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Case Report

Unique association of congenital lobar emphysema with ventriculomegaly and encephalocele

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Abstract

Congenital lobar emphysema (CLE) is a rare developmental disorder characterized by over distension of pulmonary lobes or segments. Often CLE presents a diagnostic and therapeutic dilemma. Here, we report a case of incidentally found CLE at fetal autopsy of terminated pregnancy due to ventriculomegaly and occipital localized encephalocele detected on sonography in the second trimester. A 41- year-old woman at gestational age of 20 weeks 4 days revealed with ventriculomegaly and occipital localized encephalocele in obstetric sonography. In fetal autopsy beside meningocele, CLE was detected. CLE is a rare malformation characterized by over-distension of pulmonary lobes. The emphysematous lungs of infants younger than six months can result from foreign body aspiration. Major differential diagnosis of hyperechogenic lung lesions include; congenital cystic adenomatoid malformation, upper airway obstruction, pulmonary sequestration, bronchogenic cysts and CLE. The exact diagnosis of CLE is based on combination of clinical, radiologic and histopathologic examinations. Antenatal remarkable findings highlight the significance of postnatal follow-up whether the lesion echogenicity differs in size, even gets disappeared in the prenatal course. In English literature, this case is the first CLE associated with meningocele.

Key words:

Congenital Lobar Emphysema, chranioschisis, neural tube defect, hydrocephalus

Introduction

Congenital lobar emphysema (CLE) is a rare developmental disorder characterized by over distension of pulmonary lobes or segments. Its prevalence is 1 in 20.000-30.000 live births, and the incidence is estimated to be 1 in 70.000 to 1 in 90.000 [1]. Male gender is affected more than female gender. Most affected lobes are left upper, right middle and right upper lobe, relatively. Prenatal diagnosis of CLE is extremely rare [2]. Often CLE causes a diagnostic and therapeutic dilemma. Suggested treatment is usually lobectomy for CLE. However, with the use of ventilation/perfusion scintigraphy, cases may be managed non-surgically while maintaining observation of regional anatomy and function [3]. Here, we report a case of incidentally found CLE at fetal autopsy of terminated pregnancy due to ventriculomegaly and occipital encephalocele detected on obstetric sonography in the second trimester. The final diagnosis was made by fetal autopsy.

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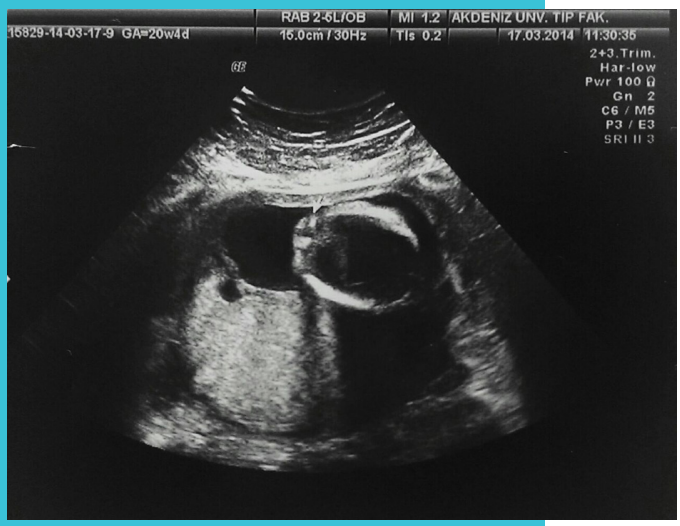
Case presentation

A 41 year-old-woman gravida 3-para 1 revealed to our institution at the gestational age of7weeks 5 days. In obstetric sonography, gestational age was recalculated by crown-rump length of 11.6 mm to 7 weeks and 2 days. Serologically Rubella IgG and IgM were relatively high. HBS-AG, HBS-AB, HBC-IGM, HCV-AB, HIV 1+2(core+envelope), VDRL-RPR, Toxoplasma IgG and IgM were negative. No clinical evidence was detected for maternal active rubella infection was revealed at physical examination such as rubelliform rash, lymphadenopathy. The second serology for rubella performed at 9 weeks; Ig G 77 IU/ml, Ig M 1.04

IU/ml. Rubella-specific IgG avidity index was measured to exclude primary rubella infection. Avidity index was found high which supported past infection. The karyotype analysis of parents revealed no chromosomal abnormality. At 17 weeks, amniocentesis was performed and no chromosomal abnormality was identified. The karyotype was reported as 46;XX, female. Fetal sonographic evaluation at 20 weeks 4 days revealed ventriculomegaly and occipital localized encephalocele (Figure 1). Termination and autopsy evaluation was performed by the permission of parents. In autopsy evaluation macroscopically chraniochisis was detected. In microscopic evaluation chronioschisis was subtyped as meningocele and left lung was compatible with the gestational age of the fetus, but in the middle segment of the right lung a 0.5 cm sized lesion characterized with marked alveolar distention was detected (Figure 2, 3). This lesion was diagnosed as CLE.

Figure 1.

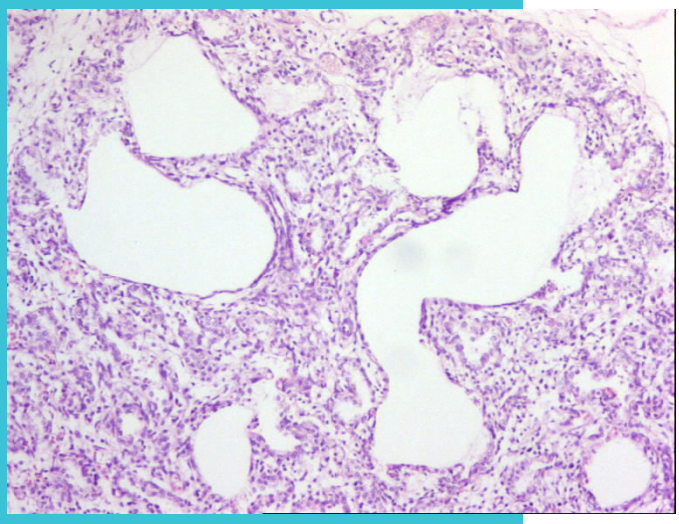


Sonographic appearance of fetus with ventriculomegaly and occipital localized encephalocele

Discussion

CLE is a rare malformation characterized by over distension of pulmonary lobes. Several etiologies have been shown for the formation of CLE. The pathophysiologic mechanism of CLE consists of disruptions of bronchopulmonary development due to abnormal interactions between endodermal and mesodermal components of the lung, meaning bronchial cartilage dysplasia or congenital deficiency of bronchial cartilage creating a ball-valve type effect that results in progressive lobar hyperinflation and distal air trapping [4]. The most accepted theory that CLE is formed as a result of intrabronchial or extrabronchial obstruction. Intrabronchial obstruction reasons are well known, including mucous plugs, bronchial stenosis or atresia, granulomas, mucosal folds and endobronchial polyp [4-7]. The abnormalities of cardiopulmonary vessels can manipulate CLE by compressing airways, especially vasculatory rings and slings, tetralogy of Fallot, patent ductus arteriosus. As a result of congenital heart disease and the subsequently increased pulmonary artery pressure vessels become dilated and compress airway tree. Esophageal duplication cysts, mediastinal cysts, CCAM, medulloblastoma, teratoma and bronchogenic cysts are known reasons for CLE [6]. Congenital cytomegalovirus infections can be reason for CLE [8-9]. Relapsing emphysematous changes can indicate an extraluminal compression [10]. Emphysematous lungs of infants younger than six months can result from foreign body aspiration (FBA).

Figure 2.

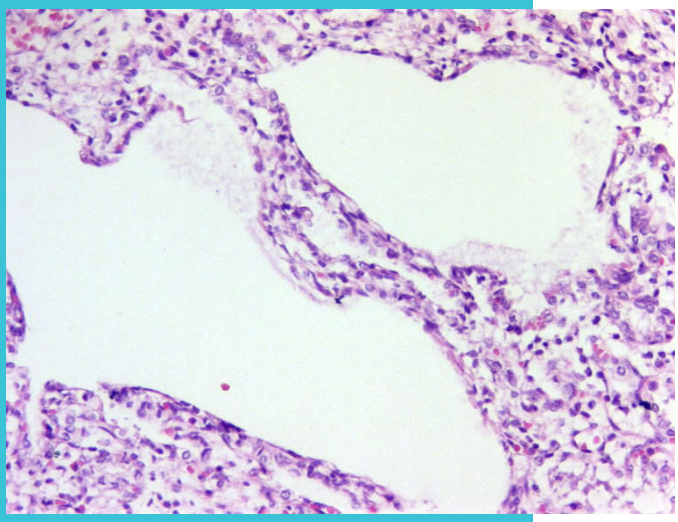


Alveolar distention of lung (H&E, x100)

Conventional radiographic imaging and computerized tomography can be suggestive for both CLE and FBA, bronchoscopy is effective in differential diagnosis of these two disorders [11]. Hislop and Reid documented a view named ‘polyalveolar lobe’ a possible eti-

ology for CLE, indicates greatly increase the numbers of alveoli in normal size, in 1970. However, the exact cause cannot be identified in the half of the patient [12]. Clinical findings and main symptoms of patients include respiratory distress, cough, tachypnea, cyanosis, poor feeding, suprasternal, subcostal or intercostal retractions, wheezing, weak lung sounds on the lesion-sided hemi thorax.

Figure 3.



Marked alveolar dilatation (H&E, x200)

And also we can examine weak and moved heart sounds due to mediastinal shift to the contra lateral side [1, 3]. Fetal lung malformations give increased echogenicity when it is compared to the liver tissue or normal lungs. Other findings may be mediastinal shift, displacement of the heart, compression to adjacent and contralateral lung tissue on sonographic evaluation. Differential diagnosis of hyperechogenic lung lesions are as follows: CCAM (congenital cystic adenomatoid malformation), upper airway obstruction, pulmonary sequestration, bronchogenic cysts and CLE [2, 13-14]. Partially increased hyperechogenic lesions may indicate CLE, CCAM, pulmonary sequestration and bronchogenic cysts compared to the upper airway obstruction which is seen as bilateral hyperechogenic enlarged mass on in utero sonography. Although microcystic or macrocystic mass appearance on fetal sonography and the echogenicity supports CCAM as an imaging diagnosis, the further investigations should be done postnatally because; some cases which are thought to suffer from CCAM, may have CLE [14]. The exact diagnosis of CLE is based on combination of clinical, imaging and histopathologic examinations. Pulmonary sequestrations can be distinguished to have vascular supply rooting from aorta, on Doppler sonography. Fetal MRI is done to readily rule out congenital diaphragmatic hernia and can be supportive technique together with CT in preoperative session. High intra-thoracic pressure leading to increased nuchal translucency in the first trimester and no obvious increases in peak systolic velocity of pulmonary veins may be the indicator for CLE, which needs further assessments and should be supported by novel findings [15-16]. The main outcome predictors are polyhydramniosis and fetal hydrops, which suggests fetal poor prognosis may results in fetal demise. Antenatal remarkable findings highlights the significance of postnatal follow-up whether the lesion echogenicity differs in size, even gets disappeared in the prenatal course [2, 13-14]. Chest x-ray and CT is effective imaging techniques in the most of cases. Hyper inflated and hyper lucent areas, mediastinal shift and diaphragmatic eversion can suggest CLE. Some cases are misdiagnosed as tension pneumothorax on chest radiograph and chest tube insertion can make the situation worse. Attenuated, stretched vascular markings in the hyperinflated area should be seen to avoid inappropriate intervention at postnatal course [17]. Our case was in early gestational age and no prenatal imaging findings were detected. The termination reason was ventriculomegaly and meningocele. Incidentally CLE was found in autopsy examination. CLE may be associated with cardiovascular anomalies in 14% of cases. Although uncommon, renal, gastrointestinal, musculoskeletal, and cutaneous malformations may also occur [4]. In English literature, this case is the first CLE associated with meningocele.

Acknowledgments
None

Conflict of Interest
Authors declare no conflict of interest

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