Physical, neurological and fundus examination were normal. No skeletal abnormalities were noted in the child, parents or any of the first degree relatives. In view of the afebrile focal seizures the child was started on carbamazepine (10 mg/kg/day), however her convulsion worsened in frequency and severity in spite of increase in dose of carbamazepine. She subsequently developed painful tingling sensations in the extremities and her right hand was flexed and adducted at the wrist.

Investigations revealed a total serum calcium of 4.9 mg/dl, with ionized fraction of 2.8 mg/dl; and serum phosphorus 9.2 mg/dl, serum alkaline phosphotase :328 IU/L, serum magnesium—2.0 mg/dl; and serum parathyroid hormone (PTH) by radio-immunoassay was 162.2 ng/dl (N=0-27 ng/dl). CT scan of the head revealed bilateral multiple symmetrical calcifications in the lentiform and caudate nuclei, and bilateral multiple calcifications in the subcortical white matter in the frontal and parietal regions. On contrast administration there was no abnormal enhancement. Skull X-ray, radiological skeletal survey and CSF examination were within normal limits. EEG was suggestive of neuronal hyperexcitability.

The association of hypocalcemia, hyperphosphatemia elevated PTH levels and basal ganglia calcifications led to the diagnosis of pseudohypoparathyroidism (PHP) without any phenotypic evidence of Albright's hereditary osteodystrophy (AHO).

She was treated with calcium supplements and 1,25 dihydroxycholecalciferol (DHCC) at a dose of 0.25 μg/day initially which was later increased to 0.75 μg/day.
There was dramatic cessation of seizures and disappearance of paraesthesia within 72 hours. This was followed by gradual normalization in serum calcium, phosphorous and PTH levels. The anticonvulsants were withdrawn and the dose of 1.25 DHCC was titrated according to her biochemical parameters. On follow up over the past one year she has been totally normal and seizure free on a maintenance dose of 0.25 μg/day of 1.25 DHCC and oral calcium supplementation of 1 g/day. Her serum calcium (total and ionic) and phosphorus are within normal limits.

Discussion

Presence of hypocalcemia and hyperphosphatemia in the absence of hypomagnesemia or chronic renal failure leads to a suspicion of hypoparathyroidism. The differentiation between the hormonopenic and PHP rests on the demonstration of either absence or presence of end organ resistance to the action of PTH. Untreated PHP almost uniformly show elevated serum levels of PTH and in the absence of hypomagnesemia or chronic renal failure, the diagnosis is fairly certain, as in our case.

Clinical findings of skeletal and developmental anomalies like a stocky build, round face, brachyactyly and mental retardation, are suggestive of AHO. If present in the patient or in any first degree relative the likelihood of PHP increases but these are not essential for the diagnosis. The skeletal defects of AHO are progressive and may not be apparent until the child is more than 5 years of age(1,2).

Basal ganglia calcification also occurs in a number of conditions not affecting the calcium-phosphorus metabolism such as the Gardener’s syndrome, Turner’s syndrome, basal cell nevus syndrome and familial calcification of the basal ganglia(1,3). Basal ganglia calcification not visible by radiography may be discovered fortuitously in 0.7% of routine CT Scan of the brain(4). Calcification of the basal ganglia occurs in nearly 50% of patients with PHP(1,3) it has rarely been described below 5 years and the earliest age at which radiological detection is possible is unclear. Calcification may also occur in the dentate nuclei and cerebral cortex as was seen in our case. The average age of presentation of PHP is 8 years and symptoms are often present for many years before a correct diagnosis is made.

Although tetany is the commonest symptom almost all types of seizures have been described including generalized tonic-clonic, focal jacksonian progression and petitmal attacks(3). Anticonvulsant therapy with diphenylhydantoin or phenobarbital has been shown to suppress tetanic manifestations by suppressing neuronal excitability but they do not correct the metabolic defect(5). Cognitive defects are also frequently encountered and are usually permanent, especially if significant hypocalcemia occurs in the perinatal period(1,3). Restoration of normal biochemical parameters, especially normocalcemia, with calcium supplements and 1.25 DHCC or high dose ergocalciferol (Vitamin D2) therapy controls the seizures although the basal ganglion calcification and lenticular opacities, if present, are irreversible(3,6). Regular follow up and monitoring of biochemical parameters is required for early recognition of complications like vitamin D intoxication and hypercalcemia(1,3).

REFERENCES


Isosexual Male Precocity Due to a Leydig Cell Tumor

S.K. Advani
N.M. Khan
S.S. Borwankar
P.B. Badhe
B.K. Kulkarni
S.D. Bhandarkar
K.V. Moralwar

A ten-year-old boys presenting with isosexual precocity had a Leydig cell tumor of the right testis. The case is presented as Leydig cell tumor is an uncommon cause of male precocious puberty(1,2). Further a Leydig cell tumor replacing the entire testis is very rare.

Case Report

A ten-year-old boy was brought to the Puberty and Growth Clinic for tall stature and a large penis. These abnormalities had been noticed by the mother about 8 months earlier, and during the intervening 8 months, the patient developed a muscular, hirsute male habitus and had erections and nocturnal emissions. On examination, the height was 142 cm (>95th percentile of ICMR standard(3), height age: 14 years), weight 41.3 kg (weight age: 15 years) and BP 110/70 mm Hg. The pubic hair were of Tanner Stage P4, the stretched penile length was 7.5 cm and the penis was of adult thickness. The right testis was 10 ml in volume with a normal configuration, whereas the left testis was 2 ml in volume and normal in consistency. The build was muscular and there were hair on the face, chest and arms. There was no gynecomastia.

Investigations showed the following: bone age 15 years (Greulich and Pyle's standards)(4), serum testosterone 25 ng/ml (normal 0-0.6 ng/ml in children), serum estradiol 45 pg/ml (normal male 30 pg/ml), serum FSH 7.9 mIU/ml (Normal: male prepuberty: 1-9 mIU/ml–Leeco Std: 2nd IRP–78/549), serum LH 5 mIU/ml

From the Departments of Endocrinology, Pediatric Surgery and Pathology, Seth G.S. Medical College and K.E.M. Hospital, Parel, Bombay 400 012.

Reprint requests: Dr. Naznin M. Khan, 17-C, Elco Arcade, Hill Road, Bandra, Bombay 400 050.

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