Abstract We present the case of a 60-year-old man with a primary pulmonary melanotic schwannoma treated with surgery and who developed an orbital and myocardial relapse 2 years after the initial diagnosis. Melanotic schwannomas are rare pigmented tumours that tend to arise from the peripheral nerves near the midline. A primary lung presentation, as in our case, is extremely rare. In more than half of cases, the Carney triad of myxomas of the heart, skin and breast, spotty pigmentation and endocrine hyperactivity is present. A thorough pathological study is pivotal for a correct diagnosis. The main differential diagnosis is with metastases of malignant melanoma. The biological behaviour is unpredictable. Treatment should include radical surgery if possible; the role of chemotherapy and radiotherapy is uncertain due to the rarity of the tumour.

Keywords Carney syndrome · Melanin · Melanoma · Sarcoma · Schwannoma

Introduction

Melanotic schwannomas are infrequent soft tissue tumours that usually arise from peripheral nerves near the midline [1]. Primary pulmonary involvement is very rare. Due to their infrequency, the biologic behaviour of these neoplasms is uncertain. We report the case of a patient with a primary pulmonary schwannoma treated with surgery and who developed a late orbital and myocardial relapse. We also review the literature regarding the pathological diagnosis and clinical characteristics of this rare tumour and the different treatment recommendations.

Case report

An incidental solitary nodule in the upper lobe of the left lung was diagnosed in an asymptomatic 60-year-old man in February 2004. He had no previous medical history of interest and had never smoked. His family history was unremarkable. A left upper segmentectomy was performed, which disclosed an encapsulated, well-circumscribed pigmented tumour of 3.7 cm maximum diameter. Microscopically, the tumour was highly cellular, although it was difficult to make out cellular detail because of the heavy melanin pigment deposits. The cells, which were of mesenchymal origin, varied in shape from epithelioid to spindled and blended gradually from one to another. The cells showed no mitoses or frank atypias, and the proliferation index, measured by Ki-67, was <1%. Occasionally, there was vague cell palisading, which resembled a schwannoma or neurofibroma. On immunohistochemical studies, the cells expressed S-100 protein and a melanoma-associated antigen (HMB-45). No Psammoma bodies were found. A thorough clinical history and physical examination revealed no previous or synchronic malignant melanoma, and all staging studies performed were negative. The diagnosis of primary pulmonary melanotic schwannoma was given. No adjuvant treatment was given, and the patient began follow-up in our clinic.

Two years after the initial diagnosis, in June 2006, an engrossed myocardial wall was discovered in a routine computed tomography (CT) scan. The patient had no cardiac symptomatology. A cardiac magnetic resonance imaging (MRI) showed a grossly dilated left ventricle with an engrossed myocardium secondary to small infiltrative...
masses. The left ventricle ejection fraction was severely depressed (14%). There was a single 3 × 4-cm mass in the right ventricle. All these lesions showed melanin production in MRI images. However, an initial endomyocardial biopsy produced no conclusive results.

In parallel, the patient referred intermittent headache and a binocular diplopia. An orbital MRI showed several discrete orbital bony lesions that were again melanin positive. A new whole-body CT scan performed disclosed several intraconal masses in both orbits (Fig. 1), enlarged mediastinal and left supraclavicular nodes and a pericardial effusion with a severely engrossed myocardium. A fine-needle aspiration of the supraclavicular node only showed scant mesenchymal cells with no atypias. The orbital biopsy was refused because of the high anaesthetic risk. However, a new endomyocardial biopsy was done, which revealed a diffuse infiltration by mesenchymal spindled cells, with no atypias or mitoses and a Ki-67 index <5% (Fig. 2). The cells were melanin-positive in a granular fashion, and immunohistochemical studies showed frank positivity for vimentin, CD68, S-100 (Fig. 3) and HMB-45 expression. The cells were similar to those of the initial diagnosis.

The diagnosis of myocardial, orbital and supraclavicular relapse of melanotic schwannoma was reached. The patient had begun to show signs of heart failure, and medical treatment with diuretics and angiotensin-converting enzyme (ACE) inhibitors was started. As anthracyclines and ifosfamide were contraindicated, we began systemic treatment with continuous temozolomide (100 mg/m²/day). Unfortunately, the patient’s condition worsened quite rapidly, and he died of refractory heart failure 6 weeks after. The family did not give permission for an autopsy.

Discussion

Melanotic schwannomas are rare pigmented tumours of neural origin that tend to arise from the spinal or autonomic nerves near the midline and were originally described by Millar in 1932 [1]. The term melanocytic schwannoma was coined by Fu et al. in 1975 [2]. Despite the similarity in their names, the lesion is a distinctive neoplasm of adult life that differs significantly from classic schwannoma in age of presentation and clinical behaviour [1–4]. Although these lesions are of neural origin, a number of cases have been reported in the stomach, bone and soft tissues. Other unusual sites include the heart, liver and skin [3]. Primary pulmonary involvement, as in our case, is extremely rare.

More than half of cases have evidence of Carney syndrome [4, 5], which includes myxomas of the heart, skin and breast, spotty pigmentation (due to lentigines, blue nevus and the distinctive epithelioid blue nevus [6]), and endocrine hyperactivity (Cushing’s disease, acromegaly or sexual precocity). About 20% of patients have multiple tumours, and in such patients, there is an ever higher probability that other manifestations of the complex will be present [3]. However, our patient had no evidence of the complex. The tumour typically develops at a younger age in patients with Carney syndrome, and it appears to have an overall worse prognosis due to the higher relapse rate and metastatic potential [4, 5]. Clinical presentation is varied and nonspecific and usually depends on tumour location and growth rate. Due to its neural origin, pain and neurologic deficits in the affected part are the most common presentations [2, 3]. The presence of melanin gives the