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Prenatal Diagnosis of Fetal Cystic Hygromas Associated with Generalized Lymphangiectasis

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Ultrasonography has made possible the prenatal diagnosis of many congenital fetal abnormalities. This report describes two cases of bilateral cystic hygromas of the neck associated with generalized lymphangiectasis that were diagnosed by ultrasound. Ultrasonic scans revealed moderate polyhydramnios; thick, edematous placenta and edematous fetus with large cystic mass occupying both sides of the neck and extending to the upper chest wall, ascites, and pleural effusion at gestational ages of 21.5 and 24 weeks, respectively. In one case, chromosomal study from amniotic fluid cell culture revealed X chromosome monosomy, often associated with lymphatic anomalies. The prenatal diagnosis was confirmed at birth: both infants delivered prematurely, were stillborn, and showed gross evidence of cystic hygromas of the neck. In this lymphatic defect, chromosomal analysis may be used for the diagnosis and in genetic counseling for subsequent pregnancies.

Lymphangiomas and cystic hygromas are congenital malformations of the lymphatic system. These comparatively rare disorders are composed of lymph-containing endothelium-lined spaces which vary in size from channels of capillary dimensions to cysts of several centimeters in diameter (1). In the process of embryonic development, various areas of the lymphatic analogue may grow abnormally, become sequestered, and fail to achieve efficient anastomosis with large lymph channels. When the obstructed lymphatic bed lies in an area of loose areolar tissue, cysts develop. The cystic form tends to arise in the neck and axillary areas. When the obstructed lymphatics lie in the area where muscle and fibrous tissues intermingle with areolar tissue, the lymphangiomatous form occurs (1). The incidence of cystic lymphangioma is unknown, but Goetsch (2) reported that of 981 tumors of all kinds, 1.2% were hygromas. About 50% of these lesions are present at birth and up to 94% are evident by two years of age (3). Bilateral cystic hygromas of the neck associated with widespread lymphangiectasis are a more severe form of this disorder. They are incompatible with life and are found only in the abortus and stillborn fetus (4-7).

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The present report describes two cases of bilateral cystic hygromas of the fetal neck with generalized lymphangiectasis which were diagnosed prenatally by ultrasonogram. In one case, a chromosomal study from amniotic fluid cell culture was also performed. Our report also reviews the pathogenesis and relationship between these lymphatic defects and X chromosome monosomy.

Case Reports

Case 1

A 23-year-old black woman (gravida 3, para I, abortion I) came to the prenatal clinic on September 17, 1976 with her uterus enlarged to approximately 10-12 weeks of gestation. Her last menstrual period was June 12, and a pregnancy test was positive. In 1975, she had delivered vaginally a male infant weighing 6 pounds, 4 ounces. Her second pregnancy ended in spontaneous abortion a year later. Her history and initial physical examination were unremarkable except for trichomonas vaginitis. All prenatal laboratory studies were normal.

When she returned one month later, the size of her uterus was compatible with 16 weeks of gestation, and fetal heart tones were detected by ultrasonic stethoscope.

On November 26, the uterine fundus had risen to the size of a 30-week gestation. Fetal heart tones were faint but audible. Polyhydramnios was suspected. An ultrasonographic examination taken on December 10 revealed polyhydramnios and a live fetus with bilateral large cystic

masses projected posteriorly and bilaterally from the neck, generalized edema, ascites, and pleural effusion. The probable diagnosis was cystic hygromas of the fetal neck with generalized lymphangiectasis.

By December 24, the patient reported that there had been no fetal movements for three to four days. Fetal heart tones were not audible with an ultrasonic stethoscope. Repeat ultrasonographic examination on December 28 failed to demonstrate motion of the fetal extremities or heart by Real-time examination.

On January 7, 1977, the patient delivered a stillborn female infant weighing 1600 gm. The infant was grossly macerated and extremely edematous with ascites and pleural effusion. Bilateral cystic masses were present in the neck (Fig. 1). Autopsy findings were compatible with cystic hygromas and generalized lymphangiectasis. No other organ abnormality was found.

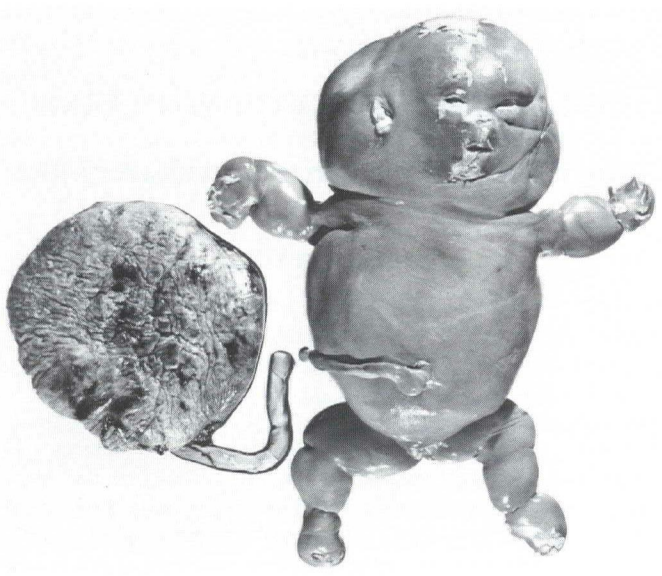

Fig. 1
Case 1
A macerated stillborn female infant with bilateral cystic hygromas in the neck with generalized lymphangiectasis.

Case 2

A 35-year-old black woman (gravida 5, para 4) visited the prenatal clinic on April 18, 1978. Her last menstrual period was on January 10, and her previous medical and obstetric history was unremarkable. She had delivered four full-term infants vaginally after uneventful pregnancies. Physical examination was also unremarkable except for exogenous obesity; she weighed 111 kg. The patient declined genetic amniocentesis for maternal age. Prenatal laboratory studies were within normal limits.

The patient returned monthly, presented no specific problems, and her examinations were unremarkable. An ultrasonogram taken on July 13 revealed a single fetus in breech presentation with large cystic masses occupying both sides of the neck and extending to the upper chest wall. The placenta and fetus were edematous with ascites and pleural effusion. With the experience gained from ultrasound study of the previous patient, bilateral cystic hygromas of the fetal neck with generalized lymphangiectasis were diagnosed. Fetal cardiac activity was observed through Real-time ultrasound. However, motion of the fetal body and extremities was not observed. On July 31, no cardiac motion was observed through Real-time ultrasound. Amniocentesis was performed for chromosomal study, since this severe form of lymphatic abnormality is reported to have X chromosome monosomy. The study confirmed X chromosome monosomy (Fig. 2).

On August 3, the patient delivered vaginally a stillborn female infant weighing 780 gm. The infant was grossly macerated with large cystic hygromas in the neck (Fig. 3). Autopsy findings were compatible with cystic hygromas of the neck with generalized lymphangiectasis. No other organ anomaly was found.

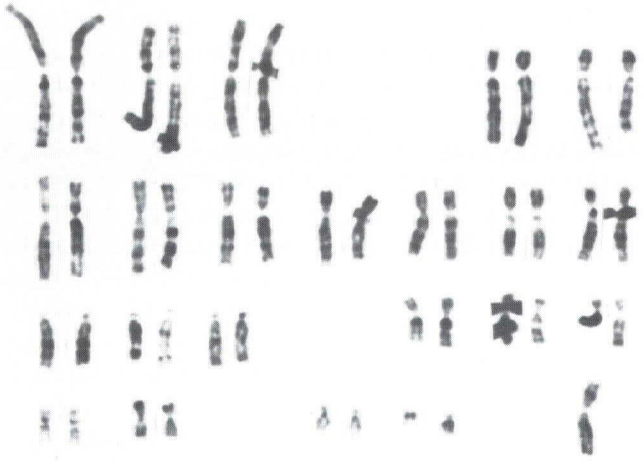

Fig. 2
Case 2
Karyotype from amniotic fluid cell culture revealing X chromosome monosomy.

In both cases, ultrasonic scans were performed using a commercially available linear B scanner and Real-time unit. Initial examination demonstrated the presence of a single live fetus in breech presentation of approximate gestational ages of 21.5 (Case 1) and 24 weeks (Case 2). In both cases, moderate polyhydramnios was noted, and the placenta was thick and edematous. Large cystic masses projected posteriorly and bilaterally in the cervical region of the fetus (Fig. 4). Similar multilocular cystic masses were

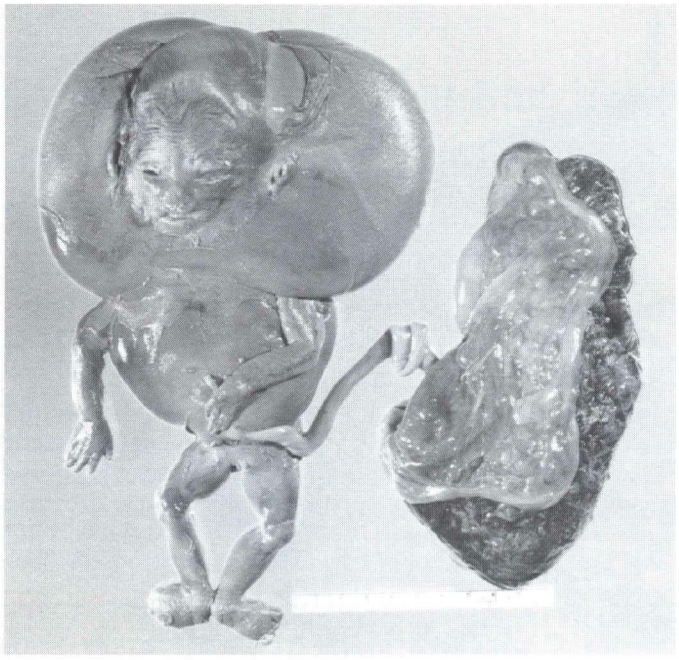

Fig. 3
Case 2
A macerated stillborn female infant with bilateral cystic hygromas in the neck with generalized lymphangiectasis.

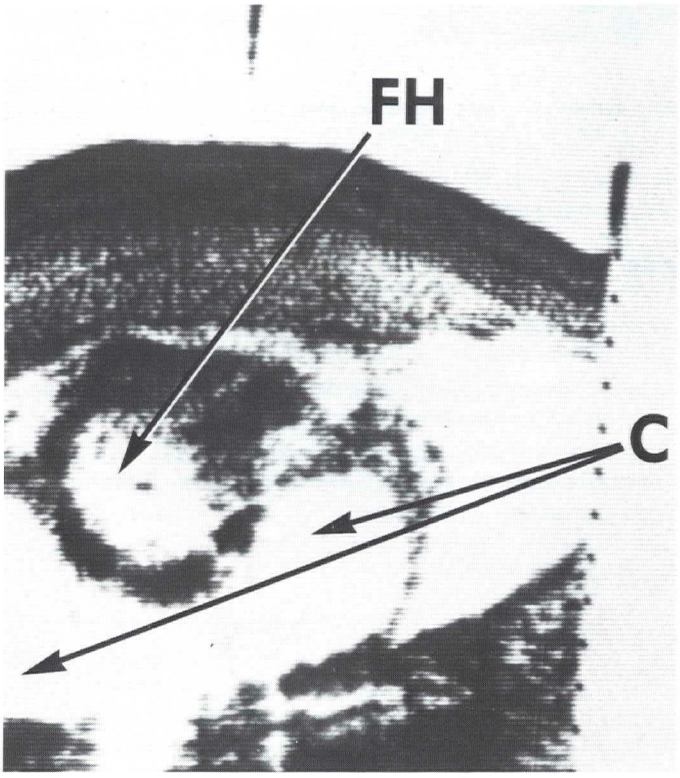

Fig. 4
Transverse scan of ultrasonogram obtained 13 cm above the umbilicus demonstrating bilateral cystic masses in cervical region. FH: fetal head, C: cystic masses.

found in the entire wall of the trunk (Fig. 5). Other findings included ascites, pleural effusion, and generalized edema of the skin. In both cases, Real-time ultrasound detected fetal heart motion initially, although no motion of fetal body or extremities was demonstrated.

Discussion

Ultrasonography has now made it possible to diagnose many congenital malformations of the fetus prenatally. The present case reports are examples of a rare congenital malformation that can be diagnosed by ultrasound because of its characteristic features.

Two cases of fetal cystic hygromas reported separately by Morgan, et al (3) and Shaub, et al (8), based on ultrasonic findings, were diagnosed as occipital encephalocele or meningomyelocele. In our two cases, cystic lesions projected posteriorly and bilaterally from the fetal neck, and multilocular cystic lesions extended into the entire wall of the fetal trunk. Ultrasonic findings of generalized skin edema, ascites, and pleural effusion can also be used to differentiate cystic lymphangioma from meningomyelocele or encephalocele.

In 1962, Verger, et al (2) reported a chromatin negative female infant with hygromas of neck and edema of the extremities. In 1966, Singh and Carr (5) reported this lymphatic abnormality in a series of XO abortus. It has been estimated that 97% of XO conceptus abort spontaneously (2).

Apparently, a few of these abnormal conceptus reach viable gestation. In milder forms of lymphatic abnormalities with XO chromosomes, reabsorption of fluid from cystic hygromas of the neck seems to be responsible for the neck webbing present in Turner's syndrome. Peripheral lymphedema frequently found in the newborn with Turner's syndrome also suggests that this chromosome anomaly is associated with a disorder in the lymphatic system. The chromosome anomaly from amniotic fluid cell culture in our second case revealed X chromosome monosomy. However, in one case of cystic hygroma, ultrasonically diagnosed by Adam, et al (6), chromosome analysis revealed normal female karyotype. In one other case reported by Frigoletto, et al (7), chromosomal analysis from amniotic fluid cell culture revealed 45 X and 46 XX mosaic karyotype. In this lymphatic defect, chromosomal analysis may be used in genetic counseling for subsequent pregnancies.

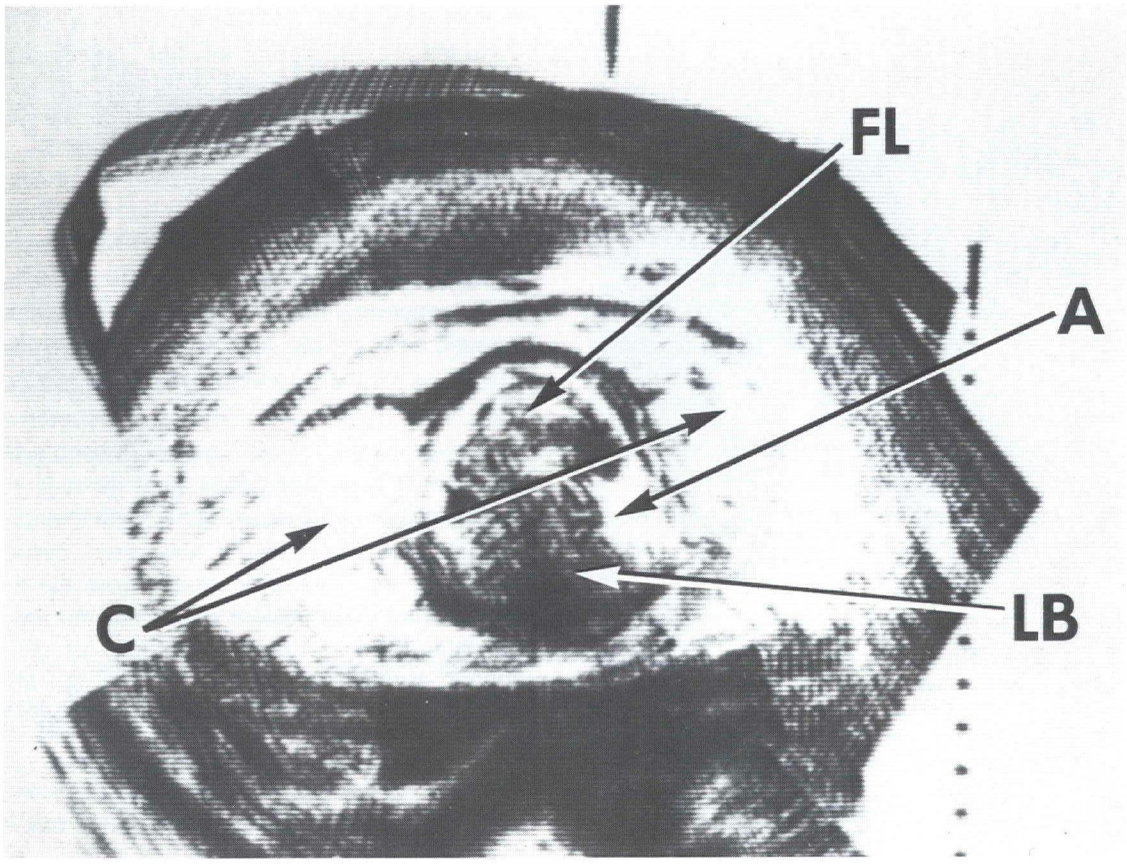

Fig. 5
Transverse scan obtained at level of umbilicus demonstrating multilocular cystic masses in entire wall of the trunk. FL: fetal liver; LB: loops of bowel; A: ascites; C: cystic masses.

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