

PostScript

LETTER

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Familial spontaneous pneumothorax

We would like to report a series of four newborns in an extended family who have had spontaneous pneumothorax soon after birth without any obvious cause (fig 1). There is no history of pneumothorax in anyone else in the family.

Case 1

A female infant was born in good condition at term by emergency caesarian section after a

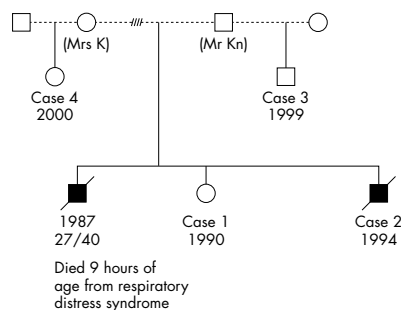


Figure 1 Genetic map showing the positions of the neonates with spontaneous pneumothorax in the extended family.

normal pregnancy. She had a spontaneous right sided tension pneumothorax within a few minutes of birth. The pneumothorax was drained. There was no evidence of surfactant deficiency on any of the initial chest radiographs. After discharge she went on to have three other episodes of spontaneous pneumothorax until 3 years of age. At 11 years of age she is now clinically well and does not show any signs of collagen disorders. Investigations have excluded cystic fibrosis, homocystinuria, and α_1 antitrypsin deficiency.

Case 2

A male infant was born at 36⁺ weeks gestation by elective caesarian section after a normal pregnancy. He was born in good condition but had a spontaneous tension pneumothorax on the right side within a few minutes of birth. His chest radiograph did not show any signs of surfactant deficiency or infection. He was ventilated, and the pneumothorax was drained. However, he became progressively hypoxic and died within a few hours of age. Post mortem examination showed normal lung size with histological evidence of hyaline membrane disease.

Case 3

A term male infant was delivered in good condition by elective caesarian section because of maternal orthopaedic problems. However, he went into acute respiratory failure because of right sided pneumothorax within a few hours of birth. There was no evidence of surfactant deficiency on the chest radiographs. He improved with drainage of the pneumothorax and ventilation. α_1 Antitrypsin has been excluded. At 2 years of age, he is well and does not have increased skin elasticity and joint hypermobility.

Case 4

A female infant was born at term in good condition. She had a spontaneous non-tension pneumothorax within an hour of birth. This resolved spontaneously over the next few days. There was no evidence of any parenchymal lung disease on the chest radiograph. On follow up she is well at one year.

Discussion

Familial spontaneous pneumothorax was first described by Farber in 1921.¹ Since then it has been described in young adults and teenagers

in extended families.^{2,3} The cause is thought to be rupture of apical bullae.^{2,3}

Recognised causes of familial spontaneous pneumothorax include genetic disorders, such as cystic fibrosis, homocystinuria, and α_1 antitrypsin, and collagen disorders, such as Ehlers-Danlos syndrome.⁴ Spontaneous pneumothorax in newborns is usually a complication of ventilation or parenchymal lung diseases such as surfactant deficiency and meconium aspiration.⁵

There have been suggestions of mendelian inheritance of this trait—autosomal dominant with variable penetrance⁶ and X linked recessive.⁷ However, all these reports are in adults in extended families.

Because of the absence of clinical, laboratory, or radiological evidence of a primary cause for these pneumothoraces, we suggest that it is a familial trait transmitted as autosomal dominant with variable penetrance (which should explain the absence of any family history). In that case, both Mr Kn and Mrs K have to be non-penetrant carriers.

This series highlights the importance of close postnatal monitoring of babies with a family history of unexplained neonatal pneumothorax. This also has obvious implications for antenatal genetic counselling.

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