**Case Report**

**Syndromic or Sporadic? Atrial Myxoma as a Component of Carney’s Complex**

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**Introduction**

Incidentally detected atrial myxomas warrant attention to whether a myxoma is syndromic or sporadic in origin. Atrial myxoma is one of the diagnostic criteria of carney’s complex, a rare hereditary disorder associated with neoplasia, and endocrine over-activity that may involve multiple endocrinopathies of which thyroid disease is the most common. Differentiating between syndromic and sporadic myxomas is important as syndromic myxomas have a higher recurrence rate necessitating more frequent follow up and tend to present in atypical locations. This diagnostic challenge is demonstrated by the presentation of a 41-year-old Caucasian female who presented with three months of atypical chest pain, palpitations, exertional dyspnea, and pedal edema.

The patient’s history revealed multiple complaints with extensive evaluations over a period of nine years, including, morbid obesity (BMI 42.7 kg/m²), hypertension, hyperthyroidism, anemia, and abdominal discomfort. Of late, she complained of headache with decreased visual acuity and colorful flares, and symptoms of sciatica. Her family history was positive for breast cancer.

In addition, her prior evaluations revealed elevated anti-thyroid antibodies (anti-thyroid peroxidase and anti-thyroglobulin) consistent with Hashimoto’s thyroiditis, hepatomegaly with 3 × 2 × 2 cm hypoechoic right liver lobe mass and a stable 5 mm left pleural pulmonary nodule. Magnetic resonance imaging of her brain showed slight supra-sellar expansion and partially empty sella with bilaterally dilated optic nerve sheaths more prominent on the right.

**Presentation**

Physical examination revealed no fever, blood pressure 148/96 mmHg, heart rate 82 beats/min, respiratory rate 16/min, and O₂ saturation 96% on room air. The thyroid gland was diffusely enlarged with multiple nodules.
Lungs were clear. Cardiac point of maximum impulse was normal, and auscultation detected no abnormal findings, including no tumor plop. There was mild non-pitting edema below the knee.

**Diagnosis**

The patient's electrocardiogram (ECG) revealed normal sinus rhythm with mild T wave inversions in the inferolateral leads. Pertinent laboratory findings: thyroid stimulating hormone 2.1 mIU/L (ref 0.4-4.0 mIU/L), hemoglobin 8.8 g/dl (ref 12-15.5 g/dl), and serum calcium 8.4 mg/dl (ref 8.5-10.2 mg/dl). Ambulatory ECG monitoring (14 consecutive days) revealed frequent premature atrial and ventricular complexes with no sustained arrhythmias. Transthoracic echocardiography (TTE) demonstrated a 2 × 2 cm spherical, pedunculated mass in the left atrium (LA), mild concentric left ventricular (LV) hypertrophy, normal LV diastolic function and LV ejection fraction of 65%. Transesophageal echocardiogram (TEE) confirmed the intra-atrial mass consistent with myxoma in the LA with its stalk attached to the supero-anterior interatrial septum (Figure 1). There was no apparent vascularity. There were no masses in the left atrial appendage or other cardiac chambers. Coronary angiography revealed normal coronary arteries and left ventriculography showed normal ejection fraction (60%).

![Figure 1](image_url)

**Figure 1.** Two-dimensional transesophageal echocardiogram revealing a 2 × 2 cm mass (thick arrow) in the left atrium above the aorto-mitral curtain (thin arrow); LA: Left Atrium; AO: Aorta.

A diagnosis of LA myxoma was made and a gelatinous mass adherent to the superior lateral free wall of the LA was excised at surgery (Figure 2). The diagnosis was confirmed by histopathological examination.

**Comment**

The majority of atrial myxomas is isolated and only 5-10% are syndromic in association with Carney's complex [1]. Carney's complex is a rare, autosomal dominant, genetic disorder with multiple neoplasia, lentiginosis, and endocrine over-activity that may involve the pituitary gland, adrenals and testes. Various thyroid gland abnormalities have been described, ranging from follicular hyperplasia to cystic changes and carcinoma. Thyroid nodular or cystic changes are the most common endocrine disorder in association with Carney’s complex with a frequency of 75% [2].
Atrial myxomas in Carney’s complex are histologically indistinguishable from the sporadic forms, yet their clinical presentation and course are distinct [3]. The diagnosis requires either two of twelve criteria or one criterion plus either a positive family history or a PRKAR1A gene mutation [4]. Our patient’s myxoma was an incidental finding during evaluation of her chest pain. A history of endocrine abnormality and a diffuse multinodular goiter raised suspicion that the myxoma was part of a complex rather than sporadic. Adherence of the tumor to the LA free wall is rare and such atypical location has been described in syndromic cases [5,6]. The patient’s evaluation failed to identify any evidence of hepatocellular and/or pituitary adenoma, spinal schwannoma, or bronchogenic cysts, all of which were potential sources of the patient’s complaints and have been described in literature in association with Carney’s complex.

Figure 2. Gross specimen showing left atrial gelatinous myxoma after complete surgical excision.

Carney’s complex has been commonly referred to as a constellation of myxomas, endocrine over-activity, and mottled skin pigmentation [7], with the latter being the most commonly identifiable abnormality. It has also been reported that the diagnosis of Carney’s complex requires only two criteria such as thyroid nodules and cardiac myxoma [4]; therefore, our patient’s manifestations are consistent with Carney’s complex rather than merely incidental findings and that would make the patient’s left atrial myxoma a syndromic rather than a sporadic one. Syndromic atrial myxomas are usually encountered in younger patients of median age 22 years [1] compared to mean age of 55 for sporadic cases [3]. Fifty percent of myxomas arise from the interatrial septum [8] and are usually attached to the fossa ovalis. Origin from the anterior mitral leaflet and left atrial appendage have also been described [9]. Our patient’s myxoma was adherent to the superior-lateral wall of the LA between the appendage and the mitral valve. Most patients with cardiac myxoma present with one or more of the triad of constitutional, embolic or obstructive manifestations [10]. Our patient presented atypically with chest pain and palpitations in the absence of an obstructing mass [11] or coronary abnormalities.
Conclusion

Determining whether atrial myxoma is sporadic or part of Carney's complex can be challenging given the various expressivity of the complex and the unpredictability of the temporal emergence of diagnostic signs. The importance of distinguishing between sporadic and syndromic myxomas is the high recurrence rate of syndromic cases (22%) even after surgical resection, compared to 1-3% for sporadic tumors [8,12,13], necessitating post-operative follow-up and careful screening of family members. Increasing awareness of the multiple clinical manifestations of Carney's complex and the diagnostic criteria can enhance physicians’ accuracy in identifying syndromic cardiac myxomas and highlighting the need for clarification of guidelines on the diagnosis, management, and prognosis of Carney's complex.

Conflict of Interest

The authors declare no potential conflicts of interest.

References