Adult-onset diabetes insipidus caused by congenital midline brain abnormalities

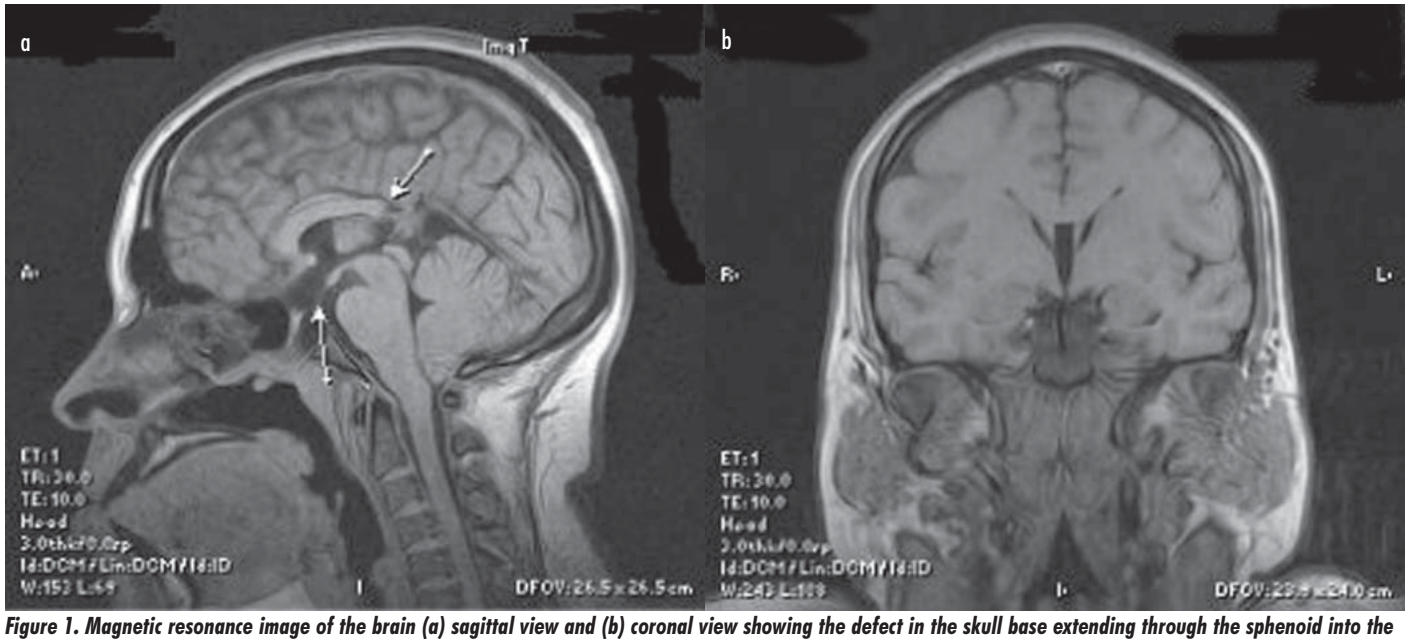

Figure 1. Magnetic resonance image of the brain (a) sagittal view and (b) coronal view showing the defect in the skull base extending through the sphenoid into the nasopharynx with extension extrusion of a sac containing CSF with stretching of the hypothalamus, the optic chiasm and pituitary gland through the defects. Optic nerves appeared normal. Anterior portion of the corpus callosum was present but posterior portion was absent (white arrows).

In transsphenoidal encephalocele there is a defect in the sphenoid bone which can be associated with nasopharyngeal mass or optic nerve abnormalities (Smith et al, 1986; Bodensteiner et al, 2003). An association with agenesis of the corpus callosum has been reported (Bale and Reye, 1976). Most cases are sporadic. Up to 70% of patients with congenital midline malformations of the brain have some degree of pituitary dysfunction (Cameron et al, 1999).

This case highlights the need to fully assess endocrine function in patients with midline congenital structural abnormalities. Radiologists should recommend referral for a specialist endocrine opinion in patients in whom midline brain malformations are detected. BJHM

Case Report

A 30-year-old man presented with a 4-month history of polyuria and polydypsia. There was no significant past medical history and no significant family history as he was adopted.

Clinical examination revealed an obese gentleman with a body mass index of 45 kg/m^2. His blood pressure was 130/80 mmHg and fundus examination was normal. Examination of his external genitalia revealed a small penis and testicular volume of 10 ml each. His full blood count, glucose, electrolytes, liver and bone profile were normal. Serum prolactin = 463 mU/litre, insulin-like growth factor = 15.7 nmol/litre (normal range (NR) =13–45 nmol/litre), free thyroid hormone level = 9.8 pmol/litre (NR = 10–23 pmol/litre), testosterone = 1.7 nmol/litre (NR = 10–40 nmol/litre), cortisol = 484 nmol/litre rising to 880 nmol/litre after Synacthen. Luteinizing hormone level was 0.1 u/litre rising to 1.7 after luteinizing hormone-releasing hormone, thyroid-stimulating hormone = 0.98 mU/litre rising to 8.9 mU/litre after thyrotropin-releasing hormone stimulation. These results suggested partial anterior pituitary deficiency affecting the gonadal and thyroid axis.

Urine failed to concentrate more than 100 mmol/litre during an 8-hour water deprivation test. After desmopressin, urine osmolality rose to 535 mmol/litre. Plasma arginine vasopressin levels were undetectable throughout the test indicating cranial diabetes insipidus. A magnetic resonance imaging scan of the brain (*Figure 1*) showed absence of the posterior portion of the corpus callosum. A transsphenoidal encephalocele was demonstrated with stretching of the hypothalamus, optic chiasm and pituitary gland through the defect.

Bale PM, Reye RDK (1976) Epignathus, double pituitary and agenesis of corpus callosum. *J Pathol* **120**(3): 161–4

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Cameron FJ, Khadilkar VV, Stanhope R (1999) Pituitary dysfunction, morbidity and mortality with congenital midline malformation of the cerebrum. *Eur J Pediatr* **158**(2): 97–102

Smith PJ, Hindmarsh P, Kendall B, Brook CG (1986) Dysgenesis of the corpus callosum and hypopituitarism. *Acta Paeditr Scand* **75**(6): 923–6

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