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ESSENTIALS Cardiac myxoma Cardiac myxomas are rare benign tumours that grow in the lumen of the atria, usually the left. Most are sporadic, but they can be asso- ciated with the Carney complex, where unusual freckling is typically the most obvious clinical clue. Symptoms and signs most commonly mimic those of mitral sten- osis. Systemic emboli occur in about 40% of cases. Constitutional effects predominate in a few patients who present with what seems to be an obscure multisystem disorder. In many patients, specific car- diovascular signs are inconspicuous or absent: an audible ‘tumour plop’ in early diastole, analogous to a mitral opening snap, is often reported only after the diagnosis is established. The diagnosis is almost always made by echocardiography. Treatment is by urgent surgical removal. Recurrence is uncommon, provided excision has been complete, except in Carney complex. Other tumours of the heart The most common tumour seen in adult patients is the benign papillary fibroelastoma, which should be surgically removed only if it has been discovered in the search for a source of otherwise unexplained embolism. Primary cardiac sarcomas are found more often in the right heart than in the left. Surgical resection is often attempted for obstructive symptoms, but recurrence and metastasis are common, and long- term outcome is very poor. Microscopic secondary deposits within the myocardium can often be found in patients who die of metastatic cancer, but these are rarely of clinical importance. Intraluminal spread of cancer to the heart by direct extension up the inferior vena cava is a particular fea- ture of renal cell carcinoma. Cardiac myxoma Cardiac myxomas are benign, typically golf ball-sized, tumours that grow in the lumen of the atria, usually the left, attached by a stalk to the atrial septum. They are not common, but are important because they can present in several ways to general physicians, and because most can straightforwardly and permanently be removed by heart surgery. They are easily demonstrated by conventional transthoracic echocardiography, and it is usually the echocardiographer who makes the diagnosis; seldom has the patient been referred with this possibility in mind. Estimates of the prevalence of such a rare con- dition are necessarily approximate and range from 1 to 5 per 10 000 in autopsy series, or 2 per 100 000 in the general population, with a sex ratio of 2:1 in favour of women. As a cause of left atrial obstruc- tion, myxomas are 200–400 times less common than mitral stenosis. Most patients are between 30 and 60 years of age, but there are re- ports of tumours occurring in

infants and in older people. Most myxomas are sporadic, unassociated with other diseases, but there is at least one Mendelian syndrome involving myxoma, best named the Carney complex. This is caused by mutation in the protein kinase A regulatory subunit-1-α gene (Carney complex type 1) or mutations in other genes and characterized by lentiginosis, multiple myxomas (most of them cardiac), skin fibromas, and various kinds of endocrine overactivity, which has included Cushing's syndrome caused by pigmented adrenocortical hyperplasia, acromegaly, and Sertoli cell tumour. Unlike the usual kind of atrial myxoma, myx- omas in Carney's syndrome may arise anywhere in the heart, are commonly multiple, and frequently recur. Inheritance of this rare disease is autosomal dominant, with centrofacial freckling as the most obvious outward marker of the phenotype. This freckling often involves unusual areas, for instance, the lips, conjunctiva, and vulva. Pathology Cardiac myxomas are benign. Local invasion is unknown and meta- static growth is exceptional, despite the lesions' situation in the bloodstream. They take the form of polypoid masses arising from a stalk, ranging in size from 3 cm to as much as 10 cm or more, with a smooth or lobulated surface and gelatinous consistency. They are frequently covered with more or less adherent thrombus. More than 75% occur within the left atrium, with the base of the pedicle arising from the fossa ovalis or its rim. Occasionally, they arise from the base of the mitral valve leaflets, from the posterior part of the left atrium, or from within the right atrium. Sometimes they grow in both atria, in the form of a dumbbell. Ventricular myxomas are exceptional and seen almost exclusively as part of Carney's syndrome. Left atrial 16.10 Tumours of the heart Thomas A. Traill

 myxomas are not generally as large as those in the right atrium at the time they are first detected. The latter may almost fill the right atrium before they begin to obstruct systemic venous return. The histology is that of a loosely woven, sparsely cellular, con- nective tissue tumour with very infrequent mitotic figures. Several cell types are identifiable, including undifferentiated stellate and pol- ygonal cells, as well as smaller numbers of fibroblasts, smooth muscle cells, and endothelial cells. Among these are found macrophages and plasma cells, and rarely other mesodermal tissues, including bone. Cytogenetic studies fit with the general presumption that these in- dolent masses are indeed neoplastic, but immunohistochemical studies of differentiation markers do not clearly define the cell type of origin. It is suggested that the source is a primitive multipotential mesenchymal cell and that the predilection of these tumours for the atrial septum reflects the abundance of such cells in this region. Clinical features Left atrial obstruction The most common symptoms and signs mimic those of mitral stenosis, with left ventricular inflow obstruction as the chief patho- physiological change. The presenting symptoms are progressive breathlessness, orthopnoea, paroxysmal nocturnal dyspnoea, fluid retention, and atrial arrhythmias. Examination suggests rheumatic heart disease, and before the routine use of ultrasonography a few such patients were referred for mitral valve surgery and the lesion was first diagnosed at operation. Some patients may develop pul- monary hypertension before the diagnosis becomes apparent. Systemic embolism Systemic emboli occur in about 40% of patients and are frequently the first manifestation of disease. In contrast to mitral stenosis, such emboli often occur while patients are in sinus rhythm. Emboli may be sizeable, large enough even to occlude the aortic bifurca- tion, and, besides thrombus, they frequently contain tumour ma- terial, hence histological examination may be diagnostic. When systemic emboli are removed from patients, they should always be sent for histological analysis. Typically, patients with systemic em- bolism are referred for echocardiography, and the diagnosis is then easily made. Constitutional effects Constitutional effects of the neoplasm predominate in a few patients who present with what seems

to be an obscure multisystem disorder. Symptoms and signs include fever, weight loss (which is more con- spicuous than in mitral stenosis and often occurs without severe left atrial obstruction), Raynaud's phenomenon (rare), finger clubbing (rare), a raised erythrocyte sedimentation rate (present in about 60% of patients), and abnormal serum proteins with elevated immuno- globulin levels. These changes are usually attributed to abnormal proteins secreted by the tumour, although the nature of these has not been determined. Other haematological abnormalities include an- aemia, which may be due to mechanical haemolysis, polycythaemia, associated particularly with right atrial tumours, leucocytosis, and thrombocytopenia. Such constitutional changes may prompt an ini- tial diagnosis of infective endocarditis in patients who have heart murmurs, or lead to the suspicion of autoimmune rheumatic or vas- culitic disease, or of occult cancer. Physical signs In many patients, specific cardiovascular signs of myxoma are in- conspicuous or absent. In others, they vary from a prominent first heart sound to obvious changes similar to those of mitral valve disease. These include apical systolic murmurs, somewhat more common than diastolic rumbles, and—in some patients—signs of pulmonary hypertension, with accentuated pulmonary closure and tricuspid regurgitation. Some may have an audible 'tumour plop' in early diastole, analogous to a mitral opening snap, but this is often reported only after echocardiographic diagnosis. On combined echocardiographic and phonocardiographic recordings, the plop is seen to coincide with the end of the tumour's downward movement into the ventricle, usually a short time after mitral valve opening. A rare but specific feature of the condition is variation of the aus- cultatory findings with change in posture; this may be particularly obvious in right atrial tumours. Investigations Chest radiography and electrocardiography do not help to distin- guish myxoma from mitral valve disease. Left atrial enlargement is common but seldom marked, and signs of pulmonary venous hypertension are infrequent. Calcification within the tumour is rarely demonstrable. Myxomas may be identified as filling defects on CT examinations of the chest. Echocardiography While the first account of left atrial myxoma diagnosed during life was not until 1951, it is now exceptional for the diagnosis to be made first at autopsy. This is chiefly attributable to the wide avail- ability of echocardiography, which has proved itself both reliable and specific for recognizing these tumours. It is no accident that the echocardiographic appearance of these lesions was among the first clinical reports by ultrasonographers in 1959. Fig. 16.10.1 illustrates a typical two-dimensional transthoracic echocardio- gram from a patient with left atrial myxoma, and Fig. 16.10.2 images obtained from a transoesophageal approach in a different patient. A video recording would demonstrate the mobility of the mass as it flops to and fro within the atrium, restrained only by its peduncle. Transoesophageal echocardiography affords the opportunity to examine the tumour and its attachment with great precision; generally, this extra clarity is unnecessary, but on occasion the transoesophageal technique is helpful if there is difficulty in differentiating tumour from an atrial thrombus. The differential diagnosis of left atrial myxoma is seldom diffi- cult. Large masses may occasionally be difficult to distinguish from left atrial ball thrombus—a lesion that is even rarer than myxoma. Smaller left atrial masses may be papillary fibroelastomas or in- fective vegetations caused by endocarditis. These can usually be dis- tinguished by their clinical context. Masses in the right atrium may also be due to thrombus, sometimes propagated from the inferior vena cava, or occasionally venous extension of abdominal cancers, particularly renal cell cancer. In a few patients, abundant strands of the Chiari network of right atrial trabeculation may give rise to similar echocardiographic appearances. Myxoma is the only neo- plasm of the heart to be found within its lumen: other cardiac neo- plasms grow within the walls.

section 16 Cardiovascular disorders 3546 Other imaging Myxomas can also be identified by MR imaging (Fig. 16.10.3). Angiography has no role in diagnosis of myxoma, although it may be required as a prelude to surgery in the older patient in whom there is, or might be, coronary artery disease. Treatment and prognosis Atrial myxoma is treated by urgent surgical removal (Fig. 16.10.4). The risk is low, comparable to that of surgery for mitral valve disease. It is important to ensure complete re- moval of the base by excising a full-thickness button of the (a) (b) (c) (d) (e) (f) Fig. 16.10.1 A large left atrial myxoma (T) recorded from the parasternal long-axis (a, b) and from the apical four-chamber views (c, d) in systole (a, c) and diastole (b, d). During diastole, the tumour prolapses into the left ventricle (LV), completely obstructing the mitral valve orifice (b, d). (e) M-mode recording at the level of the mitral valve of a prolapsing left atrial myxoma. A dense array of wavy tumour echoes is seen behind the anterior leaflet of the mitral valve. (f) Continuous wave Doppler recording of tricuspid regurgitation velocity of the same patient. Peak velocity is 5 m/s, corresponding to a 100 mm Hg transtricuspid gradient. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. From Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition,

16.10 Tumours of the heart 3547 Fig. 16.10.2 A longitudinal transoesophageal view of a large left atrial myxoma (T). In systole (left) it remains inside the left atrium, while in diastole (right) it enters the mitral valve orifice, virtually occluding the orifice. Ao, aorta; LA, left atrium; LV, left ventricle. From Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition,
 Fig. 16.10.3 A large left atrial myxoma is attached to the interatrial septum (*). (Top) Steady-state free precession (SSFP) cine sequence in the HLA (left) and left ventricular outflow tract (LVOT; right) views. The myxoma prolapses through the mitral valve in diastole, causing significant obstruction to flow and mimicking mitral stenosis. (Bottom) T1-weighted turbo spin echo (TSE) images with fat saturation, highlighting the tumour against the low signal of the moving blood. From Myerson SG, Francis J, Neubauer S (eds) (2013). Cardiovascular magnetic resonance (Oxford specialist handbooks in cardiology), by permission of Oxford University Press.

section 16 Cardiovascular disorders 3548 atrial septum, the resulting defect being repaired with a small patch. Functional results of surgery are good. Some patients are left with mitral regurgitation, but this is seldom severe. Recurrence is un- common, provided excision has been complete, except in Carney’s syndrome. In these patients, regular echocardiographic follow-up is required, at intervals of 6 months. The rare occurrence, after ex- cision, of the usual kind of myxoma generally occurs within the first 2 years; thereafter, follow-up can safely be infrequent. Other tumours of the heart Although each individually is rare, taken together the other tu- mours of the heart have an incidence that roughly equals that of myxoma. They include benign lesions, seen especially in children; sarcomas; and secondary involvement by metastasis or direct tu- mour extension. They are generally first recognized or suspected during echocardiography. MRI, or occasionally echo-directed transvenous biopsy, usually yields the diagnosis. Benign cardiac tumours Papillary fibroelastoma The most common tumour seen in adult patients is the papillary fibroelastoma, a small pedunculated mass that hangs off one of the left-sided valve leaflets, usually the mitral valve. Its echocardiographic appearance is very characteristic. The size of the mass and presence of a peduncle distinguish this small tumour from the usual kind of Lambl’s excrescence, but

histologically they are identical and, like Lambl's excrescences, papillary fibroelastomas probably arise through organization of fibrinous material that collects at the trailing edges of the valve leaflets. Their importance lies in the fact that they have been labelled as a potential source of systemic embolism, and that some authors have recommended they should be removed as a matter of routine. The evidence to support this view is thin, and the author's recommendation is to remove them only if they have been discovered in the search for a source of otherwise unexplained em- bolism. If they are an incidental echocardiographic finding, then it is safe to leave them alone; aspirin treatment is recommended. Fibroma, rhabdomyoma, hamartoma, and haemangioma These are tumours of childhood, rhabdomyoma being the character- istic cardiac tumour in patients with tuberous sclerosis. In contrast to myxomas and fibroelastomas, they grow within the myocardium, not into the lumen of the heart. Rhabdomyomas are usually asymp- tomatic, and when they are they should be left alone, since most re- gress spontaneously. Fibromas and hamartomas are both very rare, presenting with arrhythmias (particularly ventricular hamartomas or Purkinje cell tumours) or with haemodynamic abnormalities caused by their mass effect (Fig. 16.10.5). They require surgical excision, and when this is feasible the long-term results of treatment are very good. Haemangiomas, also very rare, tend to grow and to develop multiple feeding vessels, so surgical excision is usually recommended. Cardiac sarcoma Primary cardiac sarcomas are found more often in the right heart than in the left (Fig. 16.10.6), and can have one of several cell types. Fig. 16.10.4 Left atrial myxomas that were removed surgically. Note the difference in appearance: the top myxoma has a smooth surface while the bottom one is villous, more friable, and prone to tissue fragmentation and embolism. Courtesy of Rute Couto MD, and Rui Rodrigues MD, from Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition, © European Society of Cardiology, by permission of Oxford University Press. Fig. 16.10.5 Apical four-chamber view from a young child showing a voluminous fibroma (arrow) located in the interventricular septum, causing left ventricular outflow tract obstruction. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. Courtesy of Rui Anjos, MD, from Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition, © European Society of Cardiology, by permission of Oxford University Press.

16.10 Tumours of the heart 3549 Hemangiosarcoma is the most common, typically developing in the right atrium. Rhabdomyosarcoma may develop in the ventricular septum or in the right ventricular outflow tract, as may the still rarer osteosarcoma, or tumours that are undifferentiated. Since these tu- mours often present with mechanical effects, typically obstruction at the atrial or outflow tract level, surgical resection is often attempted. However, recurrence and metastasis are common, and long-term outcome is very poor. Cardiac involvement by other malignancies Microscopic secondary deposits within the myocardium can often be found in patients who die of metastatic cancer, but intramyocardial secondaries of a size large enough to be of clinical importance are very rare (Fig. 16.10.7). By contrast, pericardial involvement by lymphoma, or by cancers of the lung, breast, pancreas, and other tumours is not un- common, and may sometimes be the first presentation of the tumour (see Chapter 16.8). Treatment is analogous to that of malignant pleural effusions, with drainage, creation of a window, or intrapericardial chemotherapy, depending on the rest of the clinical situation. Intraluminal spread of cancer, by direct extension up the inferior vena cava, is a particular feature of renal cell carcinoma. Diagnosis by echocardiography is generally obvious, as the tumour has a very characteristic appearance as it waves like seaweed in the right atrium and even dangles through the rest of the right heart. It may

prove possible to resect the cava, along with the kidney and the tumour mass, under circulatory arrest. Fig. 16.10.6 Apical four-chamber (left) and two-chamber (right) views of a left atrial mass (arrows) that corresponded to a primary sarcoma removed completely with surgery. LV, left ventricle; RA, right atrium; RV, right ventricle. From Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition,
 Fig. 16.10.7 A bulky right-sided metastasis (T) causing right ventricular outflow obstruction in a patient with a primary germ cell tumour, which presented with syncope. A small pericardial effusion is also present. Ao, aorta; LA, left atrium; LV, left ventricle; RA, right atrium. From Lancellotti P, Zamorano J, Habib G, Badano L (eds) (2017). The EACVI textbook of echocardiography, 2nd edition,

section 16 Cardiovascular disorders 3550 FURTHER READING Cardiac myxoma Greenwood WF (1968). Profile of atrial myxoma. Am J Cardiol, 21, 367–75. Lee KS, et al. (2017). Surgical resection of cardiac myxoma—a 30-year single institutional experience. J Cardiothorac Surg, 12(1), 18. Pucci A, et al. (2000). Histopathologic and clinical characterization of cardiac myxoma: review of 53 cases from a single institution. Am Heart J, 140, 134–8. Stratakis CA, KIrschner LS, Carney JA (2001). Clinical and mo- lecular features of the Carney complex: diagnostic criteria and recommendations for patient evaluation. J Clin Endocrinol Metab, 86, 4041–6. Wilkes D, McDermott DA, Basson CT (2005). Clinical phenotypes and molecular genetic mechanisms of Carney complex. Lancet Oncol, 6, 501–8. Other tumours of the heart Burke A, Jeudy J Jr, Virmani R (2008). Cardiac tumours: an update. Heart, 94, 117–23. Chen TW, et al. (2019). Primary cardiac sarcomas: a multi-national retrospective review. Cancer Med, 8, 104–10. Lee E, et al. (2018). Primary cardiac tumors associated with genetic syndromes: a comprehensive review. Pediatr Radiol, 48, 156–64.

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