Pulmonary Hypoplasia

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
265430

Mode of Inheritance
? Autosomal recessive

Description
Pulmonary hypoplasia, an extremely rare condition, incompatible with life, is the underdevelopment of the lung. Affected individuals develop respiratory distress from birth. The lungs are very stiff and there is little chest movement even with artificial ventilation. Hypoxia, hypercarbia, and a metabolic acidosis can develop. Pulmonary hypoplasia occurs commonly in association with congenital diaphragmatic hernia, oligohydramnios (mostly related to renal dysfunction), skeletal dysplasias, fetal hydrops, malformations of the CNS, and neuromuscular diseases. It results from either absence of fluid to fill the lungs in development, or from absence of space to grow. Primary isolated bilateral pulmonary hypoplasia is rare and familial occurrence exceptional.

Primary pulmonary hypoplasia as a result of congenital acinar dysplasia (CAD) is exceedingly rare and is diagnosed by exclusion of all known etiologies of secondary pulmonary hypoplasia. In CAD, the acini (the respiratory bronchioles, alveolar ductus, and alveoli) fail to develop. The lungs at term will have the appearance of the pseudoglandular phase of 16 weeks gestation.

Epidemiology in the Arab World

Palestine
Rosenak et al. (1991) reported a term amelic female infant born to an apparently non-consanguineous Moslem couple. The parents had four other normal children. Afterwards, in 2 subsequent pregnancies, 2 amelic fetuses were diagnosed by transabdominal ultrasonography in the 18th and 12th week of gestation. Pregnancies were terminated and on autopsy both amelic fetuses had severe lung hypoplasia and aplasia of the peripheral pulmonary vessels. The first fetus also had apparently low-set ears and micrognathia, whereas the last had hydrocephaly and left cleft lip beside the lung hypoplasia and aberrant pulmonary artery.

Saudi Arabia
Al-Hathal et al. (1998) conducted a retrospective analysis of all neonates with respiratory distress at birth due to congenital diaphragmatic hernia (CDH) who were treated with neither extracorporeal membrane oxygenation nor nitric oxide being used. Analysis included all neonates with CDH and respiratory distress at birth who were treated between August 1, 1992 through March 1, 1997. There were 21 patients, 11 male and 10 female. There were 17 full-term and 4 premature infants and surgery was performed in 18 infants. Al-Hathal et al. (1998) reported that one infant died during preoperative stabilization from severe pulmonary hypoplasia and pulmonary hypertension and one infant died postoperatively from the same conditions.

Al-Senan et al. (2003) reported a family with one proven case and two suspected cases of isolated congenital acinar dysplasia. The index case was a female infant born at 37 weeks gestation to healthy and unrelated parents. Antenatal ultrasound (US) examination in the 32nd week of gestation showed mild oligohydramnios with no congenital malformations. There was no history of premature rupture of membrane or intrapartum fetal distress. The mother had one abortion, one intrauterine fetal death, and 2 infantile deaths. The remaining siblings are alive and well. Of
the 2 infantile deaths, the first infant was a female, product of a full-term normal delivery, which developed severe respiratory failure of unknown etiology shortly after birth, and died at 2 months of age. The 2nd infant, also a female, developed severe respiratory failure soon after birth and died at 75 days on mechanical ventilator. At 6 hours of age, the index infant developed progressively severe respiratory distress with hypoxemia and hypercapnia, which required intubation and mechanical ventilation. The chest x-ray showed reduced lung volume and increased pulmonary vascularity. Gradually, the condition of the infant worsened that requires higher ventilator settings. The infant did not tolerate trials of weaning from mechanical ventilation, and each time showed deterioration despite the administration of steroids to facilitate weaning. Microscopically, the lung biopsy showed pulmonary hypoplasia with arrest of lung growth at the acinar stage, and the electron microscopy showed plenty of lamellar bodies in type II pneumocytes. Shortly thereafter, at the age of 3 months, the infant expired. Al-Senan et al. (2003) reviewed the world cases with primary pulmonary hypoplasia as a result of congenital acinar dysplasia and suggested that these observations may raise the possibility of X-linked dominant type of inheritance for this disease.

United Arab Emirates
Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January 1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Sporadic conditions accounted for 26% of the cases. In their study, Al Talabani et al. (1998) observed two cases of pulmonary hypoplasia in families from the United Arab Emirates. Recurrence was not reported in other members of the families. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities in the whole United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi, the capital of United Arab Emirates.

References

Contributors
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