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CASE REPORT

An unusual presentation of congenital adrenocortical carcinoma: a case report and review of the literature

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Abstract

We describe a case of congenital non-functional adrenocortical carcinoma in a male infant who presented with recurrent pneumonia, paraparesis and sclerotic skeletal metastasis. To the best of our knowledge such presentation has never been reported.

Keywords: *Congenital adrenocortical carcinoma; paraparesis; sclerotic skeletal metastasis.*

Introduction

Adrenocortical neoplasms are rare tumors of children with an incidence ranging from 0.3 to 0.38 per million children less than 15 years old[1]. Clinically, adrenocortical neoplasms are functional in most cases and may present with virilization, precocious puberty, or Cushing syndrome due to increased levels of hormones produced by the tumor[2,3]. These neoplasms are extremely rare in infants and to date only 25 cases (excluding this case) of congenital adrenocortical neoplasms have been reported in the medical literature[1,3–9]. We present a case of congenital non-functional adrenocortical carcinoma in a male infant with a unique clinical and radiological presentation.

Case report

An 8-month-old male baby had been suffering from recurrent episodes of irritability with respiratory discomfort and was noticed to have progressive decrease in the movements of the extremities for almost 2 months. He was diagnosed to have pneumonia in a primary health care center and was treated accordingly for 2 weeks. The weakness in the limbs was attributed to the prolonged illness. The irritability and respiratory distress recovered after initial treatment but the weakness progressed and a week prior to presentation at our hospital he developed a cough with increased effort of breathing and abdominal distension. There was no history of fever, rashes, bladder or bowel complaints, bleeding, cyanosis, alteration in sensorium or seizures. Birth and developmental history were normal. At presentation, the baby was febrile (37.8°C) and tachypnoic (68/min) with tachycardia (170/min). There was retraction of intercostal and subcostal regions and paradoxical movement of the chest wall. Bronchial breath sounds were heard over the right interscapular and scapular region. Neurologically he was conscious, had no cranial palsy or signs of meningeal irritation. There was hypotonia in all 4 limbs with power of 3/5. Abdominal examination revealed hepatomegaly with a liver span of 9 cm. His cardiovascular system was essentially normal except for tachycardia. There was a small 1.5×2 cm nodular swelling in the right paraspinal region at the level of the inferior angle of the scapula, which was present since birth according to the mother. It was initially pea-sized but had increased in size over the last month.

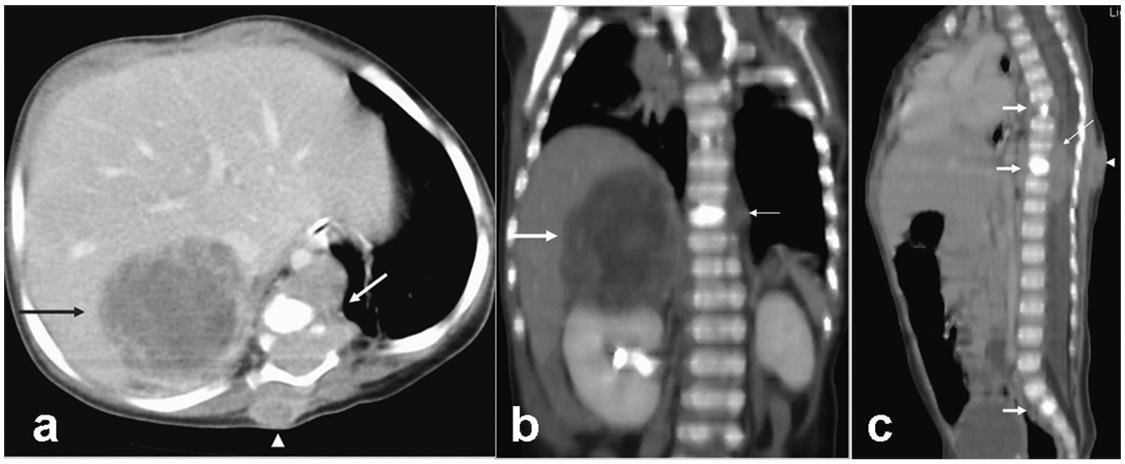

Figure 1 **CECT shows a heterogeneous mass in the right suprarenal region (black arrow) with central necrotic areas, and left paravertebral soft tissue showing intraspinal extradural extension (white arrow). Note abnormal soft tissue mass in the subcutaneous fat and skin (arrow head) (a). Reconstructed coronal image shows a right suprarenal mass (thick arrow) with poorly defined fat planes with the liver. Note the paravertebral soft tissue (thin arrow) (b). Reconstructed sagittal image shows sclerotic vertebral metastasis (thick arrows) with intraspinal soft tissue (thin arrow) and metastasis in the subcutaneous tissues and skin (arrow head) (c).**

Laboratory investigations revealed mild anemia (hemoglobin 9.7 g%); the total leukocyte count, platelets, serum electrolytes and renal parameters were normal.

A chest radiograph revealed air space consolidation in the right lower zone with the right dome of the diaphragm elevated. An ultrasound scan of the abdomen demonstrated a heterogeneous, hypoechoic, lobulated mass in the right suprarenal location displacing the right kidney inferiorly and indenting the liver superiorly. There was no evidence of calcification or any obvious cystic change.

Contrast-enhanced computed tomography (CECT) chest and abdomen showed a $4.4 \times 4.7 \times 5.25$ cm heterogeneous mass lesion in the right suprarenal region with central necrotic areas displacing the inferior vena cava anteriorly (Fig. 1a,b). A soft tissue mass measuring approximately $1.5 \times 1.5 \times 2.25$ cm was present in the left paravertebral region at D9 level associated with erosion and sclerosis of the adjacent vertebral body. The soft tissue mass was extending through the ipsilateral neural foramina with an intraspinal extradural component from D8 to D10 levels compressing the dorsal spinal cord (Fig. 1a,c). Sclerotic vertebrae were also seen at multiple other levels (Fig. 1a,c). In addition, soft tissue of similar density was seen in the subcutaneous fat and skin from D8 to D10 levels in the midline posteriorly (Fig. 1a,c). There was complete collapse and consolidation of the right lower lobe.

Ultrasound-guided fine-needle aspiration biopsy (FNAB) from the right adrenal mass revealed a highly cellular aspirate that was predominantly dissociated with a few poorly cohesive clusters of markedly pleomorphic tumor cells (Fig. 2a). The cells were polygonal in shape, had abundant finely vacuolated cytoplasm with frayed cell margins. The nuclei were rounded, eccentrically placed and showed variable anisokaryosis and prominent nucleoli. Occasional mitotic figures were also seen. The background showed necrosis. No spindle cells or ganglion-like cells were noted. No Homer-Wright rosettes or neurofibrillary material was seen. The above negative findings excluded the possibility of phaeochromocytoma and neuroblastoma. The aspiration smears from the posterior midline soft tissue nodular mass showed a similar picture, hence confirming metastasis from the same tumor (Fig. 2b). In view of the high cellularity, background necrosis, occasional mitosis and metastatic deposit, the overall features were of malignant adrenocortical carcinoma.

The parents were counseled in detail about the widespread nature of the illness and the prognosis. The family declined any aggressive treatment and so the baby was sent home with palliative care advice.

Discussion

Adrenocortical neoplasms are rare tumors in children with an incidence ranging from 0.3 to 0.38 cases per million in children less than 15 years old, and only 25 cases of congenital adrenocortical neoplasm have been reported in the medical literature to date, 19 of which were adrenocortical carcinomas[1,3–9]. The term adrenocortical neoplasm is preferred in children as, unlike adults, there are no histopathological criteria for distinguishing adenoma from carcinoma[2,3]. The presence of metastasis or vascular invasion indicates malignancy[3,10]. These tumors are reported to be associated

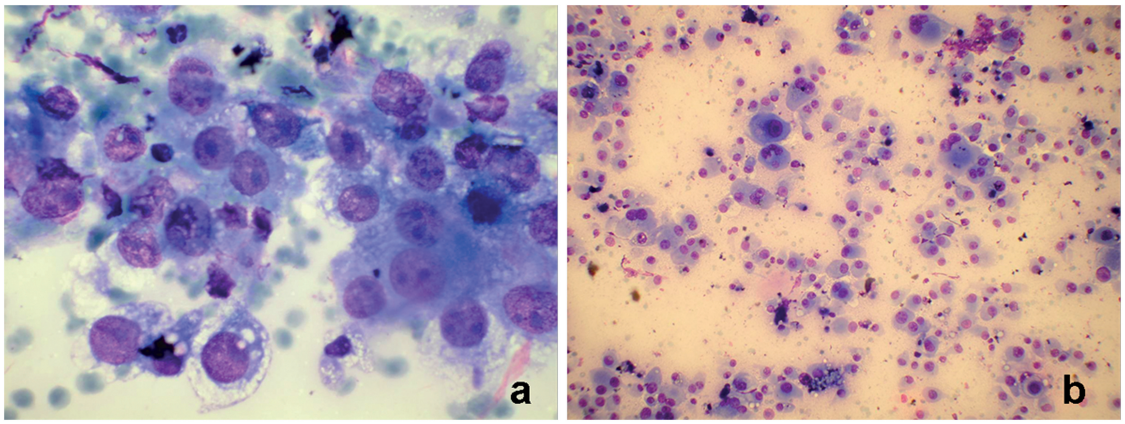

Figure 2 **FNAB from the right adrenal mass shows poorly cohesive clusters of pleomorphic tumor cells with large vesicular nuclei and macronucleoli, abundant fragile vacuolated cytoplasm. Occasional mitosis is noted. May-Grunwald Giemsa (MGG) stain, original magnification × 400 (a). Aspiration smears from the subcutaneous nodular mass shows dissociated malignant cells with similar morphology to the adrenal lesion. MGG, original magnification × 200 (b).**

with hemihypertrophy, Beckwith-Wiedemann syndrome, familial adenomatous polyposis and Li-Fraumeni syndrome[1–3]. Recently overexpression of IGF-II and H19 genes in Beckwith-Wiedemann syndrome, mutations in the APC gene and p53 tumor suppressor gene in familial adenomatous polyposis and Li-Fraumeni syndrome, respectively, have been proposed for genesis of adrenocortical carcinomas[11].

Clinically, adrenocortical neoplasms are functional in most cases, and usually present with virilization, precocious puberty, or Cushing syndrome due to increased levels of steroids or androgens produced by the tumor[2,3]. A palpable abdominal mass is the next most common presentation[3]. Of the 25 published cases, 16 patients presented with virilization, 3 patients each with Cushing syndrome and arterial hypertension, and 11 patients with palpable abdominal mass. Eight patients had metastasis at the time of presentation. The usual metastatic sites were lungs, liver, kidneys, regional lymph nodes, and rarely to skin, peritoneum, or ovaries. Twelve patients were girls, 10 were boys, and the gender of 3 patients was not reported. The right adrenal was involved in 13 cases, left in 7, bilateral in 1 and laterality was not reported in 4 cases[3–9]. Three cases were diagnosed on antenatal ultrasonography: adenoma in 2 cases and 1 case of bilateral adrenal carcinoma on pathological evaluation[3,8,9]. Our case was a male who presented with recurrent pneumonia and paraparesis, without any signs of hypercortisolism or hyperaldosteronism or a palpable abdominal mass. On imaging there was skin, extradural and sclerotic skeletal metastasis without any vascular invasion. This presentation has never been reported in the past and thus makes this case unique.

Radiological studies are essential for evaluation and establishing the diagnosis of adrenocortical neoplasms. Radiography has a limited role, however it can reveal an abdominal mass or calcification[12]. On sonography the appearance of adrenocortical neoplasms is variable; small lesions are usually round to oval, homogeneous, solid masses isoechoic to the renal cortex but can be uniformly hypoechoic or hyperechoic; large tumors are heterogeneous with central necrotic areas and can mimic multicystic kidneys[6,8,13,14]. A complex, predominantly hyperechoic pattern with radiating linear echoes, described as the scar sign has also been reported in carcinoma[13]. In addition, tumor thrombus in the inferior vena cava, right atrium, renal and adrenal veins if present, can be detected on color Doppler imaging[15].

On CT scans, adrenocortical tumors are typically circumscribed, appear variably heterogeneous due to hemorrhage and necrosis, and may display a thin capsule-like rim. Intratumoral calcification, which can be seen in about 20% of cases, can be reliably detected[13,14,16]. More specifically, CT demonstrates the exact extent of the lesion, its relation with adjacent organs, status of the inferior vena cava and presence of metastasis.

MRI is a complimentary technique for the diagnosis and there are no signal intensity characteristics that can differentiate adenoma from carcinoma. Multiplanar capabilities of MRI can readily demonstrate the relationship of the tumor with the adjacent organs and more specifically detect inferior vena cava tumor thrombus[14]. On imaging, the demonstration of a thin tumor capsule and a stellate central zone of necrosis with evidence of hormonal function can differentiate adrenocortical carcinoma from neuroblastoma[17].

Differential diagnosis of adrenal masses in children includes neuroblastoma, pheochromocytoma, adrenal hemorrhage, and rarely, metastases. Large adrenal tumors at times may be difficult to distinguish from renal tumors such as nephroblastoma, mesoblastic nephroma, renal cell carcinoma, clear cell sarcoma, and

rhabdoid tumor of the kidney[18]. However, clinical, biochemical and radiological features of these entities are different from adrenocortical neoplasms in most cases. In equivocal situations, guided FNAB can be attempted.

Surgery is the mainstay of treatment of adrenocortical neoplasm, however large tumor size and the presence of metastasis are poor prognostic indicators. Complete resection of the adenoma is associated with favorable outcome and can be curative[1,3,5,7].

To conclude, congenital (which is very likely to be the case here), non-functional adrenocortical carcinomas may remain clinically occult and present late in the disease process. The clinical presentation may be entirely due to metastasis as in the index case who presented with paraparesis.

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