

Cystic lung in Marfan's syndrome

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ABSTRACT Various pulmonary problems have been described in Marfan's syndrome. Unusual cystic lung changes in a young girl with Marfan's syndrome are described.

Marfan's syndrome usually presents with skeletal, cardiovascular, and ocular abnormalities.¹ The basic pathological abnormality in this disorder affects the supportive elements of the connective tissue. Various pulmonary abnormalities have been described. We report widespread cystic changes in a child with Marfan's syndrome.

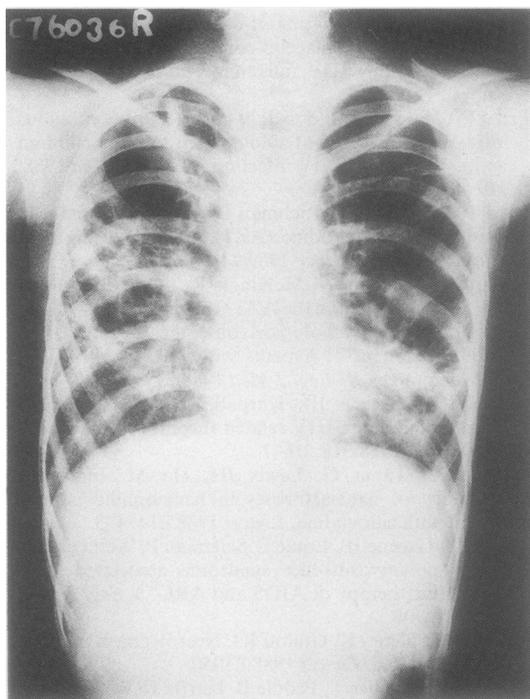
Case report

A 12 year old previously fit Indian girl was referred with intermittent low grade fever of nine months' duration, a non-productive cough, and increasing breathlessness. There was no history of contact with tuberculosis.

The patient was thin with a long face and tapering extremities. Her height was 146 cm and arm span 151 cm, and the upper segment:lower segment ratio 0.84. She had pes planus, a high arched palate, and pectus carinatum but no scoliosis. She had the "wrist sign" and "thumb sign," two skeletal signs associated with Marfan's syndrome.² On examination of the chest there was amphoric bronchial breathing in the left mid zone and scattered crepitations in all zones. No cardiac abnormality was detected clinically. Detailed ophthalmological examination, including slit lamp examination, showed nothing abnormal.

Both parents were tall with a thin habitus; the father had the thumb sign and the wrist sign. No other abnormality was detected clinically.

The blood count, results of urine analysis, the erythrocyte sedimentation rate, and the electrocardiogram were normal. Tuberculin testing gave a positive result. The chest radiograph showed bilateral, thin walled cysts of varying sizes; there was no pleural reaction and no hilar lymphadenopathy (figure). A sputum smear and three cultures were negative for acid fast bacilli and other bacteria. The child was prescribed broad spectrum antibiotics; antituberculous treatment was subsequently added empirically. She became afebrile and less breathless with this combination and antibiotics were stopped after six weeks. Four weeks later she became febrile again despite continuing



Chest radiograph showing multiple cysts and bullae in both lung fields.

antituberculous treatment but responded to another course of broad spectrum antibiotics. Chest radiographs showed no resolution. Other investigations showed a metacarpal index of 10 on radiographs of both hands (in the Marfan range)³ and aortic root dilatation of 26 mm on an echocardiogram. The serum immunoglobulin profile, the result of the sweat test, and the α_1 antitrypsin concentration were normal and a urine test for homocystinuria gave a negative result. Arterial blood gas analysis showed persistent hypoxaemia (oxygen tension 7.3-10.5 kPa). She died after developing a spontaneous pneumothorax on the right side. Necropsy was not permitted.

Discussion

Pyeritz and McKusick² recommended that at least two of four criteria—that is, skeletal, ocular, cardiovascular, and familial—should be present for a diagnosis of Marfan's syndrome to be made. The diagnosis was established in our

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Accepted 1 August 1989

patient on the basis of skeletal, cardiovascular (aortic root dilatation is the earliest cardiovascular change in Marfan's syndrome³), and familial features (skeletal changes in the father).

Various pulmonary manifestations have been described in Marfan's syndrome, including interstitial parenchymal disease and honeycombing,⁴ diffuse and apical bullous emphysema,⁵ congenital malformations of the bronchus, bronchiectasis,⁶ and spontaneous recurrent pneumothorax.⁷ Lung cysts and bullae are uncommon, however. In a recent retrospective review of 100 patients with Marfan's syndrome⁸ only five had lung cysts and all were over the age of 20. Diffuse cystic changes on the chest radiograph have been described in two adults.^{4,7} Pulmonary changes have been recorded in only 10 children and only two had cysts or emphysematous bullae, diagnosed at necropsy in both cases. The present case is the first with extensive cyst changes in both lungs at such a young age to be diagnosed during life.

Pneumonia and frequent lower respiratory tract infections occur in Marfan's syndrome.^{7,8} The positive response to the tuberculin test and prolonged respiratory symptoms suggested tuberculosis in our patient but repeated negative smears and cultures for acid fast bacilli and the recurrence of fever despite antituberculous treatment make the diagnosis unlikely. The absence of hilar gland enlargement in a child in the presence of such extensive cystic disease also makes tuberculosis unlikely, though this is impossible to exclude.

Whether the cystic changes are congenital in origin or whether they arise from premature degeneration of pulmonary parenchyma is not clear. The precise cause of the gross pulmonary disease remains uncertain; it may be a con-

sequence of abnormal collagen, causing flaccidity of the walls of the terminal bronchioles during expiration and thus obstruction and air trapping^{4,9} (abiotrophy of connective tissue⁶). It is important to recognise this complication of Marfan's syndrome as it may lead to spontaneous pneumothorax.

References

- 1 McKusick VA. *Heritable disorders of connective tissue*. 4th ed. St Louis: Mosby, 1972:61-223.
- 2 Pyeritz RE, McKusick VA. The Marfan syndrome: diagnosis and management. *N Engl J Med* 1979;**300**:772-7.
- 3 Come PC, Bulkley BH, McKusick VA, *et al*. Echocardiographic recognition of silent aortic root dilatation in Marfan syndrome. *Chest* 1977;**72**:789-92.
- 4 Lipton RA, Greenwald RA, Seriff NS. Pneumothorax and bilateral honeycombed lung in Marfan syndrome. *Am Rev Respir Dis* 1971;**104**:924-8.
- 5 Turner JAM, Stanley NN. Fragile lung in the Marfan syndrome. *Thorax* 1976;**31**:771-5.
- 6 Bolande RP, Tucker AS. Pulmonary emphysema and other cardiorespiratory lesions as part of the Marfan abiotrophy. *Pediatrics* 1964;**33**:356-66.
- 7 Dwyer EM, Troncale F. Spontaneous pneumothorax and pulmonary disease in the Marfan syndrome. Report of two cases and review of the literature. *Ann Intern Med* 1965;**62**:1285-92.
- 8 Wood JR, Bellamy D, Child AH, Citron KM. Pulmonary disease in patients with Marfan syndrome. *Thorax* 1984;**39**:780-4.
- 9 Pyeritz RE. Connective tissue in lung: lessons from the Marfan syndrome [editorial]. *Ann Intern Med* 1985;**103**:289-90.