well-defined small non-enhancing lesions located at the floor of the third ventricle, exhibiting similar signal intensity as the grey matter. If a hamartoma is suspected, the hypothalamic region should be examined thoroughly, as they can be easily missed due to their small size^{2,3}.

The aim of this presentation is to report a case of hypothalamic hamartoma and emphasize the vital role of MRI in its diagnosis.

THE CASE

A twenty-year-old Bahraini male presented in 2008 with poorly controlled complex partial seizures, with right upper limb jerky movements, lasting 5 to 10 minutes in each episode. Neurological examination revealed no other significant neurological deficits. He was started on Trileptal (oxcarbazepine) 600 mg BD.

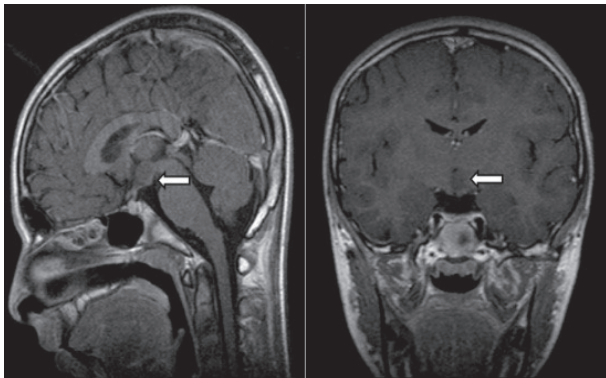


Figure 1: Sagittal and Coronal T1 Post-Contrast Sequences, a Small Lesion in the Hypothalamus. Iso-Intense to Gray Matter on T1 WI with No Obvious Enhancement

MRI brain imaging was performed with pre and post contrast sequences and revealed a small lesion in the hypothalamus, just to the left of the third ventricle in its inferior aspect, measuring approximately 1.2x1.3 cm. It is iso-intense to gray matter on T1 and slightly hyperintense to gray matter on T2 weighted images, see figures 1 and 2. No obvious enhancement was

noted on post-contrast sequences. The rest of the cerebral parenchyma was unremarkable. This lesion was compatible with the clinical history of a hypothalamic hamartoma.

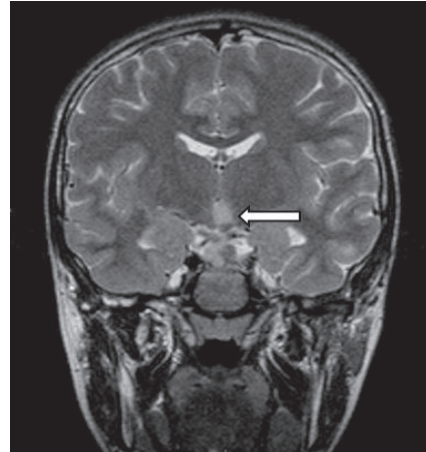


Figure 2: Coronal T2 WI Image, Showing the Hamartoma to Be Slightly Hyper Intense to Gray Matter

Despite the anti-epileptic medication, the patient was still symptomatic. He had radiosurgery in 2012. The patient developed generalized tonic-clonic seizures postoperatively. He was then started on Keppra (levetiracetam) 500 mg BD, Depakine (valproate) 500 mg BD in addition to the Trileptal.

A follow-up MRI brain performed in 2013 revealed an interval reduction in the hamartoma's size, see figure 3. The patient is currently on the same anti-epileptic regime, stable and no further attacks.

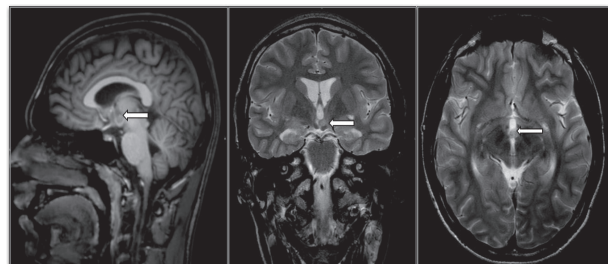


Figure 3: An Interval Reduction of the Size of the Hamartoma is Noted in the Follow-Up Study Dated 2013 after the Radio-Surgical Intervention. It Shows a Lower Signal on T2 WI, related to Radiation-Induced Gliosis

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DISCUSSION

Hypothalamic hamartoma is a rare, non-hereditary lesion originating from the tuber cinereum and inferior hypothalamus. Less than 80 cases have been reported in the literature. It usually affects patients in their first and second decade of life. It is seen in both sexes with no gender predilection⁴. Precocious puberty is the most common presentation^{5,6}.

Other clinical presentations include developmental delay, attention deficit or hyperactivity disorder and anxiety. Although uncommon, gelastic seizures that tend to develop complex partial seizures with or without secondary generalization are seen to occur in the association, often resulting in anticonvulsant drugs resistance. In some cases, visual abnormalities can be seen if the optic pathways were involved^{5,6}.

Brain MRI confirms the diagnosis of the hypothalamic hamartomas. They are seen as a homogenous small soft-tissue lesion at the floor of the third ventricle with similar signal intensity to the grey matter, with no enhancement on post-contrast images; they do not disturb the blood-brain barrier. Hemorrhage and calcifications are rarely seen⁷. In a recent study, it was found that hamartoma showed high signal intensity on T2 weighted images in 93% and low signal intensity in T1 weighted images as compared to the normal grey matter⁸. Notably, the mammillary region of the hypothalamus was involved in all cases. They can be large and exert regional mass effects; 97% of the cases showed intrahypothalamic extension, resulting in the displacement of the post-commissural fornix and hypothalamic gray matter anterolaterally.

Larger lesions tend to cause central precocious puberty. Signal intensity abnormalities of the anterior temporal white matter were seen in 16% with no associated changes of sclerosis⁸.

Craniopharyngioma, pituitary adenoma, hypothalamic/chiasmatic glioma and Rathke's Cleft Cyst are considered differential diagnosis. The presence of calcification, for example, can be used as a discriminator in cases of pediatric craniopharyngioma.

Although hypothalamic hamartomas are benign and usually stable in size, a regular follow-up every six to twelve months is recommended.

Medroxyprogesterone acetate is the treatment of choice in most cases. Medical management is used in patients with central precocious puberty. However, surgery including resection or disconnection can be helpful in cases of poorly controlled epilepsy. It can be performed via a craniotomy or trans-sphenoidal approach. Gamma knife radiosurgery has also been a common practice recently⁹.

CONCLUSION

Hypothalamic hamartomas are rare congenital malformations affecting the floor of the third ventricle. Precocious puberty is the most common presenting feature. MRI brain plays a major role in the diagnosis of hypothalamic hamartoma.

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