CASE REPORT

Isolated intraspinal germinoma in a child with congenital adrenal hyperplasia

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Study design: Case report.

Objective: To report an intramedullary germinoma associated with congenital adrenal hyperplasia (CAH).

Method: We report a 14-year-old white male with CAH who was found to have an intramedullary germinoma of the conus medullaris. Presentation and evaluation were for progressive urinary incontinence. A brief review of relevant literature was presented.

Results: Cases where germinomas have occurred simultaneously with CAH have been identified intracranially, but to our knowledge, this is the first case of CAH and intramedullary spinal cord germinoma occurring simultaneously.

Conclusion: Although rare, primary intramedullary germinomas do occur. The association between CAH and spinal cord germinoma should be considered by clinicians.

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Introduction

Germinomas comprise 0.1–3.4% of all intracranial tumors.¹ However, to date, there are only 15 previously reported cases of intramedullary spinal cord germinomas found in the literature, which have been predominantly in the thoracolumbar cord.¹ The majority of these cases are found in the Japanese literature. Germinomas have been found in patients with 21-hydroxylase deficiency with elevated 17-hydroxyprogesterone levels, but these have been intracranial lesions.^{2–4} We present the pertinent history, physical examination findings, radiologic images and treatment of a child with known congenital adrenal hyperplasia (CAH) who presented with loss of bladder function and was found to have an intramedullary spinal cord germinoma.

Case report

A 14-year-old white male with a past history of CAH (21-hydroxylase deficiency) of the nonsalt wasting type, treated with steroid therapy, presented with a 7-month history of

progressive urinary incontinence. His initial presentation with CAH at 5 years of age was with central precocious puberty as a result of early virilization and rapid maturation of the skeleton (advanced bone age). Adrenal steroids were elevated following Cortrosyn stim test. The urinary incontinence was complicated by recurrent urinary tract infections. There were no complaints of upper or lower extremity weakness, bowel incontinence or paresthesias. He had decreased cognitive abilities requiring special education in school. His height and blood pressure were stable. His weight had increased 7 kg over the past 6 months. There was no evidence of increased or decreased reflexes. The remainder of the physical examination was otherwise unremarkable. Urodynamic studies revealed an adequate bladder capacity with diminished compliance and ineffective emptying with a significant residual volume that is, neurogenic bladder. Electrolyte studies were all within normal limits. A multiplanar T1- and T2-weighted magnetic resonance imaging was performed without contrast. A dumbbell-shaped mass at the level of the conus with both an intramedullary and extramedullary component was identified (Figure 1). This mass had high intensity on T2 and intermediate intensity on T1. The magnetic resonance imaging of patient's brain was negative for intracranial pathology. A laminectomy was performed over the lesions and following the opening of the

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Figure 1 T2-weighted spinal MRIs from the patient presented herein. Note the mass (arrow) posterior to the L1 vertebra in the sagittal (a) and axial (b) views. This proved to be a germinoma. MRI, magnetic resonance imaging.

dura and dissection of the arachnoid, the inferior mass was removed en bloc and the superior mass removed through a midline myelotomy. The histology for both masses was germinoma. At 3 months follow-up, the patient continues to catheterize his urinary bladder and his neurological examination is grossly normal.

Discussion

Extragenadal germ cell tumors have an unknown etiology. Current evidence suggests that these tumors could arise from the aberrant migration of germ cells from the yolk sac or from persistent pluripotent cells in primitive rests during development.³ Intracranial germinomas in patients with CAH have been identified in the literature. Locations include the more common pineal and suprasellar regions and the less common basal ganglia, thalamus and cerebellar vermis.⁵ Presenting symptoms of intracranial germinomas are similar to other intracranial masses including seizures, increased intracranial pressure and hemiparesis.² Central nervous system germinomas have a tendency to metastasize usually through the cerebrospinal fluid and the ventricular system. In regard to the present case, a differentiation between metastatic intracranial germinomas to the spinal cord and primary spinal cord germinomas should be made. This could be identified by the lack of hypothalamic hypopituitarism, lack of diabetes insipidus and lack of positive radiological evidence of any intracranial lesions.⁵

The pathobiology of coexistent germinoma and CAH remains to be identified. However, several reports of seminomas, another germ cell tumor and testicular masses in the patients with adrenogenital syndrome exist, and some authorities attributed this occurrence to the hormonal disturbances and the presence of abnormal pluripotential

cells.⁶ To what extent such abnormalities might also have contributed to the present association of germinoma and CAH is unclear.

Conclusion

Although rare, primary intramedullary germinomas do occur. We believe this to be the first report of a primary intramedullary germinoma in a patient with CAH. The possibilities for the occurrence of spinal tumors such as germinoma should be considered in children with CAH. Such a case may suggest aberrant migration of germ cells from the yolk sac or another primitive rest.

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