

Familial Spontaneous Pneumothorax in Two Adult Siblings with Marfan Syndrome

Toshiaki Suzuki, MD, Tadashi Akiba, MD, PhD, Ryo Miyake, MD, Hideki Marushima, MD,
and Toshiaki Morikawa, MD, PhD

A 21-year-old adult with previously diagnosed Marfan syndrome (MS) was admitted to our hospital complaining of chest pains that resulted from a complete left spontaneous pneumothorax (SP). A chest roentgenogram showed a 70% pneumothorax on the left side. Because for 8 days chest tube drainage did not ameliorate the air leak, and also because a computed tomography (CT) scan showed many cystic opacities at the apical lung, a bullectomy was performed under video-assisted thoracic surgery (VATS). The patient's family history revealed that his father and two brothers had MS. Three family members had a unilateral SP, and his two siblings were treated under VATS and showed no evidence of recurrent pneumothorax. The simultaneous occurrence of MS and SP in these two patients suggests the possibility that both abnormalities in pulmonary connective tissue (e.g., collagen I) and the tall figure are responsible for the development of SP in MS patients. Because our patients and those in published reports are few in number, no definitive conclusion can be drawn about the relationship between SP and MS. Herein we describe a rare case of familial SP in two adult siblings with MS. (Ann Thorac Cardiovasc Surg 2010; 16: 362–364)

Key words: lung, pneumothorax, thoracoscopy/VATS

Introduction

We report a case of familial spontaneous pneumothorax (SP) with Marfan syndrome (MS) in whom video-assisted thoracic surgery (VATS) was successfully performed. A case of this kind is rare and has not been previously reported as having occurred.

Case

A 21-year-old man was admitted to our hospital com-

plaining of chest pains and dyspnea. Over the preceding 6 months, he had experienced a few episodes of mild pain in the left side of his chest, but otherwise had been well and experienced no breathing difficulties. A chest roentgenogram showed a 70% pneumothorax on the left side. Because 8 days of chest tube drainage did not ameliorate the air leak and a computed tomography (CT) scan showed many cystic opacities at the apical lung, a bullectomy was performed under VATS (Fig. 1). The patient's postoperative course was uneventful, and he was discharged on postoperative day 7.

Our patient was born of consanguineous parents and exhibited many of the skeletal manifestations of MS. The MS diagnosis was therefore based on the family history of the disorder, dysmorphic features (including dolichocephaly), a prominent forehead with abundant skin, a high-arched palate, micrognathia, a narrow chest, arachnodactyly, and flat feet. Lens dislocation and mitral valve prolapse were not observed.

From Department of General Thoracic Surgery, Jikei University School of Medicine, Tokyo, Japan

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Address reprint requests to Toshiaki Suzuki, MD: Department of General Thoracic Surgery, Jikei University School of Medicine, 163-1 Kashiwashita, Kashiwa, Chiba 277-8567, Japan.
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Fig. 1. A bulla of the left lung is shown during an operation.

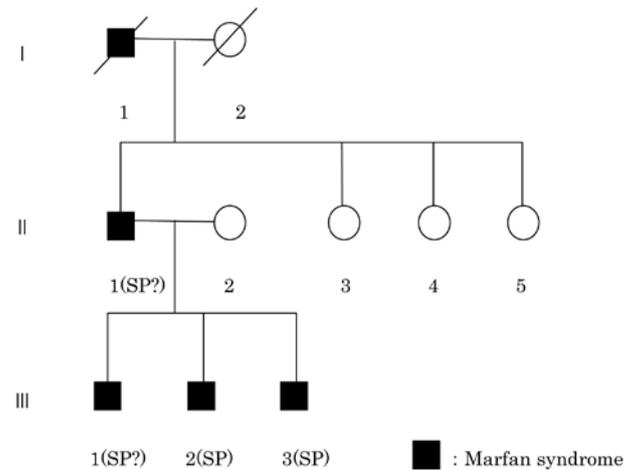


Fig. 2. Three-generation pedigrees, showing family members with Marfan syndrome (black squares) and/or spontaneous pneumothorax (SP) and their relationships with other family members.

Family History

The patient's family history, shown in Fig. 2, reveals that his grandfather died suddenly in his 40s. It is possible that he had had MS, considering that he was tall and had arachnodactyly. The patient's father, 55 years old, 198 cm tall, and weighing 83 kg, was found to have had aortic dissection 7 years previously, and he had undergone surgery to implant a blood vessel prosthesis. The surgical findings indicated that the patient had experienced a unilateral SP.

The patient's older brother, 25 years old, 184 cm tall, and weighing 60 kg, had experienced several episodes of chest pain, but he had not sought medical attention and possibly had a history of SP.

The patient's younger brother, 18 years old, 185 cm tall, and weighing 60 kg, had experienced an episode of SP and been treated under VATS in the previous year.

Comment

MS patients, however, are prone to form bullae or blebs, and SP has been described in about 10% of them. It is the most acute of pulmonary manifestations.^{1,2)} Hall et al. reported that the frequency of SP was 4.4% in patients with MS who are more than 12 years old.³⁾ A patient with MS is 10 times more likely to sustain SP than healthy individuals are.⁴⁾

SP is thought to be mainly due to an abnormality in collagen fibers, such as collagen I, which leads to focal

flaccidity and reduced tensile strength in the walls of terminal bronchioles, which results in the trapping of expiratory air.^{5,6)} However, Takamami et al. reported that there were no significant differences in collagen distribution between the lungs of MS patients and those of controls.⁷⁾ They reported that physical forces such as the tall figure in MS patients have a greater effect than abnormalities in collagen I. The origin of SP is probably multifactorial, but bullae are more likely to form and rupture because of the increased forces generated at the lung apices in tall persons.^{8,9)} Pectus excavatum or progressive scoliosis can contribute to a restrictive pattern of lung disease.¹⁰⁾

No optimal treatment is known. Sahn et al. and Hall et al. reported in a retrospective study that 5 of 11 patients with MS and SP were treated with chest drainage, 3 had no recurrence, and 6 of the 11 underwent surgery.^{3,11)} The treatment of choice depends on the size of the pneumothorax, severity of the symptoms, and level of persistence of the air leak. Both Hall et al. and Rigante et al. reported that surgery should be performed on MS patients at the first occurrence of SP because there is a high rate of recurrence after treatment with a chest tube.^{3,4)}

Sugiyama et al. reported their findings for two brothers, each of whom experienced a single episode of unilateral SP.¹²⁾

Our report is unique because three members of the same family with MS each experienced a single episode of unilateral SP, two of whom in the same generation experienced SP and were treated successfully under VATS. The simultaneous occurrence of MS and SP in

these two patients suggests the possibility that both abnormalities in pulmonary connective tissue (e.g., collagen I) and the tall figure are responsible for the development of SP in MS patients. Because our patients and those in published reports are too few, no definitive conclusion can be drawn about the relationship between SP and MS. Furthermore, MS has many types of expressions, and some documents reported the gene mutations associated with familial SP. More research of gene mutations will be needed to identify the relations between MS and SP.

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