CASE REPORT

Cretinism presenting as a pseudotumour

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SUMMARY

A 43-year-old man from a remote part of India (over 1800 km from our institution), presented with a headache of 3 years duration. He was of short stature, had delayed puberty and was mentally retarded. On evaluation he was detected to have primary hypothyroidism with markedly elevated thyroid-stimulating hormone titres. A CT of the brain revealed a large sellar mass with suprasellar extension into the third ventricle causing obstructive hydrocephalus. Surgical intervention was deferred due to absence of visual impairment and the presence of gross hypothyroidism. The clinical diagnosis of congenital hypothyroidism was confirmed by the absence of radioiodine uptake in the thyroid bed. With thyroid hormone replacement therapy, the 'tumour' underwent significant reduction in size with the resolution of hydrocephalus thereby favouring a potential pituitary pseudotumour. This was an unusual situation of a giant pituitary pseudotumour detected in an adult with untreated congenital hypothyroidism.

BACKGROUND

Short stature with a mentally challenged state is frequently attributed to congenital hypothyroidism (CH). CH is a commonly occurring condition with an estimated incidence of 1 in 2500–2800 live-births in India. Most patients have thyroid ectopia, aplasia or hypoplasia and present with varied clinical manifestations with a 2:1 female to male preponderance. Delayed diagnosis or untreated primary hypothyroidism may be associated with a pituitary pseudotumour due to thyrotroph hyperplasia. The pseudotumour generally regresses with adequate levothyroxine supplementation and hence surgical intervention should not be considered unless the condition is associated with progressive visual impairment. We report a rare case of a giant pituitary pseudotumour in a 43-year-old man of short stature in a mentally challenged state with untreated CH.

CASE PRESENTATION

A 43-year-old man from a rural area in India presented with a headache of 3 years duration, failure to gain height from the age of 8 years, poor secondary sexual characteristics and mental retardation (figure 1). He was born with delayed motor and mental milestones of non-consanguineous parents. There was no visual disturbance or vomiting, hearing impairment or neck swelling. On examination he was disproportionately short statured with a height of 132 cm (<3rd centile) with an upper segment and lower segment ratio of 1.2:1 suggestive of shortened limbs. He looked dysmorphic, with prominent temporal bones and hypertelorism. He also had proximal myopathy and a waddling gait. He did not have a palpable thyroid gland. His visual acuity and fields were normal and the Tanner staging of pubic hair was three with a testicular volume of 10 mL bilaterally. He was diagnosed as in a thyroid dysfunctional

Figure 1 Patient's image showing short stature.

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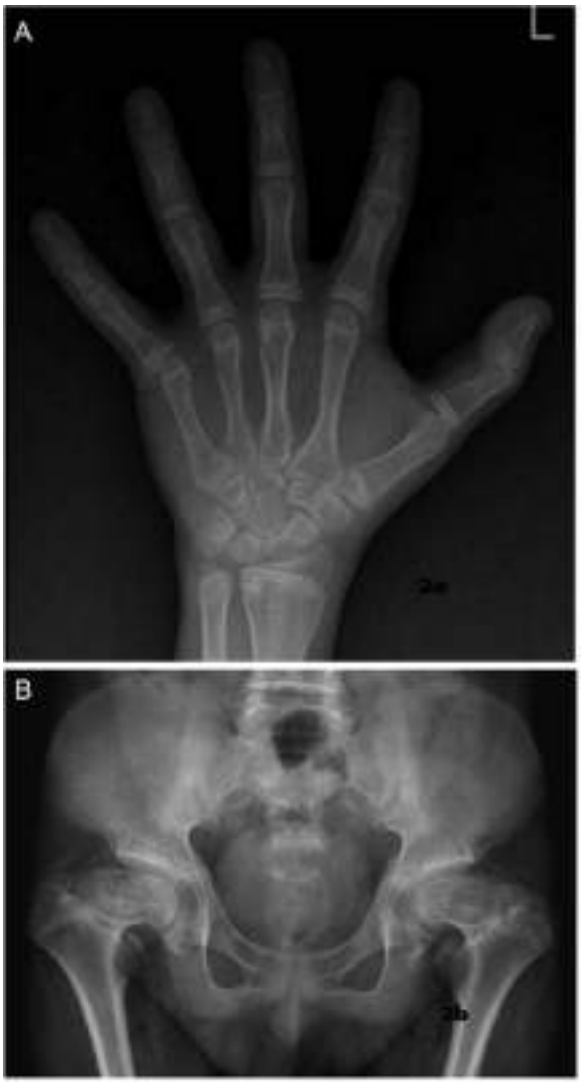

Figure 2 (A and B) X-ray of the patient's left hand showing a bone age of 12 years and dysgenesis of the femoral neck, respectively.

state at 10 years of age; however, he was not put on thyroid hormone replacement therapy.

INVESTIGATIONS

On biochemical evaluation, the patient's thyroid-stimulating hormonal (TSH) level was 775 μIU/mL (0.5–4.5) with a T4 of 1.7 ng/dL (4.5–12). Radiology of his left hand revealed a non-fused epiphysis with a bone age of 12 years (figure 2A) and dysgenesis of the femoral neck (figure 2B). In view of his headache and short stature, a CT of the brain was performed, which revealed a sellar mass of 7.1×5.9×4.2 cm (transverse×craniocaudal×anteroposterior; figure 3A and B) with suprasellar extension into the third ventricle causing obstructive hydrocephalus. His hormonal profile revealed a serum cortisol of 21.3 μg/dL (5–25), growth hormone of 0.23 ng/mL, testosterone of 108 ng/dL (250–900), luteinising hormone 2.6 mIU/mL and follicular stimulating hormone 4.2 mIU/mL. The patient's short stature and mentally challenged state led to the clinical suspicion of CH. His I131 thyroid scintigraphy showed 0.3% after 24 h, which was grossly reduced and suggestive of thyroid gland hypoplasia, confirming the diagnosis of CH.

DIFFERENTIAL DIAGNOSIS

The differential diagnoses considered were non-functioning pituitary macroadenoma, craniopharyngioma and pituitary pseudotumour caused by long-standing untreated primary hypothyroidism.

TREATMENT

The patient was advised surgery, which was deferred due to the presence of the overt hypothyroidism and absence of visual field defects. He was started on thyroxine 100 μg/day.

OUTCOME AND FOLLOW-UP

Following thyroid hormone replacement, he gained height of 4.5 cm over 3 years. A repeat neuroimaging (MRI) performed after 3 years showed a significant reduction in the size of the pituitary mass with resolution of hydrocephalus favouring

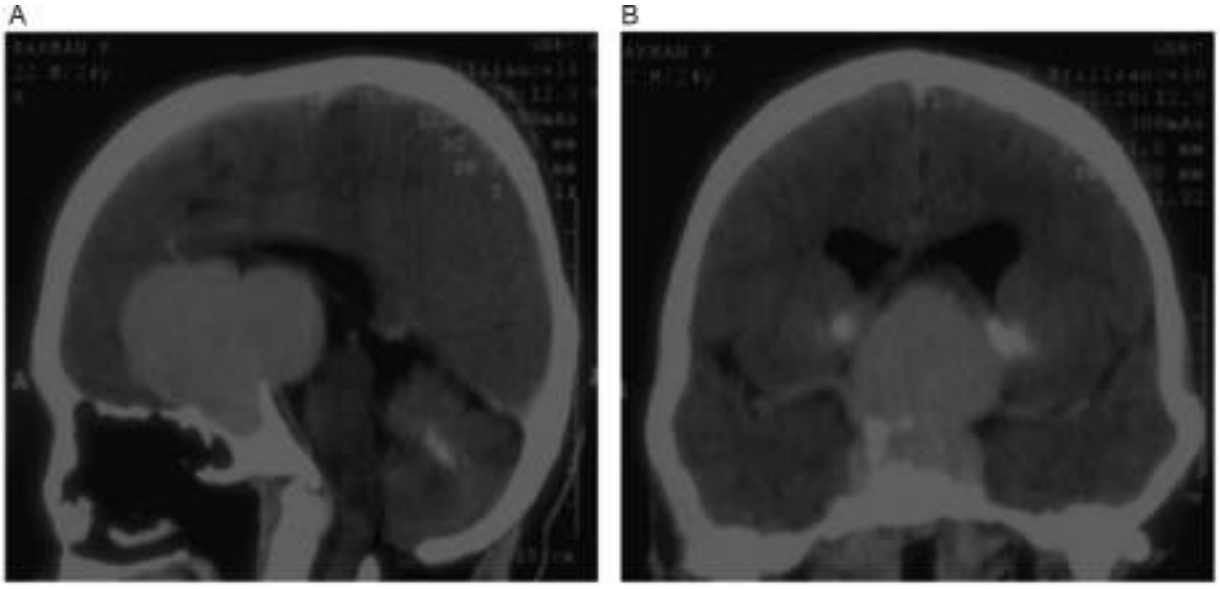

Figure 3 (A and B) CT of the brain, sagittal and coronal images showing a giant 45×58×70 mm (Tr×CC×AP) sellar mass extending into the suprasellar cistern and third ventricle, causing obstruction of the third ventricle and hydrocephalus. Tr×CC×AP, transverse×craniocaudal×anteroposterior.

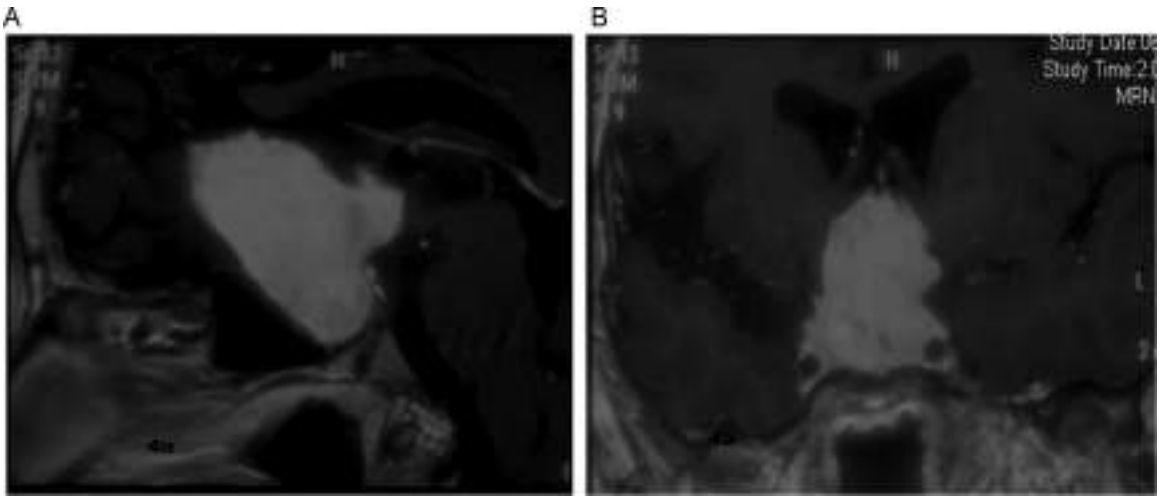

Figure 4 (A and B) T1-weighted sagittal and coronal MRI showing a giant pituitary adenoma but reduced in size to 30×41×42 mm (Tr×CC×AP) compared to the prior CT image. Tr×CC×AP, transverse×craniocaudal×anteroposterior.

thyrotroph hyperplasia causing a pituitary pseudotumour (figure 4A and B).

DISCUSSION

Pituitary hyperplasia secondary to unrecognised and untreated primary hypothyroidism has been reported in adults as well as children. The diagnosis of iodine transport defect was unlikely in our patient in view of absence of goitre and low tracer uptake. Furthermore, it appeared that the patient was unlikely to have an iodine deficiency disorder, where a sizably large goitre would have been present. The radiological diminution of pituitary mass and the mass effects such as visual field improvement after thyroid replacement therapy confirmed it to be pituitary hyperplasia, rather than pituitary adenoma. The present case is unusual, in that, despite the presence of a very large mass, the patient had no visual disturbance, and there was a significant reduction in the sellar mass on treatment with thyroxine with normalisation of TSH favouring a pituitary pseudotumour;[1] the incomplete resolution may suggest the coexistence of a non-functioning adenoma or the occurrence of thyrotrophin adenoma, as seen in earlier case reports.[2 3] Such 'thyrotrophic' adenomas are presumed to occur as the result of protracted pituitary stimulation secondary to long-standing thyroid hormone deficiency.[3] CH is a common preventable cause of a mentally challenged state.[4] Much of the aetiology is due to thyroid ectopia, aplasia or hypoplasia. Disturbances of growth, puberty and sexual function in those with CH, as seen in our participant, are explained by the secondary effects of thyroid hormone deficiency on pituitary function. Severe protracted thyroid hormone deficiency may result in thyrotrophin adenomas of the pituitary gland.[2]

Learning points

- Primary hypothyroidism can be associated with a pituitary pseudotumour on an MRI, as an incidentaloma.
- Treatment with thyroxine can result in regression of a pseudotumour, even if large, and surgical therapy will not be required.
- A universal thyroid hormone screening programme should be performed in all neonates.

Contributors DMM and NT performed the diagnosis and wrote the report. FKJ wrote the report and collected information. MPB initially saw the patient and referred him.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

1 Sarlis NJ, Brucker-Davis F, Doppman JL, *et al*. MRI-demonstrable regression of a pituitary mass in a case of primary hypothyroidism after a week of acute thyroid hormone therapy. *J Clin Endocrinol Metab* 1997;82:808–11.
2 Boyages S, Halpern JP, Maberly GF, *et al*. Effects of protracted hypothyroidism on pituitary function and structure in endemic cretinism. *Clin Endocrinol (Oxf)* 1989;30:1–12.
3 Fatourechi V, Gharib H, Scheithauer BW, *et al*. Pituitary thyrotropic adenoma associated with congenital hypothyroidism. Report of two cases. *Am J Med* 1984;76:725–8.
4 Desai MP, Upadhye P, Colaco MP, *et al*. Neonatal screening for congenital hypothyroidism using the filter paper thyroxine technique. *Indian J Med Res* 1994;100:36–42.