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Cretinism presented as a Case of Craniopharyngioma

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ABSTRACT

In Congenital hypothyroidism (CH) there is inadequate thyroid hormone production in newborn infants. Short stature with a mentally challenged state is frequently attributed to CH. Here we report a case of craniopharyngioma in an adult with untreated congenital hypothyroidism. CT scan of the brain revealed a large sella and supra sellar mass compressing the optic chiasma. MRI features of brain was found consistent with sella and supra sellar complex mass with mass effect having solid and cystic components. The clinical diagnosis of congenital hypothyroidism was confirmed by elevated TSH, low FT4, skeletal survey and ultra-sonography of thyroid gland. This was an unusual situation of a large craniopharyngioma detected in an adult with untreated congenital hypothyroidism.

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INTRODUCTION

In Congenital Hypothyroidism (CH), there is inadequate thyroid hormone production in newborn infants. It can occur because of an anatomic defect in the gland, an inborn error of thyroid metabolism, or iodine deficiency. Short stature with a mentally challenged state is frequently attributed to CH. The prevalence of cretinism in Bangladesh is 0.5% (hilly, 0.8%; flood-prone, 0.5%; and plains, 0.3%).[1] Nearly 69% of Bangladeshi population has biochemical iodine deficiency. Most patients have thyroid ectopia, aplasia or hypoplasia and present with varied clinical manifestations with a 2:1 female to male preponderance.[2-4] The pseudotumour generally regresses with adequate levothyroxine supple-mentation, but craniopharyngioma does not respond to such treatment.[5] We report a rare case of a craniopharyngioma in a 25 years old man of short stature in a mentally challenged state with untreated CH. Thyroid function test, X ray, clinical history helps to confirm CH and

Computed Tomography (CT) and MRI of brain helps to know the presence of craniopharyngioma and the extent of the lesion.

Craniopharyngioma is a slow-growing, non-cancerous brain tumour that develops near the pituitary gland and the hypothalamus.[1,6] This tumour most commonly affects children between 5 and 10 years of age; however, adults can sometimes be affected.[1] Although craniopharyngiomas are not cancerous, they may grow and press on nearby parts of the brain, causing symptoms including hormonal changes, vision changes, delayed growth, headaches, nausea and vomiting, loss of balance, hearing loss and changes in mood or behaviour.[2] The cause of these tumours is not well understood; however, researchers suspect that they begin to form during the early stages of embryo development in pregnancy and may result from metaplasia.[2,4] These tumours are closely related to another cystic mass occasionally seen in the pituitary called Rathkes cleft cyst.[5-8] Craniopharyngioma has two major pathologic variants like adamantinomatous and papillary. The adamantinomatous type is most common in children.[9,10] Treatment for craniopharyngioma varies and may involve surgery to remove the tumour, radiation therapy, chemotherapy, biologic therapy and/or hormone therapy to replace various hormones no longer produced or secreted due to the tumour or its treatment.[5] The prognosis for each patient depends on several factors, including the ability of the tumour to be completely removed and the presence of neurological problems or hormonal imbalances caused by the tumour prior to treatment, as well as caused by the treatment itself.

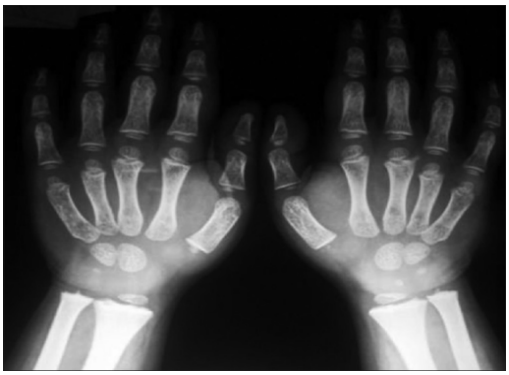

Figure 1: X ray of both hands showing unfused epiphyses and four carpal bones

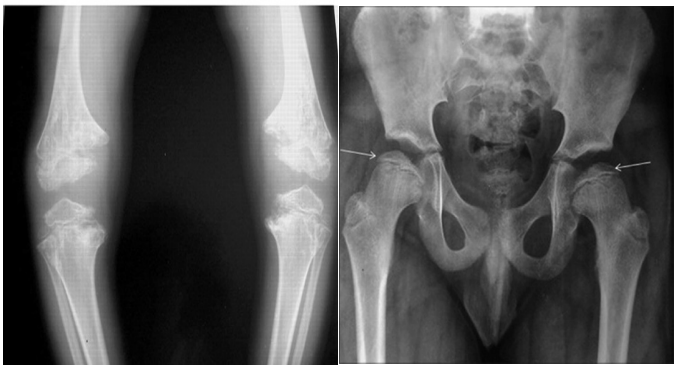

Figure 2: X ray both knee including hip joints showing unfused small epiphyses and metaphyseal irregularity

Most of the problems with hormones and vision do not improve with treatment, and sometimes the surgery can make them even worse, because it may damage the brain structures neighbouring the tumour.[6] Craniopharyngiomas tend to develop again, mostly in the first 3 years after surgery. Overall recurrence rates range from 0-17% after total removal of the tumour and from 25-63% after partial removal of the tumour with radiotherapy.[4]

The case

A 25-years-old man hailing from Demra, Narayanganj, Dhaka with clinical features of loss of vision for 5 months, failure to gain height from the age of 8 years, poor secondary sexual characteristics and mental retardation. He was born with delayed motor and mental milestones of non-consanguineous parents. He could not hear and cannot talk from childhood. On examination, he was disproportionately short statured with a height of 114 cm, weight of 28 kg, BMI of 21.5. He looked dysmorphic, with prominent temporal bones and hypertelorism. He also had proximal myopathy and was unable to walk. On biochemical evaluation, the patient's thyroid-stimulating hormonal (TSH) level was 100 µU/mL (normal reference range 0.47–5.01) with a FT4 of 5.15 pmol/L (normal reference range 9.14–23.18). X ray of his both hands revealed a nonfused epiphysis with a bone age of four (04)

years (Figure 1), and both knee, hip showed epiphyseal and metaphyseal dysplasia (Figure 2). In view of his loss of vision, a CT of the brain was performed, which revealed a large sella and supra sellar mass compressing the optic chiasma suggested craniopharyngioma (Figure 3).

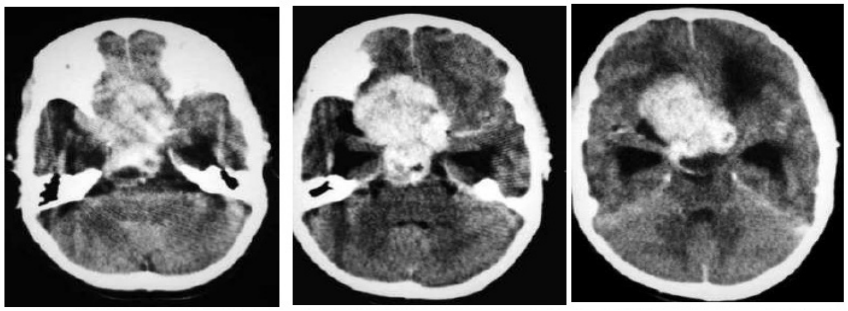

Figure 3: Axial post contrast images of CT scan shows heterogeneously enhancing sella and supra sellar mass

MRI features of brain were consistent with sella-supra sellar complex mass with mass effect having solid and cystic components suggesting craniopharyngioma (Figure 4).

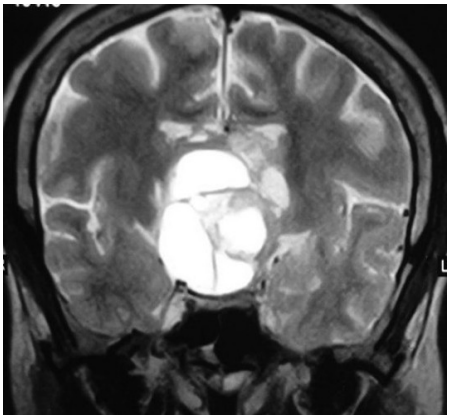

Figure 4: Sagittal and coronal images of MRI of brain shows sella and supra sellar complex mass having cystic and solid components

His hormonal profile revealed a serum cortisol of 67.12 nmol/L (normal reference range 101.2–690.0), serum prolactin 3983.59 mIu/L (normal reference range 945.0-375.0), free testosterone of 11.28 pg/mL (normal reference range 8062-54.69), luteinizing hormone 1.28mIU/mL and follicular stimulating hormone 4.76mIU/mL. USG of neck showed small sized in homogenous thyroid lobes. The patient's short stature and mentally challenged state led to the clinical suspicion of CH. The differential diagnoses considered were pituitary macroadenoma and pituitary pseudotumour caused by long-standing untreated primary hypothyroidism.

DISCUSSION

Congenital hypothyroidism (CH) is a condition of thyroid hormone deficiency present at birth. The prevalence of cretinism in Bangladesh is 0.5% (hilly, 0.8%; flood-prone, 0.5%; and plains, 0.3%) and nearly 69% of Bangladeshi population has biochemical iodine deficiency.[1] Common symptoms include decreased activity and increased sleep, feeding difficulty, constipation, and prolonged jaundice. On examination, common signs include myxoedematous facies, large fontanels, macroglossia, and distended abdomen with umbilical hernia, pseudohy-pertelorism and hypotonia. Neurologic examina-tion findings include hypotonia with delayed reflexes.[9] Affected patients with congenital

hypothyroidism could have sensorineural deafness.[9] In our case, the patient had prior history of poor secondary sexual characteristics and mental retardation and was born with delayed motor and mental milestones. He had deafness and inability to talk since childhood. On examination, he was disproportionately short stature. He looked dysmorphic, with prominent temporal bones and hypertelorism. The diagnosis of congenital hypothyroidism should be and was confirmed by finding an elevated serum TSH and low T4 or free T4 level. In our case, patient had delayed milestone of development, mental retardation, myopathy and walking difficulty. Our hypothyroid patient came for CT and MRI evaluation with history of visual loss for five months. Previous case reports[4,5,8] either revealed association of cranipharyngioma with congenital hypothyroidism only in younger children or showed craniopharyngioma in children causing hypothyroidism as well as other hormonal deficiencies related to pituitary gland. Children or adolescents with craniopharyngioma showed signs of deficiency of growth hormone, gonadotropin, ACTH as well as ADH or TSH dysfuncyion.[10] But no previous reported case showed coexistence of craniopharyngioma in an adult person with prior history of untreated congenital hypothyroidism. Our case was an unusual situation of a large craniopharyngioma detected in an adult with untreated congenital hypothyroidism.

Conflict of interest: None

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