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A Case of Central Precocious Puberty Secondary to Hypothalamic Hamartomas with Gelastic Seizure

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INTRODUCTION

Central Precocious Puberty (CPP) is caused by early maturation of the hypothalamic- pituitary-gonadal axis, characterized by sequential development of secondary sexual characteristics before 8 years in girls and 9 years in boys. We reported a case of CPP secondary to Hypothalamic Hamartomas (HH) which was initially referred for sexual precocity.

CASE

NAI, currently 6 years old, presented with monthly episodes of per vaginal bleeding at 2 months old. There was concurrent breast enlargement associated with peculiar body odor and episodes of inappropriate giggling during infancy. There was positive family history of epilepsy on the paternal side. Physically, NAI is not dysmorphic, appears tall and overweight for age with Tanner staging of B4, A1, P1. There were neither neurocutaneous signs nor virilization of the external genitalia.

Basal and stimulated LH, FSH values and ultrasound pelvis showed pubertal values with advancement of bone age noted on bone age assessment. Following this, she was commenced on monthly IM Lucrin injection which showed favorable biochemical and clinical response after 3 months of treatment. An EEG done showed abnormal epileptic records after she developed recurrent episodes of gelastic seizure since May 2016. An MRI of the brain showed the presence of a well define solid/lobulated mass in hypothalamus measuring 2.1 x 2.3 x 2.8 cm, with no mass effect to adjacent structure. An MRI was repeated after 2 years due to uncontrolled seizure despite being on oral Keppra, and showed similar findings as before.

CONCLUSION

Hypothalamic Hamartomas are rare congenital lesions presenting with classic triad of central precocious puberty, gelastic epilepsy and development delays. CPP responds well to treatment with GNRH agonist. However, gelastic seizure can differ in severity and evolution in different individuals. Majority are resistant to antiepileptics hence our patient may benefit from surgical removal of hamartomas either via transcallosal approach or minimally invasive surgery.

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A Case of Delayed Puberty and Anosmia

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INTRODUCTION

A 26-year-old lady presented at the age of 13 years with delayed puberty and faltering growth. She had a past history of left eye squint surgery for underlying optic disc coloboma, but was otherwise systemically well. She was found to have absent sense of smell. There were no prior feeding problems and she had normal intelligence. Initial assessment revealed a proportionately small-sized girl who was prepubertal, there were no obvious dysmorphic features.

CASE

Her karyotype was 46XX. Her bone age was 8 years at chronological age of 13 years. A combined pituitary function test showed poor LH and FSH response with peak responses of 0.34 IU/L and 2.62 IU/L respectively. Serum estradiol was <37 pmol/L. GH, TSH and cortisol responses were normal. MRI of the pituitary was normal. The findings of isolated hypogonadotropic hypogonadism with anosmia pointed to a clinical diagnosis of Kallmann syndrome. She was commenced on hormonal replacement therapy for induction of puberty. She complained of hearing difficulties at the age of 19 years. Pure tone audiometry confirmed bilateral conductive hearing loss. Subsequent CT temporal bones showed bilateral absence of all the semi-circular canals. The clinical diagnosis was revisited and revised to possible CHARGE syndrome as the patient fulfilled 3 major criteria of CHARGE syndrome i.e. coloboma, anosmia and absent semi-circular canals and 1 minor criterion i.e. delayed puberty secondary to hypogonadotropic hypogonadism. This was confirmed by a detectable pathogenic CHD 7 gene mutation.

CONCLUSION

The presence of non-reproductive anomalies including anosmia, coloboma and hearing defects, are red flag indicators of an underlying congenital hypogonadism. CHARGE syndrome is a major differential diagnosis of Kallmann syndrome and should be considered especially in girls. Patients with anosmia and hypogonadotropic hypogonadism should be screened for clinical features consistent with CHARGE syndrome.