Case Report

Asymptomatic Left Ventricular Malignant Psammomatous Melanotic Schwannoma

Charles Laurin, MD, Joel Claveau, MD, Sylvain Trahan, MD, Louis-Philippe Gagnon, MD, Dimitri Kalavrouziotis, MD, and Jean Perron, MD

Cardiac Surgery Department, Quebec Heart and Lung Institute, Laval University, Quebec City, Quebec, Canada
Pathology Department, Quebec Heart and Lung Institute, Laval University, Quebec City, Quebec, Canada
Department of Internal Medicine, Dermatology Division, University Hospital of Quebec, Laval University, Hôtel-Dieu Hospital, Quebec City, Quebec, Canada

ABSTRACT

Malignant psammomatous melanotic schwannoma (MPMS) is a rare type of tumour, occasionally reported to occur with mediastinal involvement. Histopathologic similarities with melanoma may lead to a wrong diagnosis, but distinguishing between types of tumours is mandatory for adequate management and prognosis. MPMS may be aggressive and manifest unpredictable behavior, with a poor midterm prognosis despite benign histopathologic features. We discuss the challenges that come with a diagnosis of MPMS, and the rationale for our treatment strategy, in this first report regarding MPMS involving the left ventricle.

Malignant psammomatous melanotic schwannoma (MPMS) represents 10% of all diagnosed schwannomas, with approximately 200 published cases. Rarely, MPMS has been described as occurring in the mediastinum, with one report outlining a case of MPMS in the right atrium. MPMS can be difficult to distinguish from melanoma because of histologic similarities. The management and prognosis differ greatly between these 2 tumours, thus necessitating an accurate final diagnosis. To our knowledge, this is the first report of MPMS involving the left ventricle (LV) of the heart. We aim to discuss the steps toward accurate diagnosis, treatment strategy, and long-term follow-up of left ventricular MPMS.

Pathology

Originating from the neural crest, the histopathologic features of malignant melanotic schwannoma are extremely variable. Malignant melanotic schwannoma is usually a grey-to-black tumour that exhibits psammoma bodies, a variable amount of melanin pigment, and similar morphology (spindled or epithelioid) to melanocytes and Schwann cells. Malignant melanotic schwannoma is divided into nonpsammomatous and psammomatous subtypes. The presence of psammoma bodies is not related to the metastatic potential of the tumour, nor are the ploidy or size. Gene suppressor PRKAR1A is absent in most MPMS, with a 95% penetrance at 50 years. Interestingly, melanoma never loses the expression of PRKAR1A. The BRAF V600E mutation is negative in MPMS, but it is typically positive in melanoma. S100 protein, HMB45, and Melan-A are also consistent features of MPMS.

Carney Complex

The Carney complex is an autosomal dominant syndrome usually described in young adults (median age of 20 years). It is associated with melanotic schwannoma and skin pigmentary abnormalities (lentigiosis, blue nevi, cutaneous myxoma, and café-au-lait spots), endocrine tumours (or endocrine
Novel Teaching Points

- This is the first report of a left ventricular malignant psammomatous melanotic schwannoma.
- The distinction between MPMS and melanoma is crucial for appropriate management.
- MPMS is a type of tumour with deceptive histologic features and unpredictable behavior. Therefore, it should be considered a malignant neoplasm and treated with total surgical resection when feasible.
- Further studies are needed to determine the role of longitudinal clinical and radiologic surveillance in improving the prognosis of these patients.

Case

An asymptomatic 74-year-old male, with no past medical history, was diagnosed with a nonspecific conduction abnormality on a preoperative electrocardiogram prior to hand surgery. Subsequent 2D transthoracic echocardiography showed a 21 x 17 mm endocardial mass within the LV (Fig. 1, E and F) and a normal LV ejection fraction. Subsequent transesophageal echocardiography (Fig. 1, G and H) and magnetic resonance imaging were performed to further characterize the tumour (Fig. 1, A and B). The lesion appeared to be encapsulated, without invasion of surrounding tissues, such as the mitral apparatus or papillary muscles. The lesion was immobile, and it was attached on one-fourth of its surface on the anteroseptal LV wall, 20 mm from the LV outflow tract. Coronary angiography showed a minimal vascularization of the lesion (Fig. 1C). No other abnormal lesions were identified by computed tomographic scan or positron emission tomography (Fig. 1D). The lesion was not calcified on any imaging modalities. The patient did not experience chest pain, palpitation, dyspnea, or syncope. Physical examination was negative for cutaneous and ocular abnormalities, and Carney complex was ruled out. No cardiomegaly was noted, and cardiopulmonary auscultation was normal. An electrocardiogram showed 45 bpm sinus bradycardia, a 194-ms PR interval, a nonspecific conduction abnormality, and a late R wave progression. Blood samples, including hemoglobin, creatinine, T-troponins, and B-type natriuretic peptide, were all in the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range. Leucocytes and high-sensitivity C-reactive protein were within the normal range.

Comment

MPMS is diagnosed at a mean age of 41 years, whereas melanoma is found in older populations (median of 70 years). Heart involvement in metastatic melanoma usually arises in the context of stage IV widespread disease, with a survival rate as low as 23% at 6 months. New kinase and immune checkpoint inhibitor therapies have significantly improved melanoma survival up to 37% at 5 years.
MPMS both express melanocytic markers, and both commonly show malignant features and necrosis. Further histologic analysis allowed us to rule out metastatic melanoma with an occult primary lesion. The presence of psammoma bodies and the absence of PRKAR1A and BRAF V600 further suggested MPMS. MPMS and melanoma both behave in a clinically malignant manner, but MPMS is associated with better survival. Accurate final diagnosis is therefore crucial to ensure appropriate medical and surgical treatment and guide long-term clinical follow-up and imaging.

Due to the paucity of late data, no guidelines currently exist regarding MPMS management and surveillance. Among 57 patients with MPMS, Vallat-Decouvelaere et al. reported a 26.3% risk of metastases, and a 53% likelihood of achieving disease-free status at 5 years. Torres-Mora et al. reported a local recurrence rate of 35%, and a distant
metastasis rate of 42% in their 40 consecutive MPMS cases, and most patients had recurrent disease within 4 years after the initial diagnosis. Lungs and pleura are the most common locations for metastatic disease. Clinical and histopathologic characteristics are poor predictors of prognosis in MPMS. Only a mitotic rate greater than 2/10 HPF has been suggested as being predictive of malignancy, but the absence of mitotic figures does not completely rule out malignant potential. MPMS should be considered an aggressive type of tumour because of a high risk of local recurrence and metastatic potential. The dearth of long-term follow-up data in MPMS may have led to the underestimation of its malignant potential, and contributed to perpetuating the notion that MPMS is a benign tumour. Therefore, we suggest that all patients with MPMS be given close clinical and radiologic longitudinal follow-up, with a high index of suspicion for local disease recurrence and distant metastases, independent of the presence or absence of malignant microscopic features.

The optimal management of MPMS is complete surgical resection. Adjuvant therapy may be needed for cases of subtotal excision, positive margins, local recurrence, or distant metastases, although its role has not been clearly elucidated for MPMS. The impact of adjuvant therapies in the presence of total resection with negative surgical margins (as in our patient) remains unknown. Further studies are needed to determine the role of longitudinal clinical and radiologic surveillance in improving the prognosis of these patients.

**Acknowledgements**

The authors thank Dr Christopher D.M. Fletcher from the Dana-Farber Cancer Institute, Boston, MA, for his contribution to the diagnosis and management of the patient described in this report.

**Funding Sources**

No funding was received for this work.

**Disclosures**

The authors have no conflicts of interest to disclose.
References

1. Torres-Mora J, Dry S, Li X, et al. Malignant melanotic schwannian tumor. Am J Surg Pathol 2014;38:94-105.

2. Gelfand ET, Taylor RF, Hendin D, Akabutu J, Callaghan JC. Melanotic malignant schwannoma of the right atrium. J Thorac Cardiovasc Surg 1977;74:808-12.

3. Shields C, Glassman S, Shields L, Raque G. Malignant psammomatous melanotic schwannoma of the spine: a component of Carney complex. Surg Neurol Int 2011;2:136.

4. Alexiev B, Chou P, Jennings L. Pathology of melanotic schwannoma. Arch Pathol Lab Med 2018;142:1517-23.

5. Stratakis CA, Raygada M. Carney Complex. In: Adam MP, Ardinger HH, Pagon RA, et al., eds. GeneReviews. Available at: https://www.ncbi.nlm.nih.gov/books/NBK1286/. Accessed December 16, 2018.

6. Vallat-Decouvelaere A, Wassef M, Lot G, et al. Spinal melanotic schwannoma: a tumor with poor prognosis. Histopathology 1999;35:558-66.

7. Endo A, Ohtahara A, Kinugawa T, et al. Characteristics of 161 patients with cardiac tumors diagnosed during 1993 and 1994 in Japan. Am J Cardiol 1997;79:1708-11.

8. Ugurel S, Röhmel J, Ascierto PA, et al. Survival of patients with advanced metastatic melanoma: the impact of novel therapies. Eur J Cancer 2016;53:125-34.