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Short reports

Familial spontaneous pneumothorax

YUKIHIKO SUGIYAMA, HIROO MAEDA, HIDEKI YOTSUMOTO, FUMIMARO TAKAKU

From the Third Department of Internal Medicine, and the Blood Transfusion Service, Faculty of Medicine, University of Tokyo, Tokyo, Japan

Spontaneous pneumothorax usually occurs sporadically in healthy young adults, who tend to be tall and to have an asthenic habitus. The underlying cause is thought to be rupture of small subpleural lung cysts or bullae that are found in the apex of the upper lobe. On the other hand, familial spontaneous pneumothorax is known to be a rare condition, with sparse published reports.[1-8] We here report the cases of two brothers who presented with spontaneous pneumothorax and had Marfan syndrome like abnormalities in the skeletal system. The results of HLA analysis of this family are also presented.

Case reports

PATIENT 1 (PROBAND)

An 18 year old Japanese boy was admitted to our hospital in November 1984 because of dyspnoea and chest pain. A chest radiograph showed a small right apical pneumothorax without any evidence of bullae. This was treated conservatively, without the insertion of an intercostal tube. Twenty days later the patient showed complete re-expansion of the affected side.

He was a slim boy 170 cm in height and 45 kg in weight. His arm span was 173 cm and the metacarpal index was above the upper limit of the normal level (8·0)—8·69 on the right and 9·62 on the left. He had the "wrist sign" characteristic of the Marfan syndrome,[9] but he had no thoracic deformities. Complete ophthalmological and cardiac examinations, including a search for ectopia lentis and anterior chamber angle abnormalities, and echocardiography revealed no abnormalities. The serum progesterone concentration was below 1·0 ng/ml and serum α_1 antitrypsin 215·0 (normal 192–264) mg/100 ml.

PATIENT 2

The brother of patient 1 had suffered from spontaneous pneumothorax and underwent thoracotomy in 1982 at the age of 20. Pathological examination of the resected bullae revealed nothing remarkable. His height was 174 cm and weight 52·5 kg. His arm span was 175 cm and the metacarpal index was 8·1 on the right and 8·9 on the left. He had no thoracic deformities and no wrist sign. Complete ophthalmological and cardiac examination showed no abnormalities. Serum progesterone concentration was below 1·0 ng/ml and serum α_1 antitrypsin 259·0 mg/100 ml.

THE FAMILY

Neither patient showed any mental retardation. Their parents and one sister had been in generally excellent health all their lives, and had no skeletal abnormalities. A radiological bone survey and complete ophthalmological and cardiac examinations, including echocardiography, revealed no abnormalities among them. The father had nine siblings and the mother had five, none of whom had had any episode of pneumothorax or manifestations of the Marfan syndrome.

In these family members, HLA-A, -B, and -C determinants were investigated by the standard typing technique, the National Institutes of Health lymphocyte toxicity method being used, with 120 different HLA antisera. The affected brothers were both A2-B15-Cw3/A11-Bw55; the unaffected sister was A2-Bw60-Cw3/A31-Bw54-Cw1. The father was A2-B15-Cw3/A31-Bw54-Cw1, and the mother A2-Bw60-Cw3/A11-Bw55.

Discussion

It is well known that patients with the Marfan syndrome may develop spontaneous pneumothorax.[10] Although patients 1 and 2 had skeletal abnormalities, they appeared not to meet the established criteria for the Marfan syndrome, since they had no ocular or cardiac abnormalities or family history.[11] Nevertheless, the fact that both patients suffered from spontaneous pneumothorax and had high metacarpal indices suggests the possibility that they had "formes frustes" of the Marfan syndrome. It is not clear whether the form of spontaneous pneumothorax common in the young has the same underlying connective tissue disorder that is seen in the Marfan syndrome. Wright *et al* found high metacarpal indices in six of 12 young patients with spontaneous pneumothorax, who did not meet the criteria for the Marfan syndrome.[12]

The results of HLA analysis might show the possibility of the presence of a disease susceptibility gene linked to HLA, non-randomly segregating with a particular HLA haplotype. Sharpe *et al* reported a relationship between the HLA haplotype A2,B40 and the occurrence of spontaneous pneumothorax by HLA analysis of a large family.[13] Our limited data on HLA make up permit no useful conclusions.

Address for reprint requests: Dr Yukihiko Sugiyama, Third Department of Internal Medicine, Faculty of Medicine, University of Tokyo, 7-3-1 Hongo, Bunkyo-ku, Tokyo 113, Japan.

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