CASE REPORT

Intimal (spindle cell) sarcoma of the left atrium presenting with abnormal neurological examination

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SUMMARY

A 43-year-old man with an unremarkable medical history presented to our hospital with 2 weeks of headaches, ataxia and confusion. CT of the head revealed a large haemorrhagic cystic lesion. A subsequent chest CT revealed a large left atrial mass. The mass was subsequently biopsied with positive immunohistochemistry staining for MDM2, FLI1 and vimentin. Real-time PCR revealed MDM2 amplification, confirming the diagnosis of intimal sarcoma. The patient underwent surgical resection and reconstruction of the atrium with subsequent discharge to short-term rehabilitation, but his symptoms continued to progress. A repeat CT of the head revealed a new cerebellar mass. He underwent a second resection, but continued to experience worsening symptoms. He was diagnosed with stage IV intimal sarcoma and referred to hospice. The patient died 5 months after initial presentation. Autopsy was performed and revealed the cause of death as pneumonia. There was no involvement of the pulmonic or aortic vessels.

BACKGROUND

There have been only five previous cases of cardiac intimal sarcoma reported in the literature, with each presenting in a unique way. These tumours more commonly present in the large blood vessels. Their course is highly aggressive, with a median survival of 3 months to 1 year.¹ Our case describes the sixth such documented case of left atrial intimal sarcoma and, to the best of our knowledge, is the first such case presenting with neurological signs and symptoms.

CASE PRESENTATION

A 43-year-old man presented with a 2-week history of headaches, ataxia and confusion, as noted by his fiancée. He had no previous medical history prior to his presentation at the hospital. His review of systems was otherwise negative, and physical examination revealed only nystagmus on upward gaze and an ataxic gait. Motor reflexes were symmetric and intact bilaterally.

INVESTIGATIONS

Initial CT of the head revealed a 4.0×3.6×3.4 cm haemorrhagic cystic lesion in the right periventricular area, with lateral ventricle extension (figure 1). The characteristic of the lesion was suspicious for metastasis from a primary malignancy. A chest CT was completed to evaluate for a primary malignancy, and revealed a 6.5×4.5 cm left atrial mass in proximity to the left superior and inferior pulmonary veins (figure 2).

A left atrial mass was excised to characterise the histology. Pathology revealed malignant epithelioid and spindle cell neoplasm with myocardial and endocardial involvement. Immunohistochemical studies indicated focal positivity for alpha-smooth muscle actin (SMA), but were negative for CD34, CD31, Fli-1, ERG, desmin, S100 and cytokeratin AE1/AE3. Fluorescence in situ hybridisation revealed 85% MDM2 amplification, with karyotype revealing 12q13–15, proving the diagnosis for intimal sarcoma.

Cerebellar pathology was remarkable for a moderate to densely cellular neoplasm with cortical infiltration of poorly differentiated large epithelioid tumour cells with moderate eosinophilic cytoplasm, and pleomorphic nuclei with prominent nucleoli and irregular nuclear borders. The immunohistochemical profile was focally positive for SMA, and positive for MDM2 expression, FLI1 and vimentin. It was negative for HMB-45, Cam 5.2, CD117, alpha-fetoprotein (AFP) and placental alkaline phosphatase (PLAP). This was characterised as metastatic spread from the primary intimal sarcoma.

DIFFERENTIAL DIAGNOSIS

Spindle cells on histology can present in many different sarcomas apart from intimal sarcoma. Synovial sarcoma is a common histological equivalent, presenting with spindle cells in either monophasic or biphasic forms. Monophasic synovial sarcoma is histologically similar to fibrosarcomas, while biphasic synovial sarcoma presents similarly to intimal sarcoma, as an admixture of both epithelioid and spindle cells.² Apart from their near identical histological presentation, their overall rare occurrence and non-specific clinical presentation make discrimination among these different conditions a diagnostic challenge. The gold standard for confirmatory diagnosis is molecular characterisation. Synovial sarcoma typically will have a low-grade histological appearance, cellular pleomorphism and minimal atypical cytological features. The presence of the t(x;18)(p11;q11) chromosomal translocation is a specific finding found only in synovial sarcoma, but there have been cases where this translocation has been absent.³ While MDM2 overexpression is observed in all intimal sarcomas, it is not enough to distinguish it from undifferentiated sarcomas or angiosarcomas. MDM2 overexpression, in combination with molecular aberration, is only present in intimal sarcoma.⁴

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Figure 1 Initial head CT scan on presentation, revealing the 4.0×3.6×3.4 cm haemorrhagic cystic lesion in the right periventricular area.

TREATMENT

The patient underwent surgical resection and reconstruction of his atrium. His postoperative course was complicated by bleeding in the posterior aspect of the left atrium, pulmonary embolism and sternal wound dehiscence. An inferior vena cava filter was placed and the patient was discharged to a short-term rehabilitation facility.

OUTCOME AND FOLLOW-UP

The patient's headaches, confusion and ataxia continued to worsen while in short-term rehabilitation. A repeat CT of the head revealed a new large right cerebellar mass with irregular haemorrhage. He underwent a right craniotomy and resection and the lesion was biopsied. Biopsy confirmed metastatic spread from the primary intimal sarcoma. However, he continued to experience new symptoms of abdominal pain, nausea, vomiting and 15 lb weight loss, with occasional dyspnoea on exertion after walking several blocks. Repeat imaging revealed new metastasis to the liver and lung. Echocardiography revealed an ejection fraction of 60.4% and evidence of a mass of

Figure 2 Initial contrast chest CT scan, revealing a 6.5×4.5 cm mass in the left atrium.

3.1×3.5 cm in the left atrium. This was deemed to be surgically unresectable and the patient was referred to hospice.

The patient had difficulty accepting the finality of his condition and elected to be discharged from hospice. He was admitted for severe sepsis due to presumed pneumonia, after which he soon expired. An autopsy was performed, revealing the cause of death was metastatic cardiac intimal sarcoma complicated by bilateral bronchopneumonia. There was no evidence of sarcoma involvement of the great vessels.

DISCUSSION

Primary cardiac intimal sarcoma is a very rare tumour, with poor prognosis. Diagnosis can be difficult to confirm, should these tumours occur in atypical locations, due to their rare occurrence and their virtual identical histological presentation to other conditions. The role of molecular cytogenetics aids us in the diagnosis and in potentially defining future therapeutic targets.⁴ MDM2 amplification, compared to overexpression, is more specific for the diagnosis of spindle cell sarcoma and is essentially confirmed in the presence of poorly differentiated sarcomas.⁴ Typically, intimal sarcoma is usually described in large vessels.⁵ Our case is the first to initially present with a neurological presentation.⁶ Each of the previous cases presented in a unique way, but none presented with an initial abnormal neurological examination. The first case described in the literature was reported in a pregnant woman at 27 weeks gestation, who died approximately 10 months after the onset of symptoms.⁷ While the median survival is at 3 months, one group described the case of a 48-year-old man who survived 11 years after initial diagnosis.⁸ Ibrahim *et al*¹ reported a case of a 69-year-old man presenting with abdominal discomfort in the setting of recent significant weight loss. Recently, a Canadian group described a case of left atrial intimal sarcoma presenting with signs of congestive heart failure.⁶ While surgical resection is essential for treatment, combination with radiotherapy or chemotherapy may be beneficial in non-metastatic patients.^{9 10}

Learning points

- ▶ Primary cardiac intimal sarcoma is a very rare tumour with an overall poor prognosis.
- ▶ Intimal sarcoma may initially present at advanced stages without symptoms of heart failure.
- ▶ The presence of spindle cells on histology is a non-specific finding, and requires molecular cytogenetics in order to confirm the diagnosis.
- ▶ While MDM2 overexpression is found in spindle cell sarcomas, it may also be found in other tumours as well, and requires both amplification and molecular abnormalities in others to confirm the diagnosis.
- ▶ Mean survival of these reported cases is between 3 months and 1 year, with the exception of one case described with survival up to 11 years, postdiagnosis.

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