### **Case Report**

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# Diagnosis of precocious puberty due to tuber cinereum hamartoma by clinicoradiologic correlation

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#### **ABSTRACT**

We present a case of a 3 year male child who presented with precocious puberty. Endocrinological studies revealed increased LH and increased testosterone. Bone age was 8 years. MR Imaging showed hypothalamic mass isointense to brain parenchyma on T1-WI and hyperintense on T2-WI, without contrast enhancement. Typical clinical presentation, endocrinological studies and MRI features strongly favoured the diagnosis of tuber cinereum hamartoma. This case is presented to emphasize the importance of clinicoradiologic correlation in the diagnosis of precocious puberty due to tuber cinereum hamartoma.

Keywords: Tuber cinereum hamartoma, Precocious puberty

#### INTRODUCTION

Tuber cinereum hamartoma is a rare non-neoplastic congenital malformation. The characteristic clinical symptoms are precocious puberty, gelastic seizures, cognitive and behavioural abnormalities. Tuber cinereum hamartoma is the commonest cause of organic central precocious puberty. The diagnosis of tuber cinereum hamartoma is based on the typical MRI findings.

#### **CASE REPORT**

A 3 year male child presented with enlargement of penis, appearance of pubic hairs, acne over the face and deepening of voice, noticed by the parents over last 6 months.

On physical examination, his height was 108cm (>95<sup>th</sup> percentile), weight 20kg (>95<sup>th</sup> percentile), enlarged penile length of 80 mm (Normal - 25-40 mm); testicles were enlarged in size with testicular volume of 15 ml

bilaterally (Normal - 1-3 ml). There was acne over forehead region (Figure 1). Voice was of low pitch and deep (adult type). Tanners stage was 3 (dense curly pubic hairs but sparing medial thigh).



Figure 1: Acne over forehead region.

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Plain radiograph of left wrist (Figure 2) revealed radiological bone age of about 8 years (Chronological age - 3 years). Endocrinological findings were as follows: Follicle Stimulating Hormone (FSH): 3.06 mIU/ml (Normal - 0.26-3.0 mIU/ml), Luteinizing Hormone (LH): 12.76 mIU/ml (Normal - 0.02-0.30 mIU/ml), testosterone: 791.56 ng/dl (Normal - 2.0-25.0 ng/dl). Thyroid function tests were normal. There was no adrenal pathology on ultrasound abdomen and testicular parenchyma was homogeneous in echotexture with the size of 34x24x18 mm on the right and 34x24x17 mm on the left. Based on physical & endocrinological findings, it was suspected as a case of central precocious puberty.



Figure 2: Radiograph for bone age. Bone age: 8 years.

MR imaging of the brain (Figure 3-6) showed 2x1.6x1.3 cm non-enhancing hypothalamic mass posterior to pituitary stalk that was isointense to brain parenchyma on T1-weighted images and hyperintense on T2-weighted images. Adenohypophysis hyperplasia was present. The diagnosis of tubercinereum hamartoma was suggested on MRI.

During follow up after 6 months of starting conservative treatment with gonadotropin-releasing hormone (GnRH) analogue (Leuprolide acetate), his progression of puberty had arrested, acne over the forehead resolved (Figure 7) and the testosterone level returned to normal 14.59ng/dl, which is normal for his age.



Figure 3: MRI brain T1 sagittal images.

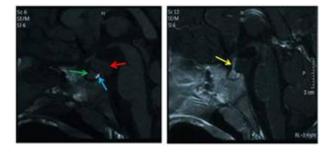


Figure 4: T1 without and with contrast (sagittal). Green: ant pituitary, Blue: posterior pituitary bright spot, Red: isointense mass (not enhancing with contrast) - posterior to pituitary stalk, Yellow: pituitary stalk.



Figure 5: T2-W sagittal image: hyperintense lesion on T2-WI.

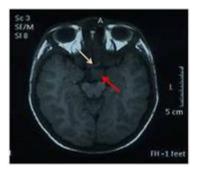


Figure 6: T1-W axial. The mass (red arrow) (isointense on T1) sparing optic chiasma (white arrow).



Figure 7: Before treatment: acne over forehead region. After treatment with gonadotropin-releasing hormone (GnRH) analogue (Leuprolide acetate), acne over the forehead resolved.

#### **DISCUSSION**

Tuber cinereum hamartoma or hypothalamic hamartoma is a rare non-neoplastic congenital malformation presenting as a mass in the hypothalamus arising from tuber cinereum, a part of the hypothalamus located between the mammillary bodies and the optic chiasm. Histologically, tuber cinereum hamartomas resemble normal hypothalamic neurons, although some dysplastic neurons and glial cells have also been described. <sup>1</sup>

Symptoms often begin in early infancy. The characteristic clinical symptoms are precocious puberty, gelastic seizures, cognitive and behavioural abnormalities. 1 Precocious puberty is defined as pubertal onset before the age of 8 years in girls and 9 years in boys. Tuber cinereum hamartoma is the commonest cause of organic central precocious puberty.<sup>2</sup> Central precocious puberty results from oversecretion of gonadotropin releasing hormone (GnRH), which in turn results in over production of Luteinizing Hormone (LH) and Follicle Stimulating Hormone (FSH). Precocious puberty was the presenting symptom in our case. Gelastic seizures are typically of short duration (2-30 seconds), characterized by uncontrollable laughter, without impairment of consciousness.3 Gelastic seizures were absent in our patient. Our patient had normal mental intelligence and cognitive functions.

Physical examination in precocious puberty includes height, weight and tanners staging. Height and weight of our patient was >95<sup>th</sup> %tile for age and Tanners Stage was 3. Investigations should include LH, FSH, testosterone levels and bone age. LH and testosterone were ↑ in our case. Bone age was 8years (chronological age 3 years).

Patients with pubertal LH (↑LH) levels should undergo MRI of brain. On MRI, tuber cinereum hamartomas appear as hypothalamic mass isointense to cerebral cortex on T1WI and hyperintense on T2WI, without contrast enhancement. Similar imaging findings were seen in our case.

Treatment options for Precocious puberty associated with tuber cinereum hamartoma may be medical or surgical. Medical treatment includes use of a long acting GnRH analogue (Leuprolide acetate 300 µg/kg) as a deep intramuscular injection every 4 weeks until puberty.

Precocious puberty is generally controlled effectively by GnRH analogues.<sup>5</sup> Our patient with precocious puberty responded well to leuprolide treatment.

#### **CONCLUSION**

Tuber cinereum hamartoma is the commonest cause of organic central precocious puberty. Clinical features and endocrinological studies should be correlated with the typical MRI findings in the diagnosis of precocious puberty due to tuber cinereum hamartoma. Accurate diagnosis is important for early treatment.

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#### REFERENCES

- Amstutz DR, Coons SW, Kerrigan JF, Rekate HL, Heiserman JE. Hypothalamic hamartomas: correlation of MR imaging and spectroscopic findings with tumor glial content. AJNR Am J Neuroradiol. 2006;27(4):794-8.
- 2. Ng SM, Kumar Y, Cody D, Smith CS, Didi M. Cranial MRI scans are indicated in all girls with central precocious puberty. Arch Dis Child. 2003;88:414-41.
- 3. Engel J, Pedley TA, Aicardi J. Syndromes of late childhood. In: Engel J, Pedley TA, Aicardi J, eds. Epilepsy: A Comprehensive Textbook. 2nd ed. Philadelphia: Wolters Kluwer Health/Lippincott Williams & Wilkins; 2008: 2525.
- 4. Shah P, Patkar D, Patankar T, Shah J, Srinivasa P, Krishnan A. MR imaging features in hypothalamic hamartoma: a report of three cases and review of literature. J Postgrad Med. 1999;45:84-6.
- de Brito VN, Latronico AC, Arnhold IJ, Lo LS, Domenice S, Albano MC, et al. Treatment of gonadotropin dependent precocious puberty due to hypothalamic hamartoma with gonadotropin releasing hormone agonist depot. Arch Dis Child. 1999;80:231-4.

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