is also atypical for PAN. It is possible that the delay in therapy because of the patient’s noncompliance contributed to the poor outcome.

The cause of pulmonary hemorrhage in this patient is unclear. Because a lung biopsy was not done, it is unclear whether pulmonary hemorrhage in our patient was because of alveolar capillary injury or a capillaritis. The later seems unlikely unless the patient has an overlap syndrome. Though cocaine has been reported to cause pulmonary hemorrhage in association with a pulmonary-renal syndrome, it is unlikely to be the cause of pulmonary hemorrhage in this patient as pulmonary hemorrhage occurred 8 days after his admission to the hospital. Infection was also excluded as a cause of hemorrhage by multiple negative blood and sputum culture results. Finally, our patient had recurrent bouts of malignant hypertension despite being administered maximum doses of four antihypertensive drugs. Recently, a case of pulmonary hemorrhage was reported in association with malignant hypertension. It is possible that this may have played role in the pathogenesis of pulmonary hemorrhage in this patient. This report suggested that malignant hypertension can cause alveolar capillary injury leading to hemorrhage.

REFERENCES

Primary Spontaneous Pneumothorax in Two Siblings Suggests Autosomal Recessive Inheritance*

Pasi A. Koististo, MD, PhD; and Aki Mustonen, MD, PhD

We report on a sister and brother with recurrent spontaneous pneumothorax without underlying connective tissue disease and without other affected relatives. The occurrence of spontaneous pneumothorax in two siblings from a large Finnish family raises the possibility of autosomal recessive inheritance of this disorder in some patients.

(CHEST 2001; 119:1610–1612)

Key words: autosomal recessive; inheritance; spontaneous pneumothorax

Primary spontaneous pneumothorax is a rare disorder mostly occurring in male subjects. Smoking, height (especially in male subjects), and family history are the best known risk factors for primary spontaneous pneumothorax. Most of the cases are sporadic, but it is also well known that primary spontaneous pneumothorax may be inherited. Familial pneumothorax may be a complication of various inherited disorders, such as α1-antitrypsin deficiency, Marfan’s syndrome, and the Ehlers-Danlos syndrome, but familial cases without evidence of connective tissue disease do occur.2–4

Primary spontaneous pneumothorax is genetically heterogeneous, and articles published so far suggest autosomal dominant inheritance with incomplete penetrance.

*From the Department of Clinical Genetics, Tampere University Hospital, Tampere, Finland. Manuscript received April 25, 2000; revision accepted November 9, 2000.
Correspondence to: Pasi A. Koististo, MD, PhD, Department of Clinical Genetics, Tampere University Hospital, P.O. Box 2000, FIN-33521 Tampere, Finland; e-mail: blpako@uta.fi

Selected Reports
polygenic, or X-linked recessive inheritance. Only one report suggested autosomal recessive inheritance.

Here, we report an observation of the occurrence of primary spontaneous pneumothorax with a putative autosomal recessive transmission in a large Finnish family.

CASE REPORT

Patient 1

The index patient is the second child of healthy, nonconsanguineous parents. She is 25 years old and a nonsmoker. She has experienced attacks of diarrhea from the age of 14 years; generally, the diarrhea has been related to eating disorders and anorexia nervosa. She has been treated for this problem. Results of gastroscopy and colonoscopy have remained normal. The age of menarche was 15 years, and no relationship between primary spontaneous pneumothorax and menstruation was present. She has not had pelvic pain, and findings of gynecologic examinations, including transvaginal ultrasound, have remained normal. From the age of 17 years, she has experienced spontaneous pneumothorax three times in the left and right lungs. A left-sided pneumothorax developed when she was 19 years, and because of slow resolution, reoperation was needed 2 years later when the apical segment of the left lung was removed. The histologic diagnosis of the removed lung tissue was inconclusive. At the age of 21 years, when she was 161 cm tall and weighed 62 kg, the patient was examined by a clinical geneticist. No minor anomalies were seen. Joints were of normal mobility. The thorax was symmetrical, and scoliosis was not noticed. Striae or varicose veins were not seen. The teeth and palate were normal, and her skin was not hyperelastic.

Patient 2

Patient 2 is a younger brother of the index patient. He is 21 years old and has been smoking for 4 years. His development was normal. He has not had GI or neurologic symptoms. At the age of 17 years, a spontaneous pneumothorax of the left lung was treated with drainage. Two years later, a pneumothorax occurred on the left lung after minor trauma. Spontaneous remission followed. At the age of 20 years, a spontaneous pneumothorax developed on the same side. Because the resolution was slow, thoracoscopic bullectomy was needed. A pneumothorax recurred 2 months later and was again treated operatively (thoracoscopic pleural abrasion and pleurodesis). The blood α₁-antitrypsin level was within reference values (1.6 g/L), and the phenotype was MM. High-resolution CT of the lungs showed a few small subpleural bullae. Ultrasound findings of the patient’s upper abdomen were normal. Echocardiography showed evidence of a bicuspid aortic valve and mild regurgitation, but no aortic root dilatation was seen. His ophthalmologic status was also normal. At the age of 21 years, when he was 183 cm tall and weighed 62 kg, the patient was seen by the clinical geneticist. No minor anomalies were seen. Joints were of normal mobility. The thorax was symmetrical, and scoliosis was not noticed. Striae or varicose veins were not seen. His teeth and palate were normal, and his skin was not hyperelastic.

Discussion

Since the first description of primary spontaneous pneumothorax >50 years ago, the best-characterized mode of inheritance is autosomal dominant with reduced penetrance in female subjects. However, one report suggested autosomal recessive inheritance in primary spontaneous pneumothorax. Gibson described three sisters with repeated pneumothoraces beginning at 28 years, 32 years, and 37 years of age, respectively.

We report here two siblings with spontaneous pneumothorax with probable autosomal recessive inheritance. Bullae were seen in both patients, and pneumothorax episodes most likely resulted from a rupture of a bulla. Because bullae and spontaneous pneumothorax are known

![Figure 1](http://publications.chestnet.org/pdfaccess.ashx?url=/data/journals/chest/21962/ on 06/17/2017)
complications in the Marfan's syndrome and Ehlers-Danlos syndrome, as well as in α1-antitrypsin deficiency, the possibility of these connective tissue diseases was carefully excluded. In addition, because of the slightly atypical clinical course of primary spontaneous pneumothorax in patient 1, several nongenetic etiologies of primary spontaneous pneumothorax, such as endometriosis, were excluded.

None of their many relatives had a history of pneumothorax or any symptoms or signs of pulmonary or connective tissue disease. In addition, chest radiographs of the affected siblings' parents showed no indication of any abnormality, and the patients were not taller than their first-degree relatives, which further strengthens the likelihood of autosomal recessive inheritance in this family (Fig 1). Determination of human leukocyte antigen haplotypes of the patients could have been of some importance in evaluating the inheritance of primary spontaneous pneumothorax in the family because an association between human leukocyte antigen haplotype A2B40 and primary spontaneous pneumothorax has been demonstrated. However, the patients refused genetic testing.

The experiences of the family reported here strongly suggest autosomal recessive inheritance of primary spontaneous pneumothorax. X-linked recessive transmission is ruled out because of an affected female family member. Excluding autosomal dominant inheritance is more difficult. However, recurrent pneumothorax rarely remains undiagnosed, and because neither the parents nor any other relatives of the patients have had primary spontaneous pneumothorax, autosomal recessive is the most likely mode of inheritance. Consistent with the possibility of autosomal recessive inheritance is that consanguinity is common in the area from which the patients and their ancestors originate. The grandparent of the patients were traced back to the late 19th century. Until this period, they were unrelated, but a common ancestor is highly likely because this sparsely populated rural area was gradually inhabited in the 17th century by a small number of people.

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**Lethal Hemoptysis Caused by Biopsy Injury of an Abnormal Bronchial Artery**

**Helmut Maxeiner, MD**

A 62-year-old man with a long history of lung disease developed atelectasis of the right middle lung lobe, caused by a protrusion in the wall of the middle lobe bronchus. A biopsy was performed in the suspicious region. This was immediately followed by massive arterial bleeding into the airways and complicated by cardiac arrest soon after. The bleeding could not be controlled by nonsurgical treatment; the patient died 24 h after the complication because of pulmonary insufficiency. Autopsy revealed the bleeding to have been caused by a biopsy injury of a bronchial artery that had run superficially in the bronchial mucosa and had produced the intrabronchial protrusion. Several other abnormal intrabronchial arteries were found peripherally in this lung.

**Key words:** bronchial arteries; bronchoscopic biopsy; bronchoscopy; hemoptysis; lethal complication

Hemoptysis may result from a variety of pulmonary, cardiovascular, or hematologic disorders. Massive hemoptysis is not frequent, but it has a high mortality and can be so rapid that it leaves no opportunity for meaningful intervention. Bleeding mostly arises from the bronchial artery plexus, which surrounds the airways and is altered in various lung diseases. There are some reports of massive hemoptysis from several types of abnormal pulmonary blood vessels; in rare cases, even the puncture of an anatomically regular, small artery can result in lethal hemoptysis. In most of the reported cases, an anamnesis of previous repeated hemoptysis is present. We investigated a case in which, to the best of our knowledge, the patient's first bronchial hemorrhage occurred in the course of a bronchoscopic biopsy that unfortunately violated a pathologic bronchial artery. Because of the rarity of such events and the considerable risk of confusing intra-bronchial abnormal arteries with tumors, a detailed presentation of our findings might be helpful for the prevention of such hazards.

**Case Report**

**History**

A 62-year-old man had a long history of severe bronchiectasis and bronchitis, complicated by frequent pneumonias. Because of

*From the Institute of Legal Medicine, Medical School Benjamin Franklin, Free University, Berlin, Germany. Manuscript received May 31, 2000; revision accepted November 14, 2000. Correspondence to: Helmut Maxeiner, MD, Institute of Legal Medicine, Medical School Benjamin Franklin, Free University, D-14195 Berlin, Hittorfstrasse 18, Berlin, Germany; e-mail: maxrne@zedat.fu-berlin.de*