Congenital bronchopulmonary sequestration presenting as a thoracic tumor: A case report

K.I. Pappa[1], N.P. Anagnou[2], A. Elsheikh[3], S.S. Bikouvarakis[1], A. Konstantinidou[4], E. Salamalekis[1]

[1]1st Department of Obstetrics and Gynecology, University of Athens School of Medicine, Alexandra Hospital,
[2]Department of Basic Sciences, University of Athens School of Medicine
[1]3rd Department of Obstetrics and Gynecology, University of Athens School of Medicine, Attikon University Hospital,
[4]Department of Pathology, University of Athens School of Medicine, Athens (Greece)

Summary

The accurate diagnosis of fetal thoracic tumors still remains unclear despite the progress in imaging technology. The differential diagnosis between tumors and congenital anomalies of the fetus respiratory system, largely depends on the diagnostic approaches involved. We report a case of a 25-year-old woman, gravida 3 para 0, who was seen at the 23rd gestational week for routine obstetric examination. The ultrasound scan detected a lung mass, occupying the whole left hemithorax with a significant shifting of the mediastinum exhibiting features compatible with cystic adenomatoid malformation (CAM). No other congenital anomalies were noted. Color Doppler ultrasound failed to detect any blood supply to the mass. Amniocentesis disclosed a normal male karyotype. Pregnancy termination was performed according to the parents' request, with the use of misoprostol and a 500 g dead fetus was delivered. The autopsy followed by detailed histological examination, disclosed the diagnosis of pulmonary sequestration. It is important to emphasize that the initial impression concerning the sonographic appearance and the size of the mass is not always in accordance with the diagnosis of the lesion and the outcome of the pregnancy. These data suggest that in cases of fetal pulmonary tumors, a thorough and comprehensive combination of imaging approaches should be employed followed by a pathologic examination of the congenital anomaly in order to establish a definitive diagnosis.

Key words: Tumor; Fetal thorax; Congenital bronchopulmonary malformation.

Introduction

Fetal thoracic tumors represent rare neoplasias. Despite the progress in imaging technology, the accurate diagnosis of fetal thoracic tumors still remains incomplete. Prenatal sonography has been used extensively for the diagnosis of fetal chest tumors, as a first stage of detection. The differential diagnosis of an echogenic lung mass (with or without a cystic component) includes congenital cystic adenomatoid malformation (CAM), pulmonary sequestration, bronchogenic cysts, thoracic neuroblastoma, tracheal or bronchial atresia, congenital lobar emphysema, and pulmonary arteriovenous malformation [1].

CAM, a rare pulmonary abnormality, is characterized by excessive growth of the terminal respiratory bronchioles. Sonographically, the diagnosis is established by the visualization of an echogenic mass within the fetal chest, associated with cystic lesions of varying sizes dispersed in the mass. Pulmonary sequestration reflects an aberrant architecture of the lung tissue, which is perfused but not ventilated and receives its blood supply primarily from the systemic circulation. Pulmonary sequestration may be located either within (intralobar) or outside of the normal lung tissue (extralobar) and may develop above or below the diaphragm. Rarely, the sequestrated lung communicates with the foregut via a patent or a non-patent tract [1].

A relatively small number of reports refer on the outcome of pregnancies following a prenatal diagnosis of pulmonary sequestration, which displays a wide spectrum of events ranging from intrauterine fetal death to the birth of asymptomatic infants. This paucity of reports reflects indirectly the difficulties involved in establishing antenatally an accurate diagnosis.

In the present study, we describe a case of pulmonary sequestration with esophageal communication, antenatally diagnosed as a thoracic tumor and discuss the issues involved in the diagnosis and management of fetal thoracic malformations.

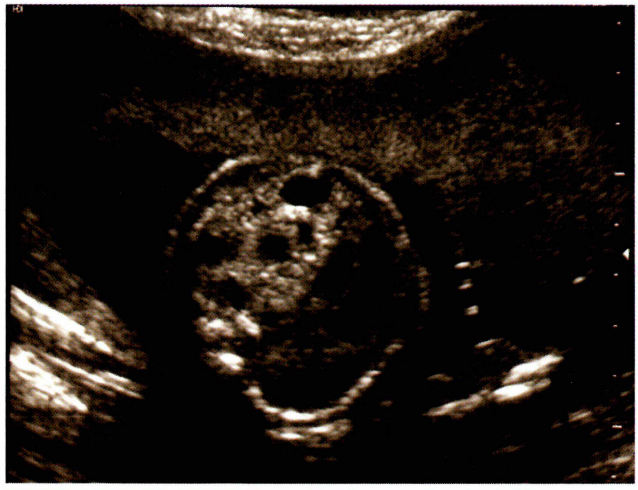
Case report

A 25-year-old White woman, gravida 3 para 0, was referred for routine obstetric ultrasound examination in the 23rd gestational week. The pregnant woman had no family history of congenital malformations, her prenatal history had been uncomplicated and she had received regular prenatal care. No significant findings on her medical history or drug abuse were documented. The ultrasound scan detected a hyperechogenic mass with a multicystic pattern as shown in Figure 1.

The mass was lying on the top of the left hemidiaphragm, expanding to the left thorax, displaying a small degree of shifting of the mediastinum to the right. No other congenital anomalies were noted. Color Doppler ultrasound failed to detect any blood supply to the mass. Amniocentesis disclosed a normal male karyotype. Based on these findings, the differential diagnosis was compatible either with a thoracic tumor or with a congenital cystic lung lesion. The patient returned to the clinic

Revised manuscript accepted for publication June 10, 2004

Fig. 1



Fig

Fig. 3

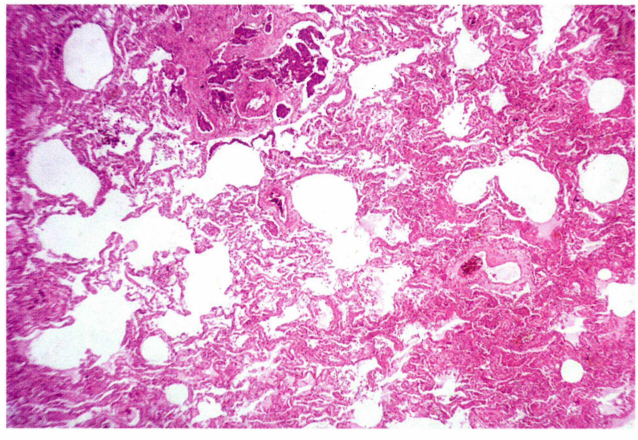


Fig.

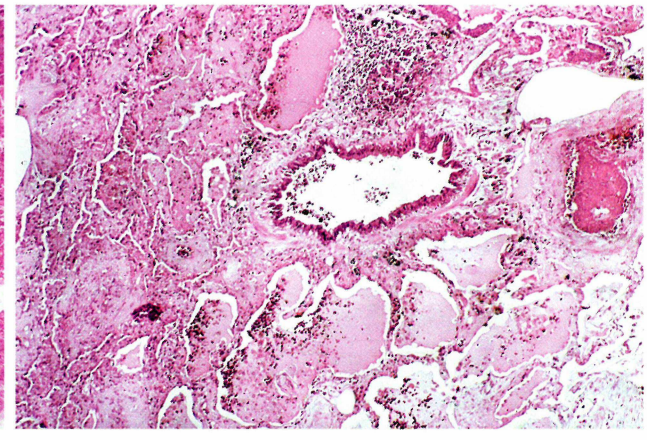


Figure 1. — Transversal sonographic appearance of the fetal thorax in the 23rd gestational week showing an echogenic mass with a multicystic component in the left lower lobe of the lung. A shifting of the mediastinum and heart structures to the right is clearly shown.
Figure 2. — Macroscopic appearance of the well-confined mass (arrowhead) communicating via a small stalk (arrow) with the lower part of the esophagus. The two hypoplastic pulmonary lobes (RL, right lobe, and LL, left lobe) are shown behind the esophagus.
Figure 3. — Lung tissue with characteristic appearance at the saccular stage of development. The saccular spaces are focally intercommunicating (original magnification x 200).
Figure 4. — Dilated bronchioli surrounded by saccular spaces. A large thrombosed vessel (upper right) and several lesions of hemorrhagic necrosis are shown. Fibrosis is also present at the left upper part of the field (original magnification x 200).

after two weeks to repeat the ultrasound scan which disclosed a rapid enlargement of the mass, occupying by that time the whole hemithorax with a significant shifting of the mediastinum, and was associated with non-immune hydrops fetalis and polyhydramnios.

The couple received appropriate information regarding the case and opted to terminate the pregnancy, despite the advice of the attending physicians to proceed further by employing additional diagnostic procedures, such as magnetic resonance imaging (MRI). Following the decision of the parents, pregnancy was terminated using misoprostol and a 500 g dead fetus was delivered.

The macroscopic appearance of the fetus lacked any abnormalities. At autopsy, a triangular pink colored irregular mass, measuring 4.3 cm x 3.5 cm x 3.4 cm, was located in the left hemithorax with its upper edge attached to the base of the left lung, but clearly separated from it. The mass was connected with a small stalk to the lower third of the esophagus at the gastroesophageal junction. Just behind the stalk, a feeding artery was running, arising from the descending aorta. Both lungs were hypoplastic and besides the hydropic features, no other significant findings were noted (Figure 2). Microscopic examination of the mass, revealed a lung tissue composition at the saccular stage of development (Figure 3). Dilated bronchioli and saccular respiratory ducts were apparent. Large vessels containing clots were surrounded by extensive areas of hemorrhagic necrosis (Figure 4). Macrophages were present and filled the terminal airways. Areas with cystic intercommunicating spaces compatible with CAM type II were also noted, although lining epithelium could not be identified due to the necrotic lesions. Interstitial fibrosis was also present (Figure 4). All these findings were compatible with typical features of pulmonary sequestration.

Discussion

The echogenicity of the normal fetal lung on ultrasound is similar to that of the fetal liver and spleen. Pulmonary abnormalities can be detected by ultrasound as early as 16 weeks of gestation. However, in our case the routine ultrasound at the 17th week did not reveal any visible lung abnormalities.

Pulmonary sequestration is characterized by a high degree of echogenicity compared to the liver and spleen since the lesion is solid and is non-aerated. Frequently, a thin echogenic margin exists around the lesion. Small cysts may also be present within the sequestrated lung or can be the prominent component of the mass [1]. Sequestrated lung is frequently found in combination with CAM or can mimic CAM on prenatal ultrasound scan [2]. Prenatally, 12-16% of pulmonary sequestration is initially diagnosed as CAM [3]. Indeed, in the present case the ultrasound pattern was echogenic and associated with cystic areas; these sonographic features were compatible with CAM type II along with the cystic areas, which were later identified histologically. Thus, the accurate diagnosis of pulmonary sequestration was definitively established at autopsy, followed by histological investigation. To overcome these diagnostic difficulties, the use of a series of novel imaging techniques have been introduced at the prenatal stage of investigation. Namely, color Doppler ultrasound is used to map the feeding vessels of the mass, while MRI, CT and angiography can be used to facilitate the differential diagnosis and establish a definitive and accurate diagnosis [1].

The malformation of pulmonary sequestration arises early in embryogenesis, when the lung starts to develop from the primitive foregut, entering a saccular stage of development [4]. Our case exhibited typical features of this stage associated with the formation of a small stalk communicating with the lower part of the esophagus.

Regarding the appearance of the mass, this seems to undergo frequently significant changes during pregnancy. Pulmonary sequestration tends to decrease in size in 68% of the cases, or even disappear gradually until birth [3]. This frequent outcome has generated several speculations for the explanation of its mechanism. A major hypothesis considers that lesions that outgrow their blood supply, decompress into the tracheobronchial tree and can lead to the obstruction of their vascular pedicle. Several investigators argue that this is a deceiving presentation, caused by the accelerated growth of the normal lung that exceeds the growth of the mass. Alternatively, others suggest a mechanism of progressive fibrosis of dysplastic tissue or thrombosis of the feeding vessel, leading eventually to mass regression. This seems to be the main reason for the necessity of repeated ultrasound examinations during pregnancy, recommended by many investigators [5].

The major complications of pulmonary sequestration in utero are polyhydramnios, non-immune hydrops fetalis, intrauterine death and preterm delivery [5]. The most important prognostic factor of pulmonary sequestration is the development of hydrops caused by large masses. Fetuses complicated with polyhydramnios and hydrops have poor prognosis and may need fetal intervention requiring lung lobectomy. In utero intervention should be the treatment of choice in selected cases, when the lesions increase in size and the mass becomes amenable or when hydrops develops. Interestingly, regression of the size of the mass has also been reported in rare cases associated with hydrops fetalis [5]. In our case, pregnancy termination was performed following the parents' decision which was influenced by the accelerated deterioration of fetal status. Thus, it is unclear in our case whether the mass would have regressed eventually, in the background of hydrops fetalis.

The treatment of pulmonary sequestration even after birth remains controversial. Conservative treatment is recommended for patients without complications, while surgical intervention is reserved when symptoms occur. Alternatively, other authors recommend routine resection of the growing mass as a preventive measure to avoid a malignant transformation of epithelial and mesenchymal masses and/or the associated pulmonary sepsis or the recurrent infections and symptoms to the mass itself, such as bleeding and pneumothorax [4].

Regardless of the in utero regression of the thoracic mass, it is advisable that neonates undergo postnatal evaluation for the definitive diagnosis and documentation of the outcome of the anomaly. Chest X-ray, ultrasonography with Doppler, CT and MRI, usually can reveal the nature and the vasculature of the lesion. Barium swallow should be performed when there is evidence of a communication with the gastrointestinal tract.

The overspread use of ultrasonography has led to early detection of congenital malformations and other fetal abnormalities, including pulmonary sequestration. A large number of cases are nowadays prenatally diagnosed accurately. The diagnostic difficulties encountered in our case using these technologies suggest that a thorough and comprehensive combination of imaging approaches should be employed followed by a pathologic examination of the congenital anomaly.

Acknowledgements

We thank Dr. Aspasia Kyroudi for invaluable discussions and suggestions and Popi Tsiara for excellent technical assistance.

References

[1] Winters W.D., Effmann E.L.: "Congenital masses of the lung: prenatal and postnatal imaging evaluation". *J. Thorac. Imag.*, 2001, *16*, 196.
[2] Borsellino A., Alberti D., Vavassori D. *et al.*: "Communicating bronchopulmonary foregut malformation involving a mixed sequestration/cystic adenomatoid malformation: a case report". *J. Pediatr. Surg.*, 2002, *37*, E38.
[3] Tsao K., Albanese C.T., Harrison M.R.: "Prenatal therapy for thoracic and mediastinal lesions". *World J. Surg.*, 2003, *27*, 77.
[4] Halkic N., Cuenoud P.F., Corthesy M.E. *et. al.*: "Pulmonary sequestration: a review of 26 cases". *Eur. J. Cardiothorac. Surg.*, 1998, *14*, 127.
[5] Roggin K.K., Breuer C.K., Carr S.R. *et al.*: "The unpredictable character of congenital cystic lung lesions". *J. Pediatr. Surg.*, 2000, *35*, 801.

Address reprint requests to:
K.I. PAPPA, M.D.
Third Department of Obstetrics
and Gynecology
University of Athens School of Medicine
Attikon University Hospital
12462 Athens (Greece)
www.kpappa@imbb.forth.gr