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CASE FOR DIAGNOSIS

A 20-year-old male with thoracic pain and a lower thoracic mass

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CASE REPORT

A 20-yr-old Caucasian male construction worker had a previous history of a road traffic accident 3 yrs before presentation. A computed tomography (CT) scan of the thoracic spine was carried out to exclude vertebral damage. No evidence of vertebral bone damage or other lesions was seen, and the patient recovered without sequelae.

A week before presentation, he noticed a stabbing pain in his right hemithorax, without dyspnoea. The pain persisted, and the patient was referred, by his general practitioner, for a chest radiograph (fig. 1). Based on these results, the patient was referred to a general hospital for further diagnostic tests. A CT scan of thorax and abdomen (not shown) revealed a large mass, which was interpreted to arise in the right upper abdomen, probably originating from the liver. A malignant tumour, or a metastatic lesion, was suspected and the patient was referred to University Medical Center Groningen (Groningen, The Netherlands).

The patient did not suffer from dyspnoea, cough or haemoptysis and there was no history of fever, weight loss, fatigue or excessive sweating. There were no neurological or gastrointestinal complaints. The patient was a nonsmoker, and did not use any medication.

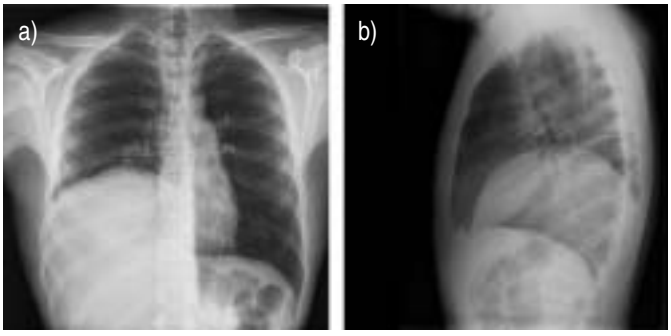

FIGURE 1. a) Postero-anterior and b) lateral chest radiographs at presentation.

On physical examination, a healthy appearing, haemodynamically stable young male, of normal posture was seen. On percussion, a dull sound was found in the right lower zone of the chest. Auscultation revealed normal cardiac sounds without murmurs, and normal breathing sounds on the left side and upper right side of the chest. Abdominal examination revealed no palpable masses or other abnormalities. No palpable lymph nodes were present. Additional physical examination revealed no other abnormalities.

Laboratory tests only showed a slightly elevated serum alkaline phosphatase of 174 $U \cdot L^{-1}$ (normal value: 13–120 $U \cdot L^{-1}$). Serum lactate dehydrogenase, α-fetoprotein and β-human chorionic gonadotropin values were all normal; therefore, an extra-gonadal germ cell tumour was unlikely.

On revision of the CT scan, there was doubt regarding the hepatic origin of the mass. Therefore, abdominal ultrasonography was performed. No focal lesions in the liver parenchyma were observed, and the liver blood flow appeared intact. In the right thoracic region, a mass was seen with variable echogenicity and rich vascularisation. Due to the high degree of vascularisation observed on the abdominal ultrasonography, no percutaneous biopsy was performed. A CT angiography was performed: first, to narrow the differential diagnosis, and, secondly, to provide the thoracic surgeon with more detailed information about vascularisation (fig. 2). At bronchoscopy, no endobronchial abnormalities were seen. Cytological examination of the bronchial lavage showed no signs of malignancy. A bone scan was normal.

Exploratory thoracotomy revealed a tumour originating from the right dorsal region, lateral from the vertebral column. A surgeon was able to remove the tumour (figs 3 and 4).

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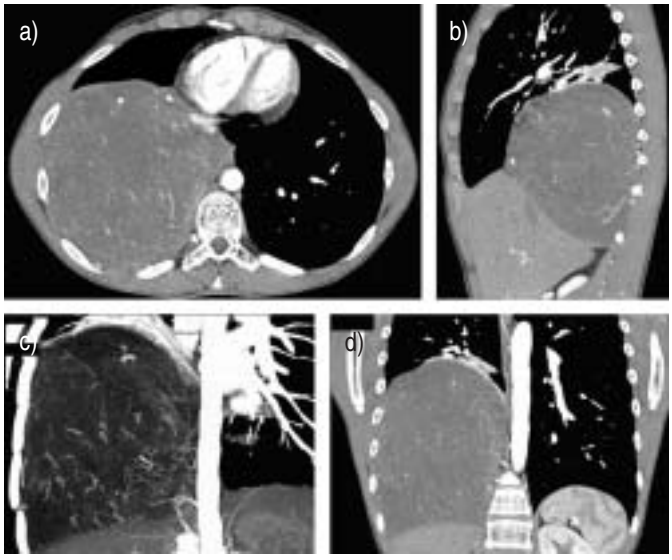

FIGURE 2. Computed tomography angiography of thorax and upper abdomen. Representative images are shown in a) transversal, b) sagital, and c and d) frontal views. A computed reconstruction is shown in c.

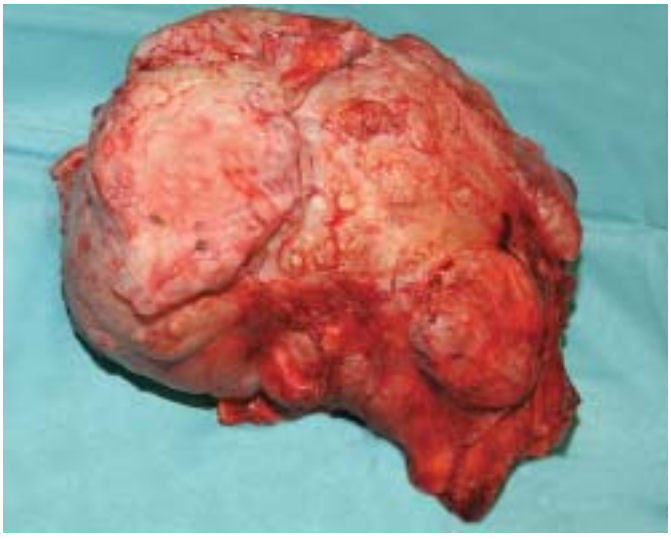

FIGURE 3. Resection specimen.

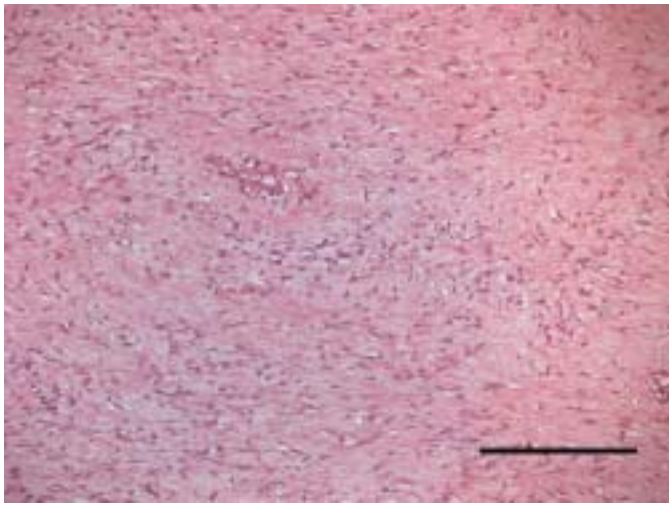

FIGURE 4. Representative microphotograph of the tumour with haematoxylin-eosin staining. Scale bar=200 μm.

BEFORE TURNING THE PAGE, INTERPRET THE CHEST RADIOGRAPHS, THE COMPUTED TOMOGRAPHY ANGIOGRAPHY, THE RESECTION SPECIMEN AND THE HISTOLOGY SLIDE, AND SUGGEST A DIAGNOSIS.

INTERPRETATION

Chest radiograph at presentation

The chest radiograph showed a loss of volume of the right lung (fig. 1). On the lateral view, the density extending from the right posterior ribs does not reach the sternum, suggesting a supradiaphragmatic lesion.

Computed tomography angiography

In the right hemithorax, a relatively hypodense mass with large blood vessels was seen, compressing the normal lung and displacing the right hemidiaphragm caudally (fig. 2). A relationship with the thoracic aorta could not be seen, nor any aberrant artery, suggesting a lung sequestration. Blood vessels originated from the intercostal arteries. No enlarged lymph nodes or suspect metastatic lesions were found.

Resection specimen

At examination, the tumour weighed 1,700 g, with reconstructed dimensions of $19 \times 17 \times 6$ cm with a resected peduncle of 6×4 cm, visible on the right lower side (fig. 3). The tumour had a lobular aspect, and was surrounded by a thin membrane with small fluid-filled cysts.

Histology

Microscopy evaluation was used to diagnose a mesenchymal tumour with small spindle cells with some variation in shape, but without atypia, mitotic figures or other malignant characteristics in a fibrous background (fig. 4). The tumour extended into the surgical margins of the resected peduncle.

Using immunohistochemistry, tumour cells were only positive for vimentin, and lacked CD34, S-100, ALK-1, Bcl-2, cytokeratins, actin and desmin staining. Few positive nuclei were seen using Ki-67 staining. The morphology, in combination with immunophenotype, is compatible with a desmoplastic fibroma of the pleura, also known as a desmoid tumour.

Diagnosis: Intrathoracal desmoid tumour with microscopically incomplete resection.

Clinical course

Although the resection was microscopically incomplete, the patient was not treated with additional surgery or adjuvant radiotherapy (see Discussion). Instead, regular magnetic resonance imaging (MRI) examinations were applied to observe any local recurrence. During 18 months of follow-up, consecutive MRIs did not show any sign of recurrence. The patient has recovered without sequelae and has caught up with his daily work.

DISCUSSION

Desmoid tumours, also known as aggressive fibromatosis, are slowly growing fibroblastic neoplasms arising from fibroblastic stromal elements. Although desmoid tumours do not metastasise, they tend to be locally invasive. The aetiology is not exactly known, but the association with familial adenomatous polyposis coli (FAP), as well as with previous trauma, has been extensively described [1, 2].

Desmoid tumours are very rare; the incidence is between 2–4 per million [3]. The primary location for desmoid tumours is extra-abdominal, with the limb girdle and extremities most commonly involved, followed by the chest wall [4]. Abdominal wall desmoid tumours are mostly seen in females, especially during pregnancy [5]. Intra-abdominal desmoids are seen in correlation with FAP. Slightly more females than males are affected. The age at diagnosis is usually between 15–60 yrs.

The present study reports a case of a large intrathoracal desmoid tumour. This type, and more so the pleural origin, is very rare and has been reported in <20 cases [6]. There may be a relationship with the traumatic chest injury which the patient suffered 3 yrs prior to presentation.

The initial differential diagnosis of large, pedunculated, intrathoracal tumours includes both mostly benign and, less frequently, malignant lesions. The most frequent malignant pleural tumour is malignant mesothelioma, which is, in general, a diffuse pleural proliferation and hardly ever presents as a pedunculated mass. Although in the past, several entities were included in the group of mesotheliomas, at present the designation mesothelioma is used for neoplastic proliferation of mesothelial cells and not for proliferation of other cells of the pleura [7]. In this case study, there were no indicators for malignancy, considering the complete lack of atypia and the scarcity of mitoses. This morphology, together with negative cytokeratin staining, makes a diagnosis of mesothelioma unlikely. Consequently, a malignant solitary fibrous tumour of the pleura, recently presented in an article in the *European Respiratory Journal* [8], could also be excluded on the basis of morphology and immunohistochemistry [8].

With respect to benign tumours to be considered in the differential diagnosis, a schwannoma of the paravertebral nerves could be excluded on clinicopathological grounds, as absence of relations to intervertebral structures and the negative S-100 staining [9]. A pulmonary sequestration was unlikely, due to anatomical presentation [10]. A solitary fibrous tumour (SFT), developing from the pleura, belongs to the differential diagnosis. Obsolete and confusing terms for SFT are localised or benign fibrous mesothelioma or benign localised fibroma [11]. A vascular peduncle may be present, especially in larger SFTs [12]. The morphology and the negative CD34 and Bcl-2 immunostaining made this diagnosis unlikely [13, 14]. The morphology of the tumour, together with the distinctive immunohistochemical results, was compatible with a desmoid tumour.

The primary management of a desmoid tumour consists of complete resection. For patients deemed inoperable, primary radiotherapy treatment is a curative option. In two recent large series, no difference was observed in disease-free survival between microscopically positive and negative resection margins after primary resection [4, 15]. However, some older and smaller studies showed a small decrease in disease-free survival time for microscopically positive margins as compared with disease-free resection margins [16]. Despite complete resection, the rate of local recurrence is ~30% [16]. Most local recurrences develop within 2 yrs after resection, but a time to recurrence of >10 yrs has also been described [15].

The use of radiotherapy as adjuvant therapy is, as yet, not substantiated. For incomplete resected tumours, the recurrence rate decreases from 39 to 25% after adjuvant radiotherapy, as was presented in a review by NUYTTENS *et al.* [16]. Adjuvant radiotherapy was not a predictor for disease-free survival in

other studies [4, 15]. Moreover, radiation-related complications, such as radiation pneumonitis, soft tissue necrosis and/or fibrosis, secondary malignancies and skin problems are well known. The application of radiotherapy for incomplete resection, therefore, remains an issue for debate. The current authors chose not to let the patient be irradiated and to follow-up the patient clinically and radiologically.

Local recurrence can be treated with re-operation or local radiotherapy. In cases with incomplete resection the radiation dose should be $\geqslant$50 Gray to decrease the risk of local recurrence [17]. The disease-free survival for patients with recurrent disease is less than that for primary desmoid tumours [15].

The benefit of additional surgery in case of positive margins is doubtful. Hence, gain in survival has not been demonstrated to be related to negative margins. Therefore, extensive surgery can be postponed until the presence of recurrent lesions [15].

Early detection of tumour relapse gives a higher chance of nonmutilating surgical resection. Follow-up should be performed with a sensitive imaging technique. MRI was preferred in the patient, as it is very specific for detecting slight differences of density between desmoid tumours and surrounding structures. Baseline MRI was chosen to be carried out at 3 months after the operation, because the clips left at operation needed to be firmly grown into the surrounding tissue. As desmoid tumours are slowly growing, 3-month intervals between MRIs were considered appropriate during the first 2 yrs, after which the intervals will become larger. At present, 18 months after diagnosis, the patient is in an excellent condition and there are no signs of relapse.

In conclusion, a patient with a large intrathoracal desmoid tumour, with microscopically incomplete resection, was presented. The differential diagnosis, the therapeutic options and the ways of follow-up were discussed in detail.

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