A rare case of congenital pulmonary lymphangiectasia, hydrothorax and ascites in a male embryo aborted at 20 weeks of gestation

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Summary

A case of a male embryo aborted at the 20th week of gestation with extensive ascites, hydrothorax, pulmonary lymphangiectasia and pulmonary hypoplasia is presented together with the pathological findings, the etiology, differential diagnosis, course and therapy of this pathologic entity. Also a short review of the literature is discussed.

Key words: Hydrothorax; Pleural effusion; Ascites; Lymphatics.

Introduction

Congenital pulmonary lymphangiectasia is a rare condition, characterized by extensively dilated pulmonary lymphatics in the bronchovascular connective tissue along the interlobular septa and in the pleura [1, 2]. This disease is usually fatal within a few hours or days of life. Its etiology is heterogenous [3-5]. It may present as part of syndromes such as Noonan, Turner or Down , but most cases are sporadic [5]. The incidence of congenital pulmonary lymphangiectasia is one in 10,000 gestations, with a 2:1 male/female ratio [1, 2].

Case Report

A 33-year-old primigravida visited our hospital at 20 weeks of an uneventful gestation for a routine check-up. The ultrasound of the second trimester revealed bilateral pleural effusion especially in the left thoracic cavity and ascites.

The first semester check-up showed that the patient was CMV (+), toxoplasma (-), coxsackie (-), rubella (±) and Prader-Willi syndrome (-). No fetal defects were mentioned in the previous ultrasound.

The patient underwent a therapeutic abortion after giving an informed consent. The aborted embryo was male and weighed 381 g; its length from head-foot was 25.5 cm and from head-hips 15.5 cm. The foot sole length was 3.3 cm. The head, thoracic and abdominal circumference were 19 cm, 16.5 cm, and 16 cm, respectively.

On gross examination, extensive abdominal distension was observed. The face (ears, nose, mouth) was normal and the eyes were closed. The extremities were normal and the external genitalia were male. The thoracic and the peritoneal cavity were full of pellucid, serous fluid and the viscera were congestive, normal in position and size. The brain weighed 43 g. The brain ventricles were full of cerebrospinal fluid and the meninges were congestive. The thoracic cavity was dilated and full of serous fluid. The lungs showed extensive, hemorrhagic infiltration especially the left lung. The heart revealed an atrial septal defect and patent ductus arteriosus, whereas the aorta and the pneumonic artery were normal. The peritoneal cavity was also dilated and full of serous fluid. The viscera were congestive and fetal weight was within normal limits for the fetal age.

Microscopic examination of the lungs revealed many cystic spaces filled with fluid, lined by endothelium and distributed under the pleura, in the interlobar spaces and round vessels (Figures 1, 2). Hydrothorax, ascites and visceral congestion were also present.

The placenta was discoid measuring 10.5 x 10.5 x 3 cm and weighing 208 g with an eccentric outgrowth of the umbilical cord. The coloboma of the umbilical cord had three vessels. Its diameter was 1.2 cm and length 10.5 cm. Furthermore, focal, dispersive, white lesions measuring 0.2 cm were found in the placenta.

Microscopic examination showed:

a) intensive vascular congestion and elevated vascularization of the chorionic villi;

b) dilatation and congestion of the lung vessels;

c) old and recent focal placenta infracts;

d) lesions of placentitis;

e) lesions of chorioamnionitis.

Discussion

Congenital pulmonary lymphangiectasia is a rather fatal disease. There are three forms of the disorder [3]:

a) isolated congenital pulmonary lymphangiectasia (poor prognosis) [4];

b) congenital pulmonary lymphangiectasia associated with pulmonary venous obstruction;

c) congenital pulmonary lymphangiectasia associated with a generalized defect in lymphatic development.

Certain genes are implicated in such a pathology e.g., FOXC2 transcription factor on chromosome 16q24.3, VEGFR3 mutation in the endothelial growth factor receptor 3, and integrin alpha-9-beta-1 receptor for extracellular matrix protein [5].

The pleural effusion is usually chylous and occurs in the right pleural space (our case occurred in the left pleural space) [1]. In a retrospective study of eight

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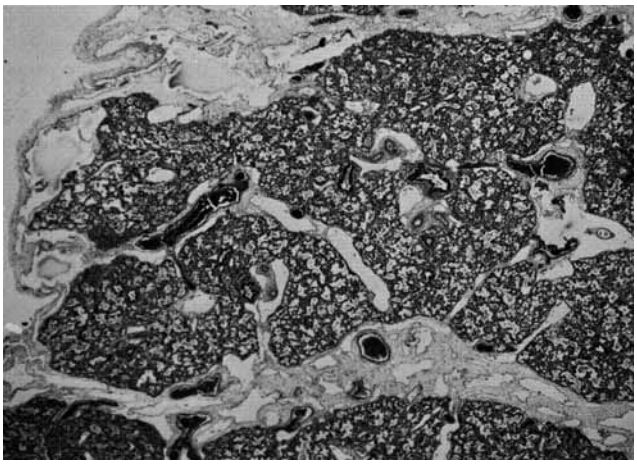
Fig. 1



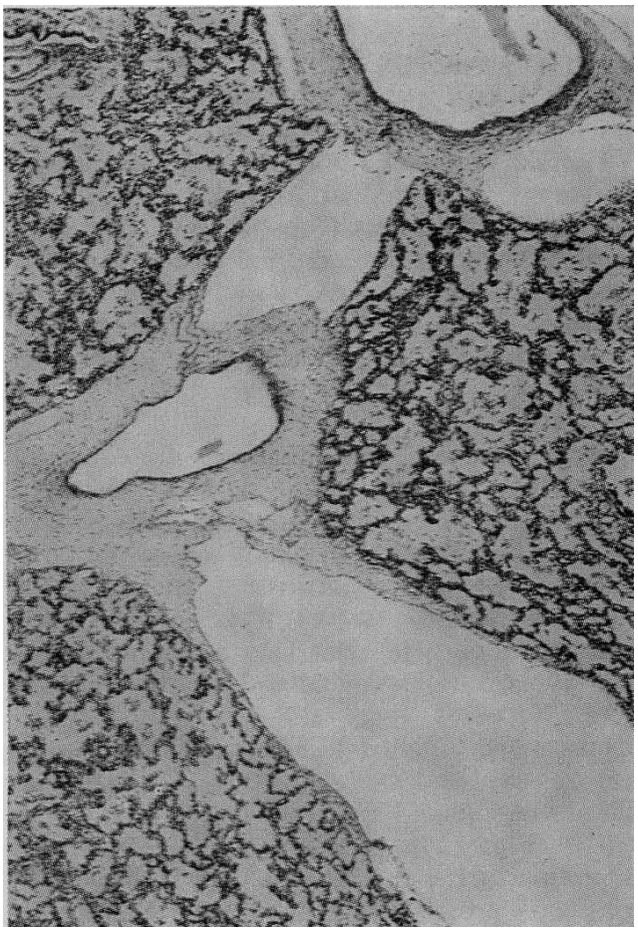


Fig. 2

Figure 1. — Histological section of fetal lung showing dilated lymphatic spaces in subpleural and interlobar spaces (hematoxytin-eosin, x 25).
Figure 2. — Histological section of interlobar spaces showing dilated lymphatics (hematoxytin-eosin, x 250).

aborted fetuses (7 males, 1 female), the fetuses weighed 463.4 g (range 177-681 g) [4]. Our case was a male embryo weighing 381 g. Six were aborted between 19 and 24 weeks of gestation for multiple malformations or anencephaly and two were spontaneously aborted [4]. Our case underwent a therapeutic abortion at 20 weeks of gestation. The main characteristics are the dilatation of subpleural and septal lymphatic space and the positive D2-40 immunostaining of lymphatic endothelium [1].

The differential diagnosis includes chromosomal, cardiac and infectious parameters. The diagnosis is confirmed by the postnatal lung biopsy. The prognosis depends on the severity of the symptoms. The infants usually die due to respiratory distress shortly after birth [6]. A subgroup of children with congenital pulmonary lymphangiectasia – if treated aggressively – may have a good prognosis for long-term survival [7]. Prenatal counseling should be proposed to the parents.

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