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 spontaneously 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. Intraoperatively the tumour was noted to invade the brachiocephalic vein and the left common
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,\(^2\) 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.

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**References**

3. Moran CA, Suster S. Primary germ cell tumors of the mediastinum: I. Analysis of 322 cases with special emphasis on teratomatous lesions and a proposal for histopathologic classification and clinical staging. Cancer 1997;80:681-90. [CrossRef]