Primary monophasic mediastinal, cardiac and pericardial synovial sarcoma: a young man in distress

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A 19-year-old male was admitted because of exertional dyspnoea. The imaging studies revealed epicardial, pericardial and mediastinal masses. The tumours could not be resected through a minor thoracotomy, only biopsies could be taken. Analyses led to the final diagnosis of a monophasic synovial sarcoma. The patient preferred a conservative and palliative approach. Three months later he died at home. Autopsy demonstrated dramatic extension of the tumour masses. We conclude this report with a discussion on primary cardiac tumours. (*Neth Heart J* 2007;15:226-8.)

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In May 2005, a 19-year-old male was admitted to our hospital suffering from exertional dyspnoea and a swollen abdomen. After a cold in January 2005 he started to lose weight and became progressively more dyspnoeic. He had not been exposed to asbestos.

On physical examination, he was afebrile, sweaty, and in some distress. He had a blood pressure of 135/80 mmHg, with no pulsus paradoxus. Furthermore, he had congested jugular veins, diminished basal pulmonary breathing sounds, hepatomegaly, ascites and ankle oedema. The electrocardiogram showed a

sinus rhythm, 90 beats/min, an intermediate axis and nonspecific repolarisation abnormalities.

The laboratory data revealed mild liver function abnormalities (serum aspartate aminotransferase 108 U/l, alanine aminotransferase 97 U/l, lactate dehydrogenase 724 U/l, bilirubin 23 µmol/l), and a moderately elevated plasma pro-brain natriuretic peptide (95.8 pmol/l).

Chest radiography demonstrated a widened cardiac shadow, bilateral pleural effusion and atelectasis of the left lung.

An echocardiogram showed a normal left ventricular systolic function and a grade 3 mitral incompetence; a large amount of pericardial effusion was seen, without signs of tamponade, and a large mass within the pericardium. Furthermore, a large mass on top of the left atrium, posterior to the ascending aorta, could be observed. In addition, several small tumours were located on the epicardial surface (figure 1).

A computed tomography and a magnetic resonance imaging scan (MRI) confirmed the pericardial and pleural effusions and clearly showed the epicardial, pericardial and mediastinal masses. A large mediastinal mass was present around the aorta and pulmonary artery (figure 2). These imaging studies, as well as the physical examinations, failed to reveal other masses in the trunk or extremities.

The patient underwent surgery to obtain biopsies or excise the tumours. He was haemodynamically unstable and single lung ventilation was insufficient due to external compression of the right main stem bronchus. Via a minor anterior lateral thoracotomy, 1100 ml of haemorrhagic pericardial fluid was evacuated and a large pericardial window was created. A massive glassy tumour (measuring 10 cm x 10 cm x 15 cm) with extensive areas of necrosis and haemorrhage was found on the diaphragm within the thickened pericardium. The restrictive epicardium was interlaced with multiple small glassy tumours that bled easily. A lymphoid mass was clogged to the aortic-pulmonary window. The tumours could not be resected, only biopsies could be taken.

The histopathology revealed a tumour composed of relatively uniform spindle-shaped tumour cells

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Figure 1. Echocardiogram: parasternal short-axis view showing pericardial effusion (PE), a pericardial mass (arrow head) and multiple epicardially located tumours (arrows).

Figure 2. MRI: transversal plane (balanced FFE sequence) showing pericardial effusion (PE), bilateral pleural effusion (PIE), atelectasis of the left lung (arrow head) and a tumorous mass encircling the aorta (Ao) and pulmonary artery (PA) (arrows).

Figure 3. Histopathological analysis showing uniform spindle-shaped tumour cells arranged in bundles, invading the microvasculature.

arranged in bundles (figure 3). Immunological analysis demonstrated diffuse expression of EMA, CD34, CD99, vimentin and focal expression of MNF116. In situ fluorescence hybridisation revealed a (X;18) translocation, specific for a synovial sarcoma. The morphology of the tumour cells, in combination with the immuno-histochemical profile of the tumour cells and the (X;18) translocation, led to the final diagnosis of a monophasic synovial sarcoma.

All treatment options were extensively considered and discussed. The patient and his parents eventually preferred a conservative and palliative approach. In August 2005, he died at home.

Autopsy revealed several tumours encircling the heart, the aorta and the pulmonary artery. The tumours extended to the left lung (leading to atelectasis) and the left thoracic wall, penetrating the right hemidiaphragm and reaching the hepatic surface. A solitary tumour was observed in the pericardium (figure 4). Additionally, thoracic and abdominal lymph node metastasis, thromboses in the innominate vein, and emboli in the right pulmonary artery were discovered, as well as pleural and abdominal fluid.

Primary cardiac tumours are rare (in a series of autopsies an incidence of less than 0.1% was found). In comparison, metastatic involvement of the heart is over 20 times more common. Patients may suffer from cardiac symptoms or the cardiac tumours may be found incidentally during evaluation for a seemingly unrelated problem.

Over 70% of primary cardiac tumours are histopathologically benign myxomas; the remainder include a variety of other tumours, of which the most common

Figure 4. Autopsy showing a tumour encircling the heart, extending to the left lung and left thoracic wall (arrow ←), penetrating the right-sided diaphragm, reaching the hepatic surface (arrow ↑) and a solitary tumour in the pericardium (arrow ↓).

are malignant cardiac tumours. Nearly all of these are sarcomas.

Primary cardiac or mediastinal synovial sarcomas are exceedingly rare.¹⁻³ Synovial sarcomas predominantly occur in para-articular soft tissues of the extremities of young adults and adolescents.⁴ Most cardiac synovial sarcomas are located in the right side of the heart. However, there are reports of a primary cardiac-related synovial sarcoma, originating either in the interatrial septum, the interventricular septum, or in the left heart or the pericardium.^{3,5} The synovial sarcomas in this case report were located in the mediastinum, in the

heart and in the pericardium. However, the origin remains unclear. Ectomesenchymal-derived soft tissue could be a common progenitor of these malignancies.

Synovial sarcoma is composed of two types of cells that can form a characteristic pattern: epithelial cells, resembling those of carcinoma, and fibro sarcoma-like spindle cells. Depending on the relative prominence of these two elements, synovial sarcoma can be classified into subtypes.

The availability of frozen tissue, procured at the time of surgery, allows for the molecular genetic identification of the t (X;18), which is found in synovial sarcoma. The translocation involves the SYT gene on chromosome 18 and either the SSX1 or SSX2 gene on the X chromosome.^{6,7}

Primary malignant cardiac tumours seldom show invasive or metastasis behaviour. These tumours proliferate rapidly and often cause death through widespread infiltration of the myocardium or obstruction of flow within the heart. Complete resection is seldom possible and if so, frequently followed by recurrent illness within two years.^{1,2} Due to the rarity of this malignancy, no standardised medical therapy exists (both older and newer regimes). Radiation therapy is known to be of value in cases where the margins are not tumour free, to reduce local recurrence rates. However, the history of primary cardiac synovial sarcoma, following surgical excision, adjuvant therapy or the natural progression of untreated patients, all yield a very poor survival. In this case a palliative approach was preferred. ■

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