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PRECOCIOUS PUBERTY, GELASTIC SEIZURES AND HYPOTHALAMIC HAMARTOMA

P.S.N. Menon
N.K. Mishra
V. Muthukumar

Hypothalamic hamartoma is a well defined rare syndrome which often presents with precocious puberty and seizures(1). With the advent of magnetic resonance imaging (MRI), even very small sized hamartomas can be picked up. Using gonadotropin releasing hormone (GnRH) analogs, precocious puberty as well as skeletal maturation can be controlled without any major side effects(2). However, the seizures which occur because of cerebral dysgenesis still pose a therapeutic problem(3). We report here a boy with hypothalamic hamartoma who presented with precocious puberty and gelastic seizures.

Case Report

A product of full term normal delivery following an uneventful antenatal period presented at 2½ years of age in 1985 with increasing size of testes and penis noticed immediately after birth and increased appetite from early infancy. Pubic hair appeared at 6 months of age and erections and ejaculations were observed at 1 yr of age. There was no axillary hair. In addition, he had behavioral abnormalities in the form of obstinacy and temper tantrums.

He was investigated in our hospital in 1985. CT scan and CT cisternography revealed a suprasellar isodense mass suggestive of hypothalamic hamartoma (*Fig. 1*). He underwent surgery in 1986 with a right frontal craniotomy with partial decompression of sella. Follow up CT scans showed persistence of the tumor. He was put on buserelin, a GnRH analog for arresting further pubertal development.

Six months later he developed secondary tonic seizures. Acne appeared at the age of 4½ yrs and chest hair appeared at 6½ yrs. He had discontinued buserelin therapy after one year due to financial problems. In 1991, at the age of 8½ yrs, he again

From the Division of Pediatric Endocrinology, Department of Pediatrics and Department of Neuroradiology, Neurosciences Centre, All India Institute of Medical Sciences, New Delhi 110 029.

Reprint requests: Dr. P.S.N. Menon, Additional Professor of Pediatrics, All India Institute of Medical Sciences, New Delhi 110 029.

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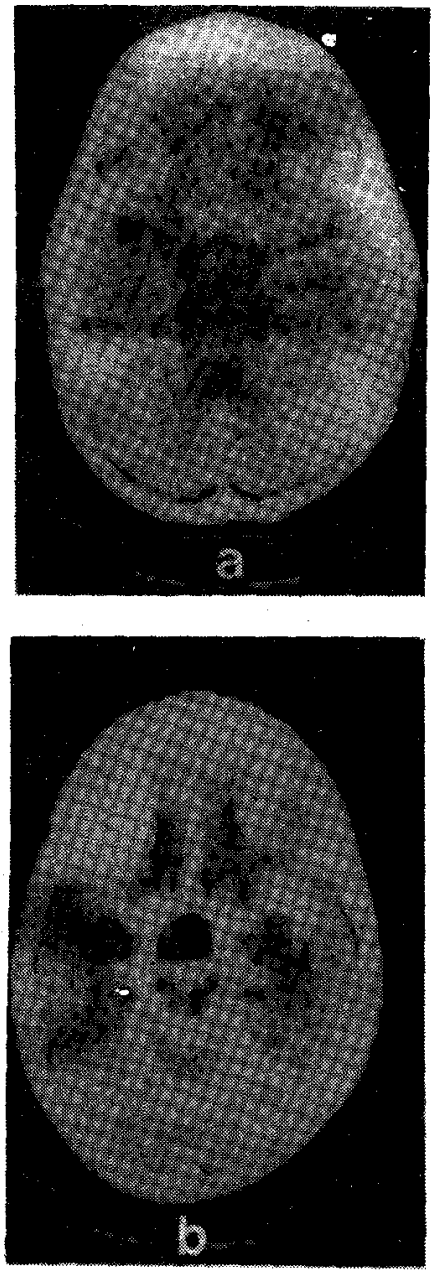

Fig. 1. (a) Contrast enhanced CT scan showing an isodense suprasellar mass. (b) CT cistemogram showing the filling defect in the suprasellar cistern by the mass.

presented for further progression of secondary sexual characters, uncontrolled seizures and behavioral abnormalities. In addition he had typical gelastic seizures (laughing spells with abrupt loss of consciousness).

At presentation, he had acne, muscular build and adult type body habitus with moustache, beard, further hair growth over the chest and voice change. There was no axillary hair. Pubertal development was Stage V of Tanner. Penis measured 10 cm in length and 9.5 cm in girth. Testis measured 25 ml on the right and 15 ml on the left. His height and weight were 142 cm and 37.5 kg, respectively, both above the 95th percentiles of ICMR standards. Upper and lower segment ratio showed shortening of lower segment (76 : 66). Neurological examination was unremarkable except for the secondary tonic and gelastic seizures. Fundus examination showed bilateral hypoplasia of the temporal half of optic nerves. Examination of other systems was unremarkable.

On investigation, bone age (TW2 method) was advanced (17.4 yrs). Follow-up CT scan of brain showed an isodense mass in the interpeduncular cistern. MRI confirmed that this isodense mass was situated in the tuber cinereum and was not enhanced by contrast. EEG showed features of generalized tonic seizures. His basal LH levels ranged between 14.3-18.3 IU/L, FSH 11.2-21.2 IU/L, testosterone 22-110 ng/ml. The peak response of LH to GnRH given IV was 27.3 IU/L. He was treated with phenytoin and diazepam for control of seizures. There was a partial response to this treatment. GnRH analog therapy was not initiated because of financial constraints.

Discussion

Precocity in boys is defined as appearance of secondary sexual characters before 9 yrs of age(4). Isosexual precocity is broadly classified into central and peripheral precocious puberty. Among the CNS tumors which produce central isosexual precocity (CIP), hypothalamic hamartoma is the most common and is being diagnosed most frequently(5). The first case of hypothalamic hamartoma was described in 1934(6). Till 1980 only 37 cases were

described and by now about 95 cases have been reported. The recent increase in the incidence is due to better imaging by CT and MRI.

Hypothalamic hamartoma is a congenital malformation characterized as heterotopic and hyperplastic tissue located in proximity to the tuber cinereum and mammillary bodies(7). It is usually small in size, ranging from 1 to 6.5 cm in diameter. It is a developmental aberration presumably resulting from the remnants of brain tissue left along the floor of the third ventricle, while the chorda withdraws(7). Histologically it consists mainly of normal brain elements.

The most common clinical presentation of hypothalamic hamartoma is precocious puberty as also observed by Zuniga *et al.* in about 74% of these cases(7). Genital changes may begin soon after birth(8). The progression is usually rapid with an occasional absence of adrenarche(9). Early epiphyseal closure results in short adult stature. The development of CIP in hypothalamic hemartoma is related to the secretion of GnRH. Electron microscopy and immunochemical studies have demonstrated the presence of secretory granules containing GnRH(11,12), indicating that it acts as an accessory hypothalamus(10,12). Alternately, this could be due to interference with the prepubertal inhibitory influences(3,12,13) or a direct effect on the hypothalamus(3).

Various forms of seizure disorders and classically gelastic seizures are associated(3). Gelastic seizures may begin at birth or later(14). Secondary generalized seizures denote cerebral dysgenesis, which may also explain the abnormal behavior in these children(3). Hypoplasia of optic nerves in this patient supported this possibility. Gelastic seizures probably occur due to the stimulation of the floor of the third ventricle(3,14).

With the advent of CT and MRI, imaging in particular the latter, small hypothalamic hamartomas can be identified(3,15). CT usually shows an isodense lesion in the interpeduncular cistern. On T1 weighted MR images the hypothalamic mass appears isointense with grey matter and on T2 weighted images it becomes hyperintense. There is no enhancement with Gadopentate reflecting an intact blood brain barrier.

The aim of therapy is to control precocious puberty and associated seizure disorders. Cyproterone acetate, medroxyprogesterone and of late, GnRH analogs are used. The latter control skeletal maturation effectively(2,12,16). Surgery is not the favored mode of treatment even though microsurgical techniques have been tried, when feasible with pedunculated hamartomas(5). Surgical intervention did not help this child and carries a high risk of morbidity.

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STRAIGHT LINE SIGN IN PATENT DUCTUS ARTERIOSUS

Harjit Singh
J.C. Chugh
A.H. Shembesh
Abdulla Al Khayat
A.A. Ben-Musa

Even though echocardiography has revolutionized the diagnostic workup in congenital heart disease, the technique is yet not routinely available in most of the developing world. A straight *X*-ray chest is a routine investigation in such situations.

In patent ductus arteriosus (PDA), the concavity between the aortic knob and the pulmonary artery is filled in to form a straight left border at the base of cardiac silhouette, in a straight *X*-ray of chest (*Fig. 1*). This, alongwith plethoric lungs and a large heart, make the classical presentation of PDA(1). The straight border, referred to as the "straight line sign", is not commonly described in pediatric and cardiology texts, as well as in books on pediatric radiology(2-4).

In order to assess the clinical value of the straight line sign in the radiological assessment of children with PDA, the present study was undertaken.

From the Departments of Pediatrics, Al Fatah Children's Hospital, Faculty of Medicine, Al Arab Medical University, Benghazl, Libya, and Al Wasl Hospital for Women and Children, Dubai, UAE.

Reprint requests: Prof. Harjit Singh, P O Box 12836, Dubai UAE

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