Familial Spontaneous Pneumothorax in Neonates

Soraisham Amuchou Singh and Harish Amin

Division of Neonatology, Department of Pediatrics, Foothills Medical Centre, University of Calgary, Alberta, Canada

Abstract. Spontaneous pneumothorax is a recognised cause of respiratory distress in the neonatal period. Spontaneous pneumothorax occurring during the neonatal period in siblings within a family is rare. We report a case of spontaneous pneumothorax occurring in two siblings in the neonatal period. [Indian J Pediatr 2005; 72 (5) : 445-447]
E-mail: amuchou@yahoo.com

Key words: Spontaneous pneumothorax; Familial; Neonate; Newborn

Spontaneous pneumothorax is a recognised cause of respiratory distress in newborn infants. Familial spontaneous pneumothorax during the neonatal period is an uncommon entity and rarely reported. To the best of our knowledge, there are only three case reports of pneumothorax occurring in more than one neonate in the same family.1,2,3 We report the occurrence of spontaneous pneumothorax in two siblings during the neonatal period.

CASE REPORTS

Case 1
A 3010 gm female infant was born at 40 weeks gestation by spontaneous vaginal delivery to a 33 years old Gravida 2 Para 1 woman after an uneventful pregnancy. Prenatal course was unremarkable except for vaginal colonisation with group B streptococcus. Prophylactic intrapartum antibiotics were administered. The duration of labour was 4 hours and membranes ruptured just prior to delivery. Apgar scores were 7 and 8 at 1 and 5 minutes respectively. Initial resuscitation included oro-pharyngeal suction and free flow oxygen. Umbilical cord arterial pH was 7.26 with base deficit of – 4. There was no maternal fever or evidence of chorioamnionitis. Shortly after delivery, baby developed grunting and cyanosis. She required supplemental oxygen via face mask with resolution of cyanosis.

Physical examination at admission revealed the following: heart rate 156 beats/min, blood pressure 64/44 mmHg, and respiratory rate 68 breaths/minute. Breath sounds were decreased with no rales or wheezes. No chest retractions were noted. Rest of the systemic examination was normal.

An arterial blood gas measurement in 40% ambient oxygen revealed a pH of 7.41, PaCO2 of 32 mmHg and PaO2 of 57 mmHg. The chest radiograph revealed bilateral small anterior pneumothoraces with normal pulmonary vasculature and normal cardiac silhouette (Figs. 1 & 2). Complete blood count was normal. She was managed with oxygen via an oxyhood. Respiratory distress
resolved within 12 hours. She did not require needle thoracocentesis or chest tube placement. Breast feeding was started at 12 hours of age. She was also treated with ampicillin and gentamicin for 48 hours. Blood cultures were sterile. Repeat chest radiograph done after 24 hours showed resolution of pneumothoraces.

Case 2 (Sibling 1)

A 41-week gestation male infant was born by forceps delivery after induction of labor for postmaturity. Fetal tachycardia and thin meconium stained fluid was noticed during second stage of labor. Birth weight was 3815 gm. The infant was vigorous at birth and the Apgar scores were 8 and 9 at 1 and 5 minute respectively. No endotracheal or oral suctioning was required at birth.

Soon after birth, he started grunting and was tachypneic. Chest radiograph showed bilateral small pneumothoraces. He was treated with oxygen by oxyhood. Respiratory distress resolved within 4 hours, following which breast feeding was commenced. At 2 years, he is a healthy boy and has not had any recurrences of pneumothorax.

DISCUSSION

We describe the occurrence of spontaneous pneumothorax in the newborn period in two siblings of a family. Spontaneous pneumothorax is a recognised cause of respiratory distress in newborn period. The estimated incidence of spontaneous pneumothorax varies from 0.3% to 1.3% based on clinical symptoms or on radiological findings respectively.

In the newborn period, pneumothorax may occur spontaneously (idiopathic) or secondary to underlying lung diseases such as respiratory distress syndrome, meconium aspiration syndrome, vigorous resuscitation, positive pressure ventilation, pulmonary hypoplasia, pneumonia and congenital pulmonary cystic malformations. Spontaneous pneumothorax at birth may result from rupture of alveoli secondary to high pressure needed to expand previously uninflated lungs or from uneven distribution of inflating pressures among groups of alveoli.

Familial spontaneous pneumothorax (FSP) was first described by Faber in 1921. FSP is extremely rare in neonates. To the best of our knowledge, only three case reports are available in the literature that involves more than one newborn in the family. One report describes a family in which 5 persons in three generations suffered spontaneous pneumothorax, four of them had spontaneous pneumothorax during the neonatal period. No etiology was identified, although transient tachypnea of newborn (TTN) or unspecified neonatal respiratory problems were diagnosed. In this family, most pneumothoraces occurred in female infants on the maternal side.

Bagchi et al reported a series of four neonates in an extended family that have had spontaneous pneumothorax soon after birth without any obvious cause. Kugelman et al describe neonatal pneumothorax associated with transient tachypnea of newborn in siblings of two families.

FSP has also been reported in adults and adolescents with male predominance. Wilson and Ellsworth reviewed the literature and identified 57 familial cases of spontaneous pneumothorax in 21 families. The affected parents and children were found in 13 families. Typically spontaneous pneumothorax in this population was due to spontaneous rupture of apical bulla. Other causes of familial spontaneous pneumothorax include bronchial asthma, tuberculosis, Marfan's syndrome, genetic disorders such as cystic fibrosis, homocystinuria, alpha1 antitrypsin deficiency and collagen vascular disorders like Ehlers-Danlos syndrome.

Attempts have been made to identify the mode of inheritance of FSP. Morrison et al and Bagchi et al have suggested an autosomal dominant inheritance with variable penetrance whereas Abolnic et al suggested an X-linked recessive pattern. Sharpe et al suggested possible linkage of HLA-A2, B40 haplotypes with spontaneous pneumothorax. Lenler-Petersen et al in their studies did not identify any relationship between familial spontaneous pneumothorax and certain HLA haplotypes.

Administration of 100% oxygen (nitrogen washout therapy) to infants with pneumothorax accelerates the resolution of air leaks and shortens the duration of extra pulmonary air collection from 2 days to 8-12 hours. The majority of the infants with spontaneous pneumothorax are managed conservatively. Only 6/80 (7.5%) symptomatic term infants with spontaneous pneumothorax require chest tube insertion or needle thoracocentesis.

CONCLUSION

We suggest that infants born to families with a history of spontaneous pneumothorax in the neonatal period should be closely observed for early signs of respiratory distress, and be evaluated and treated promptly.

REFERENCES

2. Bagchi I, Nycz J. Familial spontaneous pneumothorax. Arch Dis Child Fetal Neuropulmonol. 2002; 87 : F70