CASE REPORT

Anterior mediastinal alveolar rhabdomyosarcoma in an infant: rare site for a common paediatric tumour

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Rhabdomyosarcoma is a common paediatric soft tissue tumour. However, the anterior mediastinum is an extremely rare site for its occurrence. This report describes the imaging and histological findings of such a tumour in a 4-month-old boy.

Case report

A 4-month-old boy was admitted as an emergency in December 2010 because his mother noticed he had been breathing noisily for 1 month, and that such breathing had become increasingly severe over the last 4 days. On the day of admission, the patient's mother noticed her son had an episode of cyanosis that subsided spontaneous after a few minutes. Physical examination revealed the presence of stridor and mild tachypnoea, but there were no added sounds upon auscultation of the chest. Cardiovascular and abdominal examination yielded nil abnormal. Chest radiography revealed a large well-defined mediastinal mass compressing the trachea (Fig 1a). Computed tomography (CT) revealed a large heterogeneously enhanced anterior mediastinal mass measuring approximately 7 cm x 3 cm x 5 cm and extending to the lower neck (Fig 1b), but there was no clear fascial plane between the tumour and the displaced major intrathoracic vessels, which included the superior vena cava and the brachiocephalic vein (Fig 1c). Moreover, no cervical or mediastinal lymphadenopathy or distant metastases were detected.

Incisional biopsy of the cervical portion of the mass under general anaesthesia yielded histological features of rhabdomyosarcoma (Fig 2). No teratomatous component was identified, and molecular study found the presence of a PAX3/FOXO1 fusion transcript and confirmed the diagnosis of alveolar rhabdomyosarcoma.

Chemotherapy entailing a regimen of vincristine, actinomycin D and cyclophosphamide was started 1 week post-biopsy. A follow-up CT study 3 months later showed a markedly shrunken tumour, which remained attached to the major intrathoracic vessels. An operation performed around 4 months after the diagnosis confirmed the presence of a residual tumour over the superior aspect of the anterior mediastinum. Intra-operatively the tumour was noted to invade the brachiocephalic vein and the left common

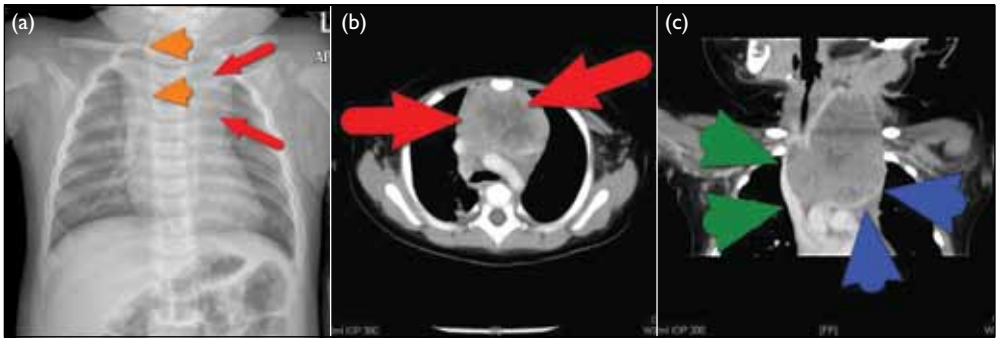

FIG 1. (a) An anteroposterior chest radiograph shows a large well-defined anterior mediastinal mass (red arrows) causing compression and rightward deviation of the trachea (orange arrowheads). No lung mass, pleural effusion, or focal destructive bone lesion was evident. (b) An axial computed tomography (CT) image obtained after intravenous injection of the contrast medium shows the anterior mediastinal mass, demonstrating heterogeneous enhancement (red arrows). No internal fat or calcification was evident. The mass abutted the aortic arch and the superior vena cava. (c) A coronal reformatted CT image obtained after injection of contrast medium demonstrates the craniocaudal extent of the anterior mediastinal mass, which extended to the lower neck, abutted and displaced the brachiocephalic vein (blue arrowheads) and the superior vena cava (green arrowheads)

Key words
Mediastinal cyst; Neoplasms; Pediatrics; Rhabdomyosarcoma, alveolar

Hong Kong Med J 2013;19:458-9
DOI: 10.12809/hkmj133714

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carotid artery. Tumour debulking was performed and the patient received postoperative radiotherapy and still undergoes chemotherapy.

Discussion

Rhabdomyosarcoma is a malignant tumour that displays features of differentiated striated muscle.[1] It is the most frequent soft tissue sarcoma of children and young adults,[1] and occurs in any anatomical location where there is skeletal muscle and in some locations where skeletal muscle is not normally found.[2] Its four histological subtypes include embryonal, alveolar, pleomorphic, and mixed. Approximately 20% of children with rhabdomyosarcoma have the alveolar subtype.[2]

The mediastinum is a rare site of rhabdomyosarcoma, which involves the thymus or the anterior mediastinum, and most commonly occurs as a complication of a teratoma[3] or as a thymic carcinosarcoma.[4] Primary mediastinal rhabdomyosarcomas unassociated with germ cells, teratomatous or malignant epithelial components are extremely rare.[5,6] Such tumours tend to have a large size and local invasion at the time of diagnosis. They have been reported to be highly aggressive, and usually give rise to frequent recurrences and rapid dissemination.[6] More studies regarding the treatment and prognosis of these tumours in infants are necessary, in view of very limited information currently available.

嬰兒中的前縱隔肺泡橫紋肌肉瘤：一個發生在罕見部位的常見小兒腫瘤

橫紋肌肉瘤是小兒常見的軟組織腫瘤。然而，前縱隔是一個極為罕見的腫瘤部位。本文報告一宗發生在一名四個月大的嬰兒身上的橫紋肌肉瘤病例，並描述了其成像和病理檢查結果。

Acknowledgement

I would like to express my great thanks to Dr Anthony WH Chan (Attending Pathologist, Department of the Anatomical and Cellular Pathology, Prince of Wales Hospital, Hong Kong) for providing the invaluable histological slides of this case report.

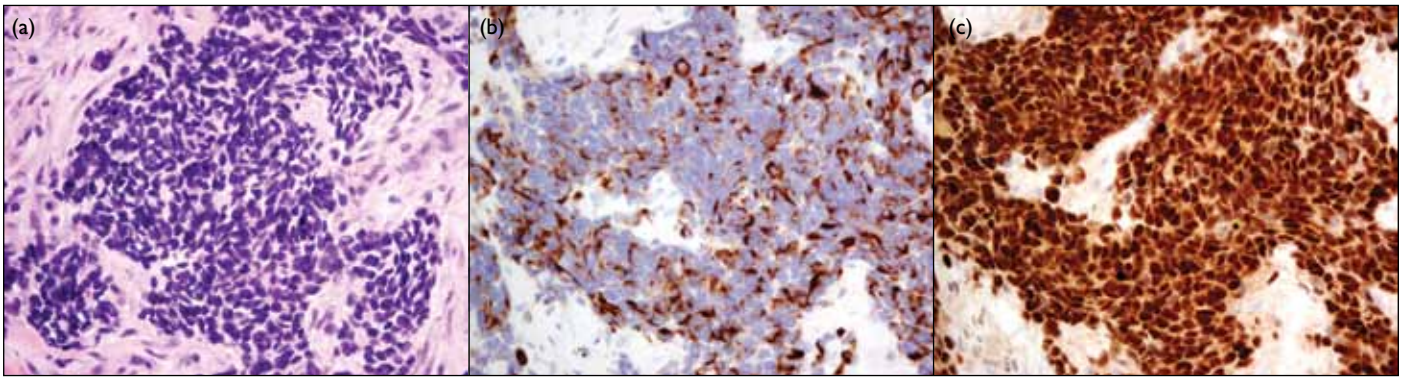

FIG 2. (a) Microscopy shows infiltrative clusters of tumour cells in the fibrous stroma (H&E, x 400). The tumour cells exhibited hyperchromatic nuclei, pleomorphism, and stippled chromatin. Histo-immunochemical staining sections show the tumour was reactive to muscle markers: (b) desmin and (c) myogenin (x 400)

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