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Carney Complex: Eleven Open Heart Operations in a Single Family

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Herein, we report on a family with Carney complex. Four members of the family underwent a total of 11 open heart operations as well as 9 other operations to treat extrathoracic masses. All the family members met at least 2 major clinical criteria and 1 supplemental criterion. We analyzed their genomic loci, including the protein kinase A regulatory subunit 1 gene. The results revealed no specific mutations, except for a common single nucleotide polymorphism. This case series of Carney complex emphasizes the importance of close longitudinal follow-up because of the high rate of tumor recurrence irrespective of the site. Clinicians should not overlook the specific features of familial myxoma.

Key words: 1. Carney complex
2. Myxoma
3. Familial myxoma

Case report

We observed a case series of 3 siblings who underwent 8 open heart operations for recurrent myxoma and conducted a thorough analysis of the cases, including the family history and chart reviews (Fig. 1). The results indicated that the mother of the siblings underwent 3 open heart operations for recurrent myxoma, as well as 3 other extrathoracic mass-removal operations. She died of malignant schwannoma with bone and lung metastasis in 2014. The proband, the first son, underwent 3 open heart operations for myxoma as well as 3 other extrathoracic operations. The proband's younger brother, the second son, underwent a total of 6 mass-removal operations, along with 4 open heart operations. Lastly, the proband's sister underwent 1 right ven-

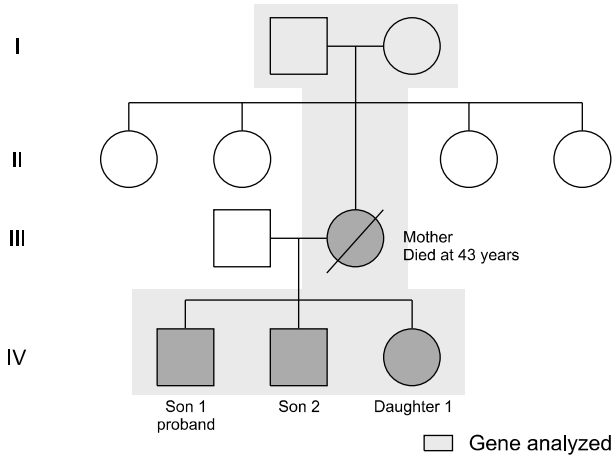

Fig. 1. Family pedigree.

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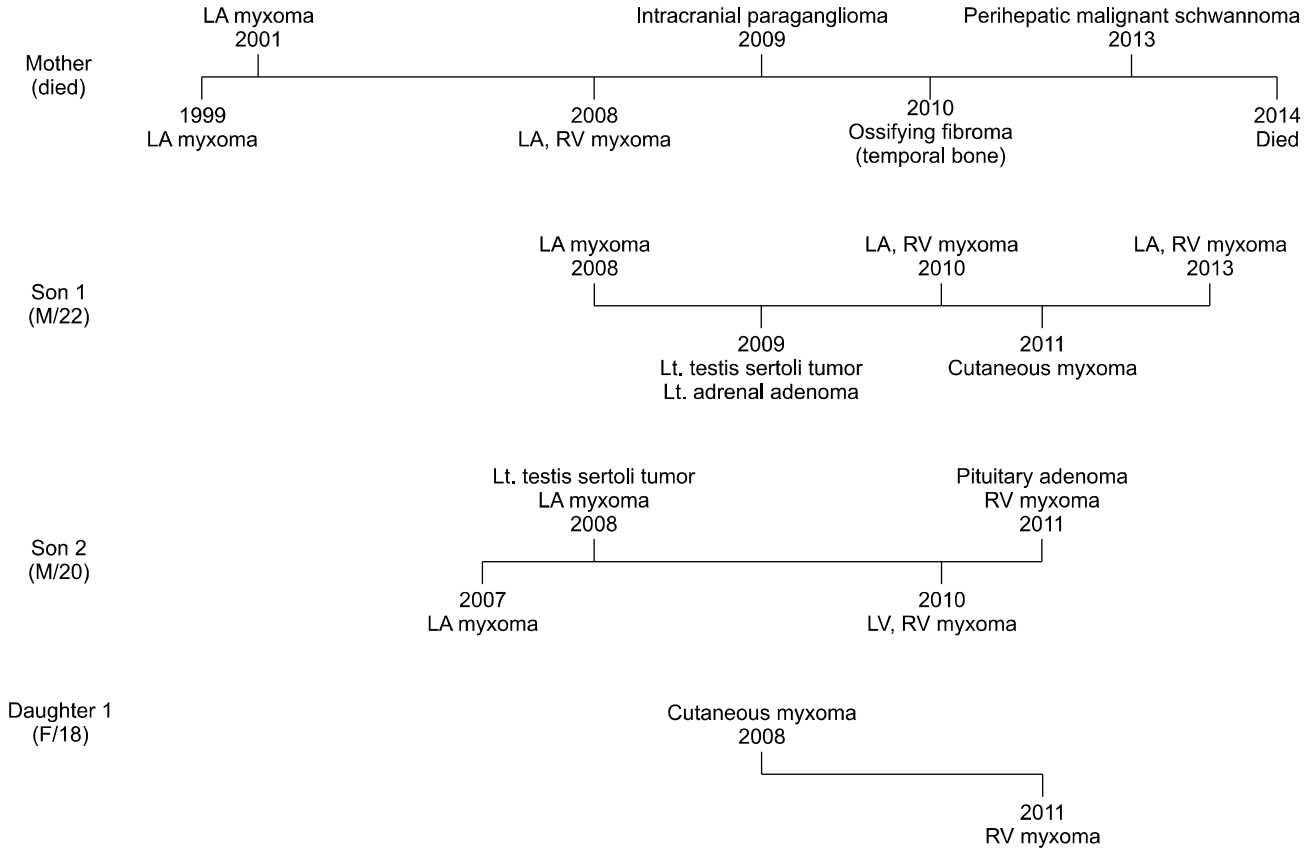

Fig. 2. Summary of surgical histories of the subjects of this study. M, male; F, female; LA, left atrial; RV, right ventricular; Lt., left.

tricular myxoma removal and 1 left chest wall myxoma removal operation. The surgical histories of the family members are summarized in Fig. 2. The 4 family members met the diagnostic criteria of Carney complex. They met at least 2 major clinical criteria (including spotty skin pigmentation, myxoma, acromegaly, or multiple endocrine tumors) and 1 supplemental criterion (an affected first-degree relative). We analyzed the genomic loci of Carney complex 1 and Carney complex 2, including the protein kinase A regulatory subunit 1 (*PRKAR1A*) gene. However, the results revealed no specific mutations, except for a common single nucleotide polymorphism.

Discussion

Carney complex is a rare multineoplastic disorder that was first described by Carney et al. [1] in 1985. It is characterized by skin pigmentation, cardiac myxoma, and endocrine overactivity [2]. It is usually inherited in an autosomal dominant pattern with variable penetrance. A heterozygous mutation of the *PRKAR1A* gene can be observed in approximately 50% of affected patients [3]. In our case series, we found no specific mutations except for a common single nucleotide polymorphism. Cardiac myxomas are the second-most common manifestations of Carney complex, following spotty skin pigmentation [4]. Compared to sporadic cardiac myxoma, familial cardiac myxoma has distinctive features. There are usually multiple rather than solitary myxomas, and they occur in atypical locations, rather than in the left atrium [5]. Additionally, familial cardiac myxoma has a high recurrence rate and usually affects younger individuals. In cases of cardiac myxoma with patients presenting the above features, the possibility that it is of the familial type should be considered, because such myxomas can behave like a malignancy, as in our case series.

This case series involving 4 members through 2 generations highlights the need for vigorous screening of the family members of patients with Carney

complex. Routine annual echocardiographic surveillance is also reasonable because of the relatively short interval between recurrences of cardiac myxoma. The high rate of tumor recurrence affecting both cardiac and extrathoracic sites highlights the importance of close longitudinal lifetime follow-up. To the best of our knowledge, no such series of cases within a single family has previously been reported.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

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