

Unusual Presentation of a Family with Thymoma: A Case Report¹

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Thymoma is an uncommon tumor of childhood and familial occurrence is very rare. Here we report two sisters and their father with thymoma identified by imaging findings.

Index words : Mediastinum

Thymus, neoplasms

Thymic lesions account for approximately 2 - 3% of all pediatric mediastinal tumors and include thymic cysts, hyperplasia, carcinoma, thymoma and thymolipoma (1). Thymomas are rare tumors of the thymic epithelium with a broad spectrum of morphological and clinical features; they comprise about 1% of mediastinal tumors. Familial occurrence of these tumors has been rarely reported (2 - 4). We report an unusual familial occurrence of three cases of thymoma in two sisters and their father.

Case Reports

Case 1

A 13-year-old girl was admitted to our hospital for further evaluation of a mediastinal mass that was detected on a chest radiograph. She was followed for idiopathic thrombocytopenic purpura and alopecia, without myasthenia gravis, over a period of ten years. An initial chest radiograph showed an anterior mediastinal mass obliterating the silhouette of the ascending aorta and upper portion of the right lateral border of the heart (Fig. 1A).

Chest CT showed a large lobulated anterior mediastinal mass (about 14 × 9 cm in size) with enhancing solid portions and non-enhancing cystic or necrotic regions, as well as a small lymph node in the left axilla (Figs. 1B and C). Abdominal CT revealed small nodules in both kidneys and a small left paraaortic lymphadenopathy (Fig. 1D). To remove the anterior mediastinal mass, a total thymectomy was performed. The final pathological result was thymoma, WHO type A, with minimal invasion into surrounding fat tissue. The findings from a ¹¹¹In octeroscintigraphy (not shown here), performed during postoperative chemotherapy, showed mild focal uptake in the left axilla, suggesting a thymoma stage IVb according to the Masaoka classification. After chemotherapy, both renal masses and the left paraaortic lymph node resolved as demonstrated by follow-up abdominal CT (not shown here); the size of the left axillary lymph node was slightly decreased on follow-up chest CT (not shown here). The patient has been followed under close observation in the outpatient department without significant complications.

Case 2

Six years ago, an older sister (six years older than the case 1 patient), without myasthenia gravis, was found to have a well-defined anterior mediastinal mass on a chest radiograph (Fig. 2A). Chest CT (Fig. 2B and C) showed a contrast-enhancing homogeneous solid mass in the thy-

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mus. Pathological investigations after thymectomy confirmed a minimally invasive thymoma ($9.0 \times 8.0 \times 4.0 \text{ cm}^3$). The postoperative period was also uneventful in this patient.

Case 3

Their father [case 1 and 2] did not have myasthenia gravis, and underwent surgical removal of pathologically proven thymoma (of epithelial type) 25 years previously; he died 14 years later due to liver cirrhosis. A

non-enhanced chest CT (Fig. 3) revealed a homogeneous solid mass in the thymus.

Discussion

Thymomas in childhood usually occur in isolation and are only rarely associated with myasthenia gravis (5). About 10% of the patients with thymoma may present with a variety of autoimmune diseases (1). Thymomas occur as one of three different histological classes: pre-

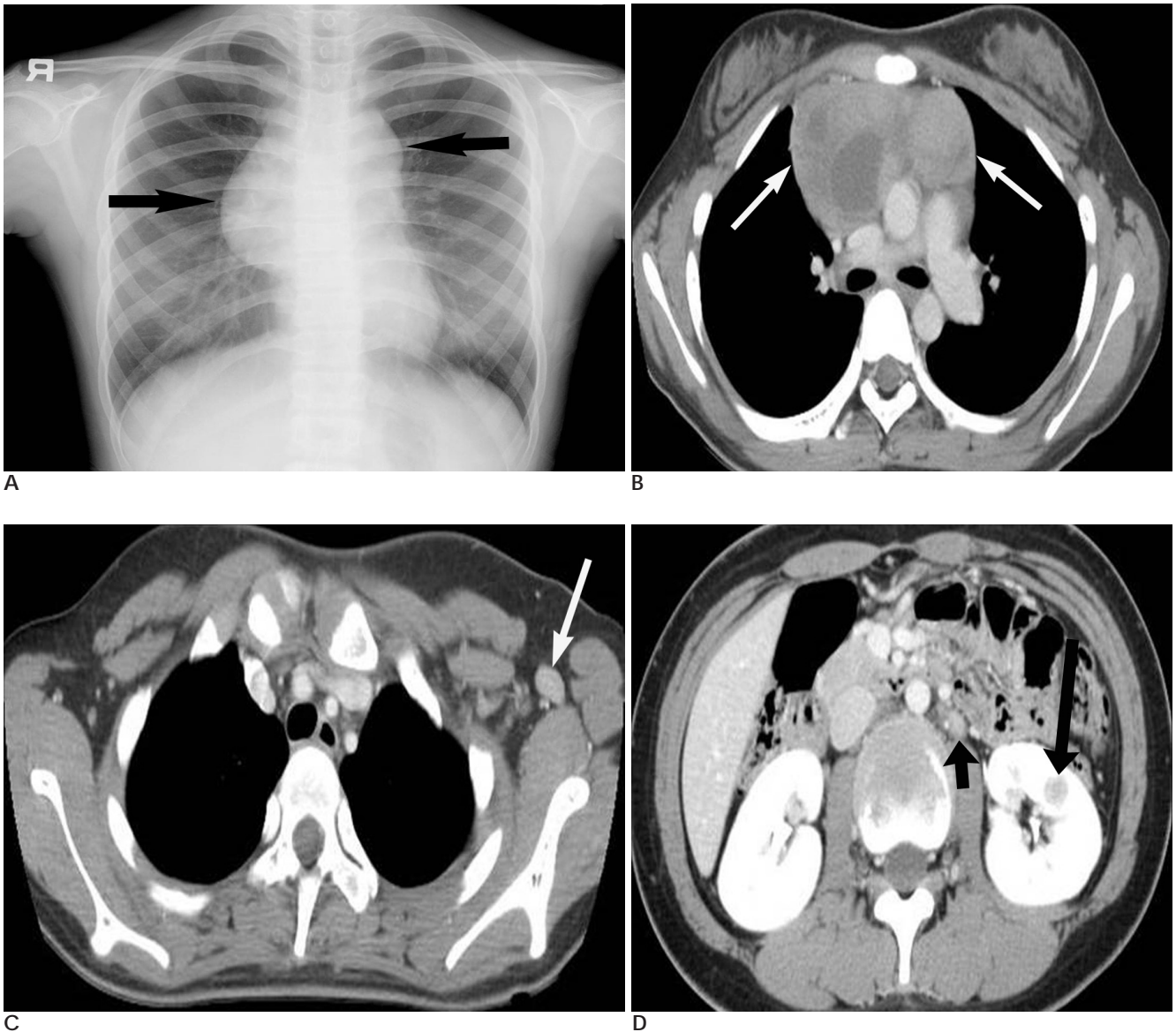
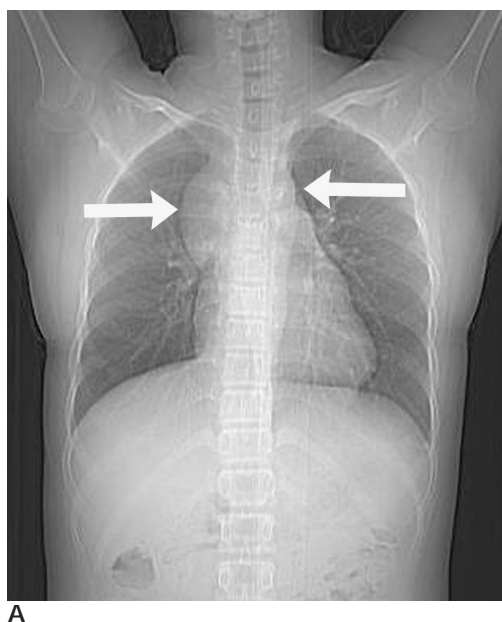


Fig. 1. Case 1. A 13-year-old girl with invasive thymoma.

A. Chest PA shows an anterior mediastinal mass (arrows) obliterating the silhouette of the ascending aorta and upper portion of right lateral border of the heart.

B, C. Contrast-enhanced chest CT demonstrates a large lobulated anterior mediastinal mass (arrows in B) with enhancing solid portions and non-enhancing cystic or necrotic regions. Note the small lymph node in the left axilla (arrow in C).

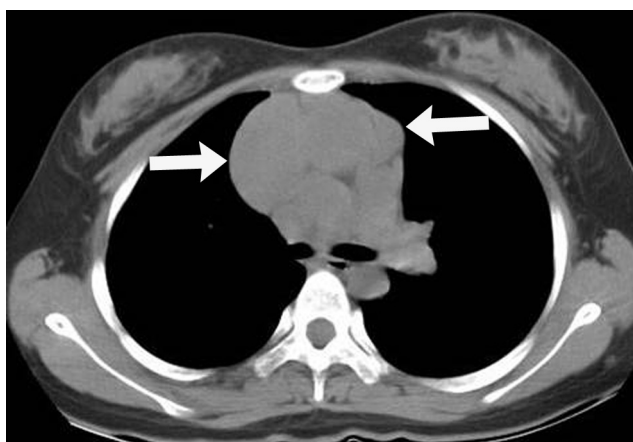
D. Contrast-enhanced abdominal CT reveals small nodules in the left kidney (long arrow) and small lymph nodes in the left paraaortic area (short arrow).



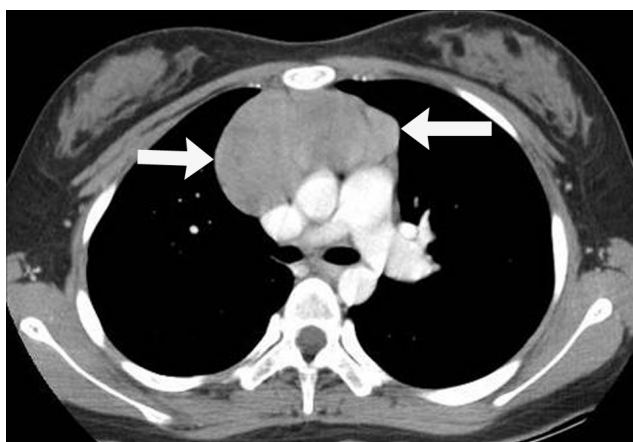
A

Fig. 2. Case 2. The older sister (six years older than in case 1 patient) with minimally invasive thymoma.

A. CT scanogram shows an anterior mediastinal mass (arrows) with a convex border at the right lateral aspect. Non-enhanced (**B**) and contrast-enhanced (**C**) chest CT scans show a contrast-enhancing homogeneous solid mass in the thymus (arrows).



B



C

dominantly lymphocytic, mixed lymphoepithelial, and predominantly epithelial (6).

The radiological findings of thymoma vary from a small focal heterogeneous mass adjoining either thymic lobe to a large lobulated tumor that replaces the whole thymus and distorts the mediastinal structures. Imaging features of thymoma, especially a large and lobulated one, are similar to the more common mediastinal lymphoma; although calcification of the untreated mass is more common in thymomas (7). Calcification in thymoma, however, is also uncommon (5). The three thymomas in our cases did not show any calcifications by imaging and pathological evaluation of specimens. If a mass is found in thymus in pediatric cases, lymphoma, Langerhans cell histiocytosis and germ cell tumors should be differentiated from a thymoma.

CT is the imaging modality of choice for staging of thymoma; it provides information on the location, size, extent of involvement of the tumor and the presence/absence or the extent of tumor invasion into adjacent structures. Variable low attenuation with moderate contrast enhancement is known to be characteristic of this

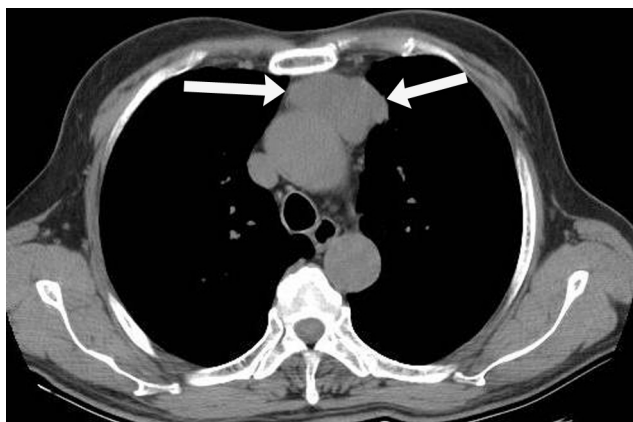


Fig. 3. Case 3. The father with thymoma of the epithelial type. Non-enhanced chest CT reveals a small homogeneous solid mass in the thymus (arrows).

tumor (5). Irregular borders between the lesion and adjacent lung or mediastinal structures suggest malignant invasion, such as vascular invasion or encasement, and pericardial or pleural invasion (8). Malignant lesions tend to be more invasive with pleural encasement, pericardial invasion and pulmonary metastasis (5).

Although our three patients were confirmed to have pathologically proven thymoma, it is very difficult to determine whether there is an association of the thymoma with a familial genetic inheritance. There are only a few reported cases of familial thymic masses. Lam et al reported invasive cystic thymoma in two siblings (an 11-year-old girl and a 9-year-old boy) with the radiographic, CT and ultrasonographic features (2). Matani and Dritsas described a lymphocytic thymoma in a 27-month-old girl and her 9-month-old brother (3). Wuketich and Zwintz reported squamous cell thymoma in a 58-year-old male and his 59-year-old sister (4). There is no prior report on the genetics of familial thymoma. Further collection of familial thymoma cases and research on the genetics of this disorder is needed. These tumors are usually aggressive, with rapid onset of symptoms and a high mortality rate (7); this is probably due to incomplete surgical resection. Rothstein et al. (1) reported that a 14-year-old girl with thymoma showed no radiographic evidence of recurrence and returned to all her usual activities at eight months after surgery. Lam et al (2) reported that the postoperative period of two siblings (an 11-year-old girl and a 9-year-old boy) with invasive thymoma was uneventful.

In summary, we present an unusual case of pediatric thymoma in two sisters and adult thymoma in their father with imaging findings.

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