Emphysema, Congenital Lobar

Alternative Names
CLE
Congenital Pulmonary Emphysema
Emphysema, Localized Congenital
Lobar Emphysema, Infantile
Lobar Tension Emphysema in Infancy

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations of the respiratory system

Incidence per 100,000 Live Births
Unknown

OMIM Number
130710

Mode of Inheritance
Autosomal dominant

Gene Map Locus
N/A

Description
Congenital Lobar Emphysema (CLE) is a serious condition of respiratory distress in neonates, in which a weakened or absent bronchial cartilage leads to bronchial narrowing. Thus, air is able to enter the lungs, but is unable to exit from it. This leads to hyperinflation of one of the pulmonary lobes, compression of the remaining lung, and displacement of the mediastinum. Typical symptoms in the neonate include wheezing, shortness of breath, and cyanosis. The condition usually involves one of the pulmonary lobes, but could involve more than one too. The condition is most common in neonates, but may appear in infants less than six months old too. Interestingly, the severity of the symptoms varies among individuals, and thus, in some affected people, the condition may never become apparent. For some reason, the condition is more common in boys than in girls and in whites than amongst non-whites.

An infant with respiratory distress is diagnosed with CLE when X-rays show over-inflation of at least one pulmonary lobe and a blocked air passage. Patients with the milder form of the disease do not require any treatment. Severe forms, however, require surgical correction, also known as lobectomy, in which the affected lobe of the lung is removed. Newer techniques, such as ventilation/perfusion scintigraphy, are now being used to manage affected patients without resorting to surgery.

Molecular Genetics
Occurrence of CLE is more often than not, sporadic in nature. However, a few cases have been reported where genetic transmission of the condition has been noted. Affected siblings, and at least one pair of affected mother and daughter, and affected father and son have been described, providing evidence for inherited factors in the etiology of CLE.

Epidemiology in the Arab World
Kuwait
Abdul Majid and Perianayagam (1990) reported nine cases of congenital lobar emphysema. In seven of these cases, the left upper lobe was involved, while in two it was the right middle lobe. Surgery was performed in all patients and was life saving in all cases. In only one case, delayed surgery led to the infant's death.

Ayed and Owayed (2003) conducted a retrospective study on 47 infants with congenital lung diseases. The mean age at the time of diagnosis was 90 days (range, 7 days to 11 months). Thirty-four patients were male (72%). Congenital lobar emphysema was seen in 26 patients.

Oman
Thakral et al. (2001) conducted a retrospective study on 21 patients (13 males, 8 females) diagnosed with congenital lobar emphysema during the period of 1988 to 1999 (11 years) in the Royal

Hospital, Oman with exclusion of those with lobar hyperinflation secondary to intraluminal or extrinsic obstruction. All cases were non-white Arabs. The majority (81%) presented within the first six weeks after birth, eight cases (38%) presented within the first 24 hours, seven (33.5%) presented at the age of one week to six weeks, and two (9.5%) had onset of symptoms within the first week of life. Two patients (9.5%) presented between six weeks to six months of age and the last two presented (9.5%) after the age of six months. The presenting symptoms varied among the patients according to severity, but all had tachynea, 18 cases (86%) had respiratory distress, fever was present in 9 patients (43%), cough in 8 patients (38%), and wheezing and cyanosis was present in 4 cases (19%) each. One baby was misdiagnosed in a peripheral hospital as a case of pneumothorax and his condition deteriorated after the insertion of an intercostal drain. Only nine cases out of the 21 were mildly symptomatic and did not need oxygen supplementation to maintain good oxygen saturation. The diagnosis was made by chest X-rays, which showed the involved lobe to be hyperlucent with few pulmonary markings, herniation of the hyperinflated lobe into the opposite side with mediastinal shift and collapse of the adjacent lobe, and the diaphragm was either normal or displaced downwards. CT scan of the chest confirmed the above findings as it showed stretched attenuated spread-out vessels in the emphysematous lobe, but it was difficult in two patients to differentiate between congenital lobar emphysema and compensatory emphysema. Other investigations done were bronchoscopy in seven patients to exclude obstruction (mucus plug, granuloma, radiolucent foreign body, or narrowing of the bronchus), and barium swallow and angiography (two patients) to rule out external vascular compression of the bronchus or trachea. It was found that the upper lobe disease was more severe than the middle lobe one as nine out of the 13 cases with left upper lobe disease were severe and four were mild, the one with right upper lobe disease was severe and two out of the seven with right middle lobe disease were severe and five were mild. Conservative treatment was possible in only three patients who had mild distress with no fever, and it failed in four cases with distress and fever. These patients then underwent lobectomy along with the remaining 14 patients. There was no mortality or significant complications after the operation except for a lower lobe bronchus injury in one patient.

Saudi Arabia

Al-Salem et al. (1990) reported three cases of congenital lobar emphysema that were treated at King Fahd Hospital in Saudi Arabia over a period of three years. All the cases were boys and their age ranged from two and a half to eight months. All presented with recurrent attacks of chest infection, which started from the neonatal period. There were no associated or cardiac anomalies. Attempts to treat the patients using antibiotics, chest physiotherapy, bronchodilators, and oxygen therapy were all unsuccessful. Meanwhile, chest X-ray made a clear diagnosis of congenital lobar emphysema possible. In two patients, the left upper lobe was affected, while in the remaining patient, it was the right upper lobe. Two of the patients underwent lobectomy, while surgery was refused in the case of the third patient. The surgeons were careful with administering anesthesia, and made sure that nitrous oxide was added to the mixture only after excision of the emphysematous lobe, in order to prevent rapid inflation of the emphysematous lobe resulting in a cardiac arrest. Both patients made a rapid recovery without any complications. Chest X-rays showed that the remaining lobes had re-expanded to fill the space created by the lobectomy.

References

Abdul Majid OA, Perianayagam JW. Congenital lobar emphysema. Kuwait Med J. 1990; 24(1):74-7. [Abs]

Al-Salem AH, Adu-Gyamfi Y, Grant CS. Congenital lobar emphysema. Can J Anaesth. 1990; 37(3):377-9. PMID: 2322975 [FT]

Ayed AK, Owayed A. Pulmonary resection in infants for congenital pulmonary malformation. Chest. 2003; 124(1):98-101. PMID: 12853509

Thakral CL, Maji DC, Sajwani MJ. Congenital lobar emphysema: experience with 21 cases. Pediatr Surg Int. 2001; 17(2-3):88-91. PMID: 11315310.

Related CTGA Records

N/A

External Links

http://www.emedicine.com/radio/topic188.htm

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=1928

http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Emphysema%2C%20Congenital%20Lobar

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