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INTERACTIVE CLINICAL CASE

Unpainful mass - a sad end

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Abstract

A soft tissue thoracic mass differential diagnosis includes congenital, inflammatory/infectious and neoplasic causes. A neoplasic aetiology - in which sarcoma is included - can be a challenging diagnosis, given its unpainful and slow growth nature. Despite being unusual, sarcoma must be considered in the presence of a gradually enlarging unpainful mass in the trunk or extremities, as this is the most common form of presentation. Quite often, the histologic exam of a soft tissue mass is essential for diagnosis and treatment planning.

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Question:

A 4-month-old female infant was evacuated from Cabo Verde for further investigation due to a thoraco-cervical, gradually enlarging, painless mass, present since birth of unknown aetiology.

Apart from the 9cm diameter mass swelling on the collarbone, without cutaneous inflammatory signs, physical examination was otherwise unremarkable.

Thoracic CT scan showed an intra and extra-thoracic mass with invasive features and superior mediastinic origin. It also revealed right axillary lymph nodes and no signs of metastasization (Figure 1). MRI showed an heterogeneous tumour lesion, 8x7x5cm along the greatest axis, invading the chest wall, the small pectoral muscle pushing the great pectoral muscle anteriorly and the presence of multiple cervical ganglionary lineups (Figure 2). Excisional biopsy was performed.



Figure 1. Contrast thoracic CT showing an intra and extrathoracic component mass with invasive features. The intrathoracic component is of superior mediastinic origin and adheres to the great pectoral muscle. Note the organized mass with thick and vascularized septa.

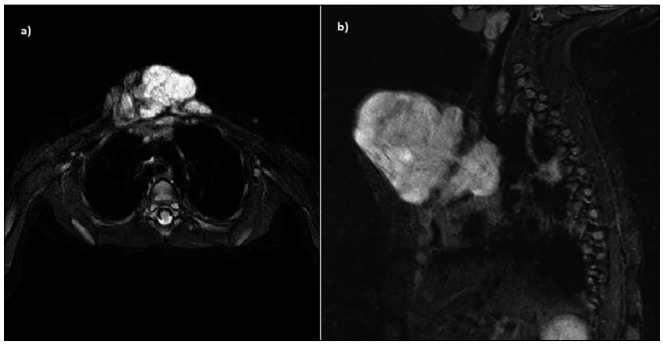


Figure 2. Magnetic resonance images of the thorax. Axial (a) and sagital (b) showing an heterogeneous tumoral lesion (cystic areas, haemorrhagic component, and calcifications), 8x7x5cm along the greatest axis of local aggressive features, and the presence of multiple cervical ganglionary lineups.

What is your diagnosis?

a) tuberculosis
b) soft tissue sarcoma
c) Hodgkin disease
d) histoplasmosis

Answer

The histology revealed a round cells malignant neoplasia with 6 mitosis per 10 high power field, without signs of necrosis. A diagnosis of a round cells undifferentiated sarcoma of intermediate grade malignancy was then made. Tumour cells were positive for CD99, a marker of aggressiveness. Gene rearrangement of BCOR, CIC, EWSR1 and ETV6 was not detected, a common feature, and an important diagnostic tool in sarcomas. Mantoux skin test, IGRA T-SPOT and HIV were negative. She began chemotherapy with tumour mass reduction followed by complete excision of the tumour mass. Despite more chemotherapy cycles together with radiotherapy, intracranial masses suggestive of metastases were documented and, unfortunately, 10 months after the diagnosis, the patient died.

DISCUSSION

Soft tissue sarcomas (STT) are a rare and heterogeneous group of malignant tumours of mesenchymal origin. They include more than 100 different histologic subtypes and comprise about 12% of paediatric cancers[1-6].

Most STT occur sporadically and not from a pre-existing benign lesion; however, most cases have unknown aetiology. Predisposing factors have been identified as exposure to radiation therapy or chemotherapy and chemical carcinogens[1-3]. Genetic syndromes such as Li-Fraumeni syndrome and neurofibromatosis type I are known to be associated with STT[1,2,4,5]. In our case report, no risk factors were found.

The most frequent symptom of a soft tissue sarcoma is a gradually enlarging, unpainful mass[1,2,4] like our patient. Patients may also have symptoms related with mass compression in surrounding structures[1].

Most of STT affect the extremities but they can occur, virtually, anywhere[1,2,4].

The differential diagnosis of soft tissue thoracic mass includes congenital, inflammatory/infectious and neoplasic causes.

In our case, considering the local aggressive aspects, the diagnostic hypothesis of lymphoma, thymic carcinoma, thymoma and germ cells mediastinal sarcoma were considered.

It is essential to differentiate between the various subtypes of sarcoma for both prognosis and treatment; therefore, histologic examination is mandatory[1-5].

Imaging is used to help define the extent of the tumour, determine the presence/absence of metastatic disease and plan surgical treatment[2,4].

Although surgery remains the mainstay of treatment, curative treatment usually cannot be achieved without adjuvant treatment. Radiation therapy is considered in the presence of residual disease and regional spread. Chemotherapy is associated with reduced risk of relapse and improved overall survival[2-4].

Data related to prognosis is limited in paediatric STT but negative factors include disease extension, higher tumour size and higher grade. Survival rate depends on STT type ranging from 15% in metastatic disease to 90% in low-grade tumours[3].

Given its unpainful and slow growth nature, its diagnosis can be a challenging process and a delay in diagnosis is common[1].

Although unusual, sarcoma must be considered in the presence of an unpainful mass in the trunk or extremities.

Although important insights into STT *pathogenesis* and new molecular diagnostic tools have emerged, most of it is unavailable in low-income countries, which are almost exclusively reliant on patient evacuation. Despite being evacuated and given prompt treatment, this infant eventually died. It highlights the importance of multidisciplinary work with both medical and surgical specialities as well as the importance of prompt evacuation and adequate evaluation, which, in our case, took 5 crucial months.

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