CASE REPORT

A fatal case of primary cardiac chondrosarcoma presenting with amaurosis fugax

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SUMMARY

A 64-year-old previously healthy woman consulted her general practitioner because of recurrent episodes of right-sided monocular transient visual loss (ie, amaurosis fugax). At first, these symptoms were followed over time, but as the attacks worsened, and were accompanied by dizziness and general discomfort, the patient was admitted to the department of neurology for further investigations. CT of the brain was normal; however, during admission, the patient developed rapid atrial fibrillation and was transferred to the department of cardiology. Transthoracic echocardiography revealed a massive tumour on the atrial side of the anterior mitral valve leaflet, partly obstructing the mitral valve inflow. The tumour was excised and a biological prosthetic mitral valve inserted. The tumour was histologically determined to be a highly malignant dedifferentiated chondrosarcoma. After 6 months, the tumour relapsed and expanded aggressively to completely obstruct the mitral valve inflow, ultimately leading to cardiac arrest and death.

BACKGROUND

Primary cardiac chondrosarcoma (PCC) is a malignant subtype of sarcoma and is presumed to arise from multipotent mesenchymal stem cells that undergo malignant cartilaginous differentiation. The most common initial symptom reported is dyspnoea on exertion as a result of congestive heart failure, intracavitary obstruction, or pericardial effusion. The presenting symptom in this case was, however, amaurosis fugax, which is defined as a painless transient monocular visual loss due to retinal artery ischaemia.¹ The annual incidence of first-time amaurosis fugax is estimated at around 7/100 000.² Approximately two-thirds of cases are idiopathic while the majority of the remainder are caused by embolic, haemodynamic, ocular or neurological diseases.³ Patients typically report amaurosis fugax as being a 'down or upcoming curtain', appearing into the visual field.³ We report the first case of PCC presenting with recurrent episodes of amaurosis fugax.

CASE PRESENTATION

A 64-year-old woman presented with six episodes of classical amaurosis fugax over the course of 2 months. The first episode presented with abrupt onset of complete anopsia of the right eye persisting for approximately 5 min followed by complete visual recovery over an additional few minutes. All subsequent episodes only involved the lower part of the visual field of the right eye, and were

accompanied by dizziness and general discomfort. The patient had also noted progressive dyspnoea on exertion, but initially attributed this symptom to stress at work and the recent loss of a close relative. No sensory disturbance, pareses or other neurological symptoms were present.

The patient had no predisposition to cancer, but had a positive family history of coronary artery disease.

INVESTIGATIONS

Initial clinical examination was normal apart from a faint diastolic murmur on cardiac auscultation. Routine biochemical investigations including troponin T were within the laboratory reference limits. Owing to the initial symptom of recurrent amaurosis fugax, the patient underwent a cerebral CT scan, which ruled out ischaemic or haemorrhagic stroke, as initially suspected. As the patient developed irregular tachycardia with a heart rate of 200 bpm, an ECG was recorded, confirming atrial fibrillation. The combination of several episodes of amaurosis fugax and atrial fibrillation finally led to a transthoracic echocardiography, revealing a massive tumour attached to the atrial side of the anterior mitral valve leaflet. This was followed by transoesophageal echocardiography (figure 1) for more accurate assessment of tumour size and effect on mitral valve function. As seen in figure 1, the tumour prolapsed across the mitral valve, threatening to release emboli to the arterial system and cause amaurosis fugax and other ischaemic phenomena. Histological sections of the tumour determined it to be a highly malignant dedifferentiated chondrosarcoma (figure 2).

TREATMENT

The initial treatment of rapid atrial fibrillation included rate control with a β -blocker (metoprolol) and anticoagulation with a vitamin K antagonist (warfarin). Shortly after diagnosing the tumour, it was decided to perform a surgical excision and, as the tumour was attached to the anterior mitral valve leaflet, a biological prosthetic mitral valve was inserted. Rhythm control with amiodarone was initiated following surgery.

OUTCOME AND FOLLOW-UP

The surgery was performed successfully and without perioperative complications. However, after approximately 6 months, the tumour relapsed. Chemotherapy was initiated but had no substantial effect on the aggressive growth of the tumour, which eventually caused obstruction of the mitral

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Figure 1 Transoesophageal two-chamber views at the level of the mitral valve, illustrating a 22×44 mm obstructing tumour attached to the atrial side of the anterior mitral valve leaflet. (A) Two-dimensional (2D) image of the left ventricle during diastole. (B) The 2D image of the left ventricle during systole. (C) The 2D colour Doppler image with intense inflow jet, which aliases across the mitral valve (peak/mean gradient 46/28 mm Hg). (D) The 2D colour Doppler image of the left ventricle during systole with mitral valve regurgitation (LV, left ventricle; LA, left atrium; LAA, left atrial appendage).

valve inflow and cardiac arrest. Owing to the underlying malignant disease, resuscitation was not attempted.

DISCUSSION

We report a fatal case of PCC in the left atrium, with amaurosis fugax as the predominant initial symptom. Sarcomas comprise the largest group of malignant primary cardiac neoplasms, with angiosarcomas being the most common subtype. In contrast, PCCs are exceedingly rare, especially in the left heart, with only nine cases reported in the English literature since the initial report by McConnell in 1970.⁴ Although right heart PCCs are more common, they are also more likely to represent metastatic lesions than primary cardiac tumours.^{5 6}

Of the previous nine cases of PCCs in the left heart, eight originated in the left atrium^{6–13} and only one in the left ventricle.¹⁴ The initial symptom has most often been

dyspnoea,^{6 7 9–13} while symptoms related to production of parathyroid hormone-related protein have also been reported.⁸

Transthoracic echocardiography remains the modality of choice for the initial evaluation of cardiac tumours and masses.¹⁴ However, a CT scan can more accurately image the heart as well as the surrounding mediastinum. Furthermore, it provides better soft tissue contrast than echocardiography, and can also depict calcification and haemorrhagic areas within tumours.¹¹ MRI better defines the extension of tumours into the interatrial septum and pulmonary veins, as well as into the pericardium.¹¹ In the present case, transthoracic echocardiography was used initially, followed by transoesophageal echocardiography for better representation of the left atrium and more accurate preoperative assessment of the tumour due to its superior spatial resolution.

Figure 2 Histological sections of the tumour stained with H&E. (A) Low-power magnification of biphasic tumour showing a dedifferentiated spindle cell component along with a well-differentiated chondroid component. (B) High-power magnification of the chondroid component with hyaline matrix and lacunae with chondrocytes. (C) High-power magnification of a spindle cell component with nuclear pleomorphism and mitoses. (D) High-power magnification of transition with a hint of ossification and poorly differentiated primitive cells.

In the initial diagnostic work up of a patient presenting with amaurosis fugax, focus should be directed at ruling out the most critical causes, namely, cardiac arrhythmia or structural heart disease, embolic phenomena leading to stroke, carotid artery disease and temporal arteritis. Carotid artery disease is prevalent in the elderly population¹⁵ and is the underlying cause of amaurosis fugax in 27% of all cases.¹⁶ However, carotid duplex scanning (whether positive or negative) does not always rule out other causes of amaurosis fugax. When encountering a patient with amaurosis fugax accompanied by syncope or dyspnoea on exertion, it is therefore critical that cardiac causes are excluded.

Regarding the management of PCC, surgery remains the first-line treatment; however, long-term survival after surgical excision is poor due to infiltrative growth with following early local recurrence and metastases.¹¹ Surgery is recommended not only to obtain a tissue diagnosis but also to relieve tumour compression with subsequent prolonged survival and improved quality of life.¹⁰

Adjuvant chemotherapy is often initiated on relapse or used as primary treatment when tumours are not suitable for radical surgery. The efficacy of chemotherapy has not been established.

The prognosis of PCC is extremely poor and limited to weeks or months.^{6 7 11} Surgical and chemotherapeutic strategies have both generally been disappointing for most patients. However, Hsing *et al*⁹ reported a case of PCC in which the combination of incomplete resection and adjuvant combination chemotherapy with etoposide, ifosfamide and cisplatin showed a relatively good response. After six cycles, the size of the mass in the left atrium was reduced without distant metastases, and the symptoms were relieved following initiation of treatment. Although these malignant tumours usually metastasise widely, systemic metastases were only present at initial presentation in one of the previous nine cases of PCC in the left heart.⁷

To the best of our knowledge, amaurosis fugax as the presenting symptom of PCCs in the left heart has never previously been reported. Thus, this symptom is rarely a sign of PCC but often heralds more common cardiac disease such as atrial fibrillation with thromboembolism. As the symptom is painless and transient in nature, patients tend to consult their general practitioner after resolution of symptoms. Reports of amaurosis fugax-like episodes from patients merit clinical attention, and

thorough investigation of the aetiology should be sought, including possible cardiovascular causes.

Learning points

- ▶ Primary cardiac chondrosarcoma (PCC) is an extremely rare cardiac tumour with infiltrative growth and a dismal prognosis.
- ▶ Amaurosis fugax is a painless transient monocular visual loss due to retinal artery ischaemia.
- ▶ Despite the temporary nature of the vision loss, those experiencing amaurosis fugax are advised to consult a physician immediately, as the symptom often heralds serious vascular events (eg, stroke, critical cardiac disease and temporal arteritis).
- ▶ Amaurosis fugax should be recognised as a critical symptom necessitating immediate diagnostic work up including: complete physical examination, ECG, carotid ultrasonography and MRI or CT scan.
- ▶ Patients presenting with amaurosis fugax should be referred for transthoracic echocardiography if cardiac disease is suspected during initial diagnostic work up.

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REFERENCES

- 1 Bioussé V, Trobe JD. Transient monocular visual loss. *Am J Ophthalmol* 2005;140:717–21.
- 2 Andersen CU, Marquardsen J, Mikkelsen B, *et al*. Amaurosis fugax in a Danish community: a prospective study. *Stroke* 1988;19:196–9.
- 3 [No authors listed]. Current management of amaurosis fugax. The Amaurosis Fugax Study Group. *Stroke* 1990;21:201–8.
- 4 McConnell TH. Bony and cartilaginous tumors of the heart and great vessels. Report of an osteosarcoma of the pulmonary artery. *Cancer* 1970;25:611–17.
- 5 Ng SH, Ko SF, Yang TS, *et al*. Primary cardiac chondrosarcoma. *Am J Emerg Med* 1996;14:285–7.
- 6 Nesi G, Pedemonte E, Gori F. Extraskelatal mesenchymal chondrosarcoma involving the heart: report of a case. *Ital Heart J* 2000;1:435–7.
- 7 Miwa S, Konishi Y, Matsumoto M, *et al*. Primary cardiac chondrosarcoma—a case report. *Jpn Circ J* 1997;61:795–7.
- 8 Kase S, Nakamoto S, Miyasaka S, *et al*. Cardiac chondrosarcoma producing parathyroid hormone-related protein. *Circ J* 2004;68:715–18.
- 9 Hsing CT, Oh SY, Lee S, *et al*. Extraskelatal mesenchymal chondrosarcoma of the heart responded to systemic chemotherapy: a case report. *Cancer Res Treat* 2007;39:131–3.
- 10 Izzo P, Ricci N, Capolupo R, *et al*. A rare case of primary chondrosarcoma of the heart. *J Cardiovasc Med (Hagerstown)* 2007;8:210–13.
- 11 Parmar C, Jojo A, Vachhani KC, *et al*. Primary chondrosarcoma of the heart. *Eur J Cardiothorac Surg* 2008;33:513–15.
- 12 Sawaed S, Adler AC, Vlodavsky E, *et al*. Primary cardiac chondrosarcoma with rapid MFH-like recurrence: case report and review of tumor type. *Tumori* 2010;96:637–9.
- 13 Zhang G, Chen X, Guo L, *et al*. Primary Cardiac Chondrosarcoma. *J Card Surg* 2012;27:186–8.
- 14 Jinno T, Morimoto T, Itoh A, *et al*. Primary cardiac chondrosarcoma with large cell pulmonary carcinoma. *Jpn J Thorac Cardiovasc Surg* 2006;54:228–31.
- 15 de Weerd M, Greving JP, Hedblad B, *et al*. Prevalence of asymptomatic carotid artery stenosis in the general population: an individual participant data meta-analysis. *Stroke* 2010;41:1294–7.
- 16 Smit RL, Baarsma GS, Koudstaal PJ. The source of embolism in amaurosis fugax and retinal artery occlusion. *Int Ophthalmol* 1994;18:83–6.

Patient's perspective

My wife's course of disease started with malaise accompanied by repeated episodes of transient visual loss. She consulted her general practitioner several times before she was admitted to the department of neurology. CT of her brain did not explain her symptoms, and the underlying cause was not suspected until a doctor recorded an ECG. The ECG showed an irregular heart rhythm and this led to an ultrasound scan of her heart, revealing the tumour. The time from initial symptoms to diagnosis was long and frustrating, and could have been shortened if relevant investigations were initiated when we first got in touch with the general practitioner. We witnessed a healthcare system that neglected my wife's initial symptom of visual loss as her vision had recovered every time we consulted the general practitioner. My hope is that my wife's story can disseminate the knowledge of this rare disease and enable doctors to react appropriately to symptoms such as transient visual loss.

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