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A Rare Case of Hypothalamic Hamartoma – MRI Study

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Abstract :

Hypothalamic hamartomas are rare congenital hamartomatous non-neoplastic lesions that involve the hypothalamic region of the brain with clinical manifestations such as gelastic seizures or with precocious puberty. Imaging features are classical and helps narrow down the differentials.

Keywords : Gelastic seizures, precocious puberty, hamartomas

Introduction :

Hypothalamic hamartoma (HH), also known as diencephalic or tuber cinereum hamartoma, is a nonneoplastic congenital malformation associated with precocious puberty, behavioral disturbances, and gelastic seizures.

HHs are an anomaly of neuronal migration that probably occurs between gestational days 33 and 41. A syndromic abnormality that occurs with HH, Pallister-Hall syndrome (PHS), is caused by GLI3 frameshift mutations on chromosome 7p13.

Case History and Imaging Findings

A four year old female patient was brought to our department for MRI study of the brain to evaluate the cause for precocious puberty. Patient reportedly had breast buds at birth. Patient was seen to have developed axillary hair and pubic hair at the age of four years, which is when she was brought for clinical evaluation.

Patient was also seen to have elevated levels of FSH and LH.

MRI Findings

MRI study of the brain shows a large well-defined non-enhancing (Fig 5) T1 isointense (Fig 1,2) T2 iso-hyperintense(Fig 4), FLAIR hyperintense(Fig 3), lobuated mass involving the pituitary stalk with a normal pituitary glandular morphology and a well seen posterior pituitary bright spot(Fig 2). There is displacement of the optic chiasma with no direct involvement of the same(Fig 4). Diffusion weighted imaging shows no restricton of free water diffusion.

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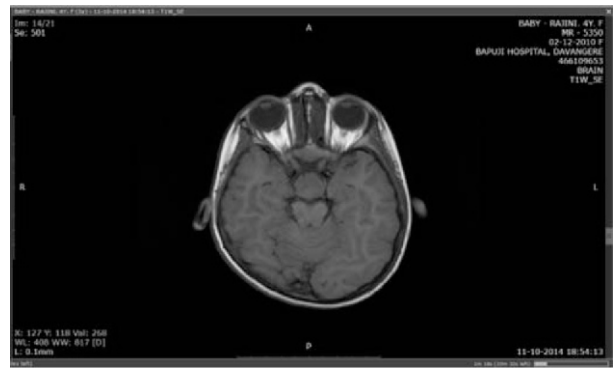

Fig 1 : Axial T1W Image shows a well defined isointense retrochiasmatic mass

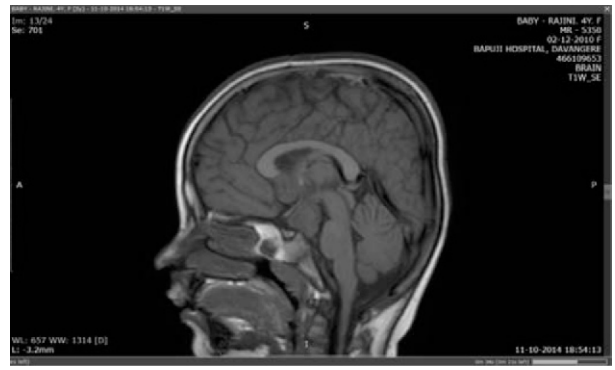

Fig 2 : Sagittal T1W image shows the involvement of the pituitary stalk to advantage.

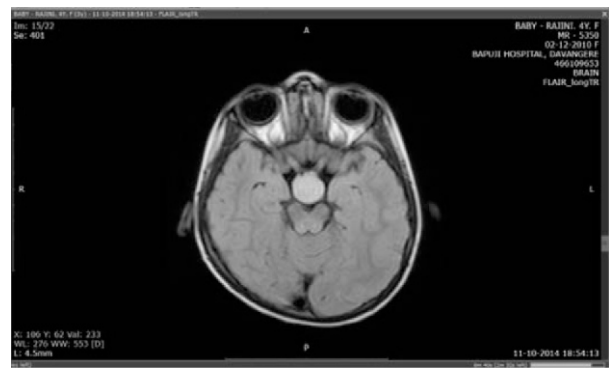

Fig 3 : FLAIR sequence shows that the mass is hyperintense

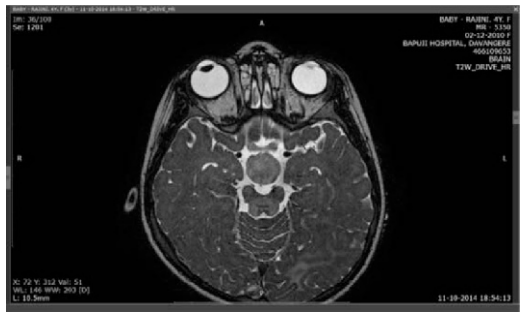

Fig 4 : TFE sequence shows that the mass is isointense to minimally hyperintense to cortex

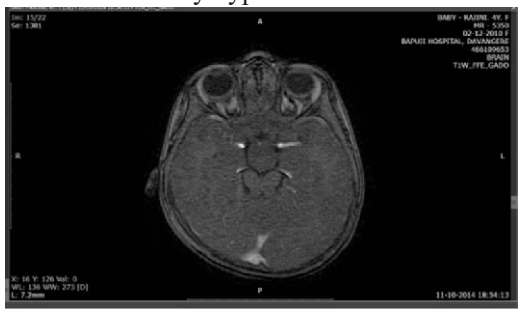

Fig 5 : Axial T1W post gadolinium scan shows no enhancement of the mass.

Discussion :

Le marquand and Russell[1] were the first to report hypothalamic hamartornas as congenital malformations consisting of tumour like collections of normal brain tissue lodged in an abnormal location.

Hamartomas are not true neoplasms. Pathologically they contain the nerve cells that resemble those of the tuber cinereum along with normal glial cells. Most of these lesions occur in hypothalamus. Subcortical cerebral cortex and periventricular regions have also been reported as other sites of occurrence of this rare lesion

Hypothalamic hamartomas are small pedunculated growths contiguous with posterior hypothalamus, between the tuber cinereum and mamillary bodies. They fill the free space between the optic chiasm and pons and usually do not distort the hypothalamus or other parts of the base of the brain unless they are very large.

They occur with equal frequency in males and females[2]. Most of patients usually present in the first or second decade of life. These patients typically present with precocious puberty. [35-70%] although the mechanism for this is not well understood. It may be from premature disappearance of normal inhibitory factors on hypothalamus. The larger hamartomas are less likely to produce precocious puberty[2].

Occasionally, visual disturbances may be present because of involvement of optic pathways. Other presenting symptoms may also include seizures and/or laughing spells. On imaging, hypothalamic hamartomas produce characteristic soft tissue masses iso-intense to grey matter. They are homogeneous and sharply marginated by the surrounding CSF, as they do not cross the blood brain barrier they do not show enhancement on post-contrast images. Calcification is rare and haemorrhage is not described in these lesions. The anatomic location of these hamartomas together with signal intensity similar to grey matter on T1 WI and a higher intensity than gray matter on PDWI and T2 WI strongly, supports this diagnosis[3].

Differential diagnosis of hypothalamic hamartoma may include craniopharyngioma, optic gliomas hyothalamic gliomas and gangliogliomas. All these tumours enhance after contrast. Calcification is common in craniopharyngiomas and optic gliomas. Hypothalamic gliomas although a close differential are usually, inhomogeneous and often show enhancement. Rarely, enhancement in hypothalamic hamartomas has been reported in patients with neurofibromatosis[4]. In cases of hypothalamic hamartomas surgical treatment is not always favourable. Medical treatment with medroxy progesterone acetate is the treatment of choice in most patients[5]. Follow up MRI is recommended every six to 12 months for demonstrating lack of growth.

Conclusion :

In conclusion, hypothalamic hamartomas are rare congenital malformations. Classical clinical presentation of isosexual precocious puberty, alone, with MR imaging features of isointense non-enhancing, mass of the grey matter intensity in the region of the hypothalamus is highly, suggestive of the diagnosis.

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