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Case Report

PRENATAL DIAGNOSIS OF CONGENITAL CYSTIC HYGROMA WITH HYDROPS FETALIS AND SEVERE FETAL DYSMORPHISM: A CASE REPORT

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ABSTRACT

Cystic hygroma is a congenital malformation of the lymphatic system characterized by single or multiloculated fluid-filled cystic lesions, most commonly located in the cervical region. It is frequently associated with chromosomal abnormalities, genetic syndromes, structural malformations, and poor perinatal outcomes, particularly when accompanied by hydrops fetalis. Early prenatal diagnosis is essential for appropriate counseling and management. We report the case of a 33-year-old primigravida who presented for her first antenatal visit at 14 weeks and 3 days of gestation. Routine antenatal investigations revealed reactive syphilis serology with a Rapid Plasma Reagin (RPR) titre of 1:16. Obstetric ultrasonography demonstrated a single live fetus with multiple congenital anomalies, including a large cervical cystic hygroma, hydrops fetalis, facial dysmorphism, hyperextension of the neck and spine, and suspected cardiac abnormalities. Considering the severity of the anomalies and poor fetal prognosis, the couple was counseled regarding the findings and medical termination of pregnancy was performed. Gross examination of the abortus confirmed the presence of a large cystic hygroma, generalized edema, facial dysmorphism, and spinal hyperextension. This case highlights the importance of detailed first-trimester ultrasonographic evaluation in detecting major fetal anomalies. The presence of cystic hygroma associated with hydrops fetalis and multiple structural abnormalities indicates a poor prognosis and warrants multidisciplinary management, genetic counseling, and informed decision-making regarding pregnancy continuation.

Keywords*: Cystic hygroma; Hydrops fetalis; Dysmorphic fetus; Prenatal diagnosis; Congenital anomalies; Fetal hydrops; Ultrasonography; Genetic counseling.*

INTRODUCTION

Cystic hygroma is a congenital malformation of the lymphatic system characterized by single or multiloculated fluid-filled cystic lesions, most commonly located in the posterior cervical or occipitocervical region of the fetus. These lesions arise due to abnormal development of the lymphatic channels and failure of communication between the developing jugular lymph sacs and the venous system [1,2]. Histologically, cystic hygromas are composed of dilated lymphatic spaces lined by endothelial cells.

Although cystic hygromas are frequently identified in first-trimester abortuses, with an incidence ranging from 0.1% to 0.5%, their occurrence among live births is relatively uncommon, with an estimated incidence of approximately 1 in 6,000 live births [3]. Prenatally diagnosed cystic hygromas are strongly associated with chromosomal abnormalities and congenital malformations. Approximately 78% of affected fetuses have an underlying chromosomal anomaly, with Turner syndrome being the most common association. Other chromosomal abnormalities include trisomy 21, trisomy 18, trisomy 13, and Klinefelter syndrome [4,5].

Advances in prenatal ultrasonography, particularly transvaginal sonography, have enabled early detection of cystic hygromas during the first trimester. Increased nuchal translucency and cystic hygroma detected during early pregnancy are associated with a significantly increased risk of chromosomal abnormalities, structural malformations, fetal hydrops, and adverse perinatal outcomes [6]. The prognosis is particularly poor when cystic hygroma is associated with hydrops fetalis, major structural anomalies, or chromosomal abnormalities, with reported survival rates as low as 2–3% among antenatally diagnosed cases [7].

We report a case of a large fetal cystic hygroma associated with hydrops fetalis, facial dysmorphism, hyperextension of the neck and spine, and suspected cardiac anomalies diagnosed at 14 weeks of gestation, which resulted in termination of pregnancy following detailed prenatal counseling.

CASE PRESENTATION

A 33-year-old primigravida presented to the antenatal clinic of our institute for her first prenatal visit at 14 weeks and 3 days of gestation. She belonged to a low socioeconomic background and had conceived spontaneously. There was no significant past medical, surgical, drug, or family history. No history of exposure to known teratogenic agents during pregnancy was elicited.

Routine antenatal investigations were performed. Maternal serological screening revealed a reactive Venereal Disease Research Laboratory (VDRL) test, with a Rapid Plasma Reagin (RPR) titre of 1:16.

Obstetric ultrasonography demonstrated a single live intrauterine fetus corresponding to a gestational age of 14 weeks and 3 days. The fetus showed multiple congenital anomalies, including marked facial dysmorphism, a dysmorphic spine with persistent hyperextension of the neck, and a large cervical cystic hygroma. Generalized fetal edema consistent with hydrops fetalis (fetal anasarca) was also present. Furthermore, the four-chamber cardiac view and fetal stomach bubble could not be clearly visualized, suggesting the presence of associated structural abnormalities.

In view of the severe fetal anomalies and poor fetal prognosis, detailed counseling was provided to the couple regarding the nature of the abnormalities, likely outcome, and available management options. Following informed consent, medical termination of pregnancy was performed using misoprostol as per institutional protocol.

Gross examination of the abortus revealed a markedly dysmorphic fetus with a large cervical cystic hygroma, generalized edema consistent with hydrops fetalis, facial dysmorphism, and hyperextension of the neck and spine. The post-termination findings were consistent with the prenatal ultrasonographic diagnosis.

Considering the positive maternal syphilis serology, both partners received intramuscular benzathine penicillin G (2.4 million units) according to standard treatment guidelines. Genetic counseling was provided regarding the possible etiological factors, recurrence risk, and the importance of early prenatal screening in future pregnancies. The patient was discharged in stable condition with advice regarding preconception counseling and folic acid supplementation (5 mg daily) before future conception. Fetal karyotyping and genetic analysis could not be performed because of resource limitations and lack of parental consent.

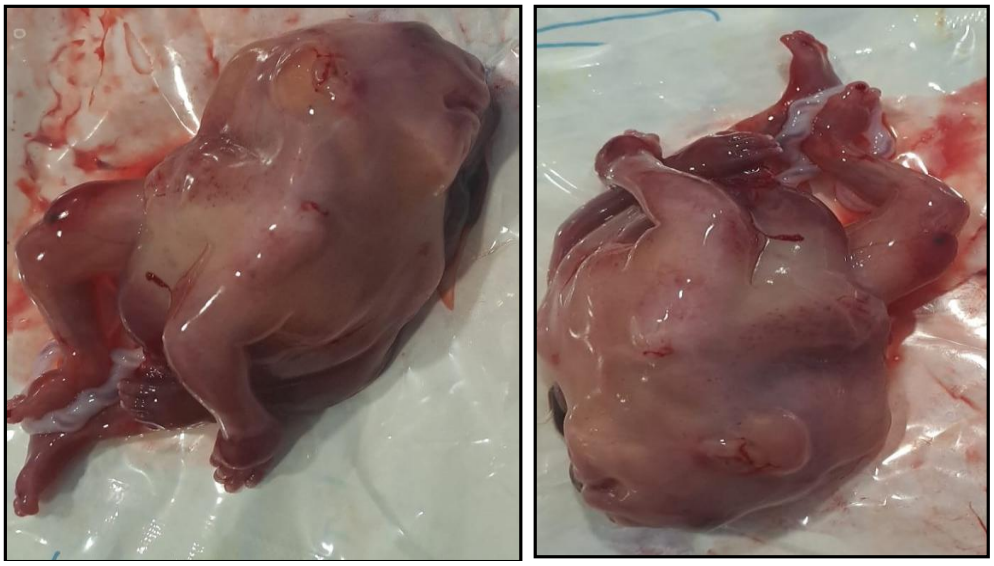

FIGURE 1, 2: ABORTUS SHOWING A LARGE CERVICAL CYSTIC HYGROMA ASSOCIATED WITH HYDROPS FETALIS, FACIAL DYSMORPHISM, AND HYPEREXTENSION OF THE NECK AND SPINE.

DISCUSSION

Cystic hygroma is a developmental malformation of the lymphatic system resulting from failure of communication between the primitive lymphatic sacs and the venous circulation during embryogenesis [8]. It most commonly presents as a septated cystic lesion in the posterior cervical region and is frequently detected during the first trimester of pregnancy owing to advances in prenatal ultrasonography. The prenatal diagnosis of cystic hygroma is clinically significant because of its strong association with chromosomal abnormalities, structural malformations, hydrops fetalis, and adverse fetal outcomes [4,5].

Approximately 50–80% of fetuses with cystic hygroma have an underlying chromosomal abnormality, with Turner syndrome being the most commonly reported association. Other chromosomal abnormalities include trisomy 21, trisomy 18, trisomy 13, and Klinefelter syndrome [4,5,10]. In addition, cystic hygroma may occur as part of several genetic syndromes, including Noonan syndrome, multiple pterygium syndrome, Fryns syndrome, and Pena–Shokeir syndrome [9,10]. Although fetal karyotyping could not be performed in the present case, the presence of multiple congenital anomalies, severe facial dysmorphism, hyperextension of the neck and spine, and hydrops fetalis strongly suggested an underlying chromosomal or genetic abnormality.

The prognosis of cystic hygroma depends on its size, presence of septations, associated structural anomalies, chromosomal status, and development of fetal hydrops. The coexistence of hydrops fetalis is considered a poor prognostic indicator and is associated with a markedly increased risk of fetal demise and perinatal mortality [6,7]. In the present case, the fetus demonstrated a large cervical cystic hygroma accompanied by generalized fetal edema, facial dysmorphism, and suspected cardiac abnormalities, indicating a very poor prognosis. Consequently, termination of pregnancy was considered after detailed counseling of the couple.

Congenital cardiac defects are frequently associated with fetal cystic hygroma and may contribute to the development of hydrops fetalis [9]. In the present case, the four-chamber cardiac view could not be adequately visualized on ultrasonography, raising suspicion of an associated cardiac anomaly. The inability to visualize the stomach bubble further suggested the possibility of additional structural abnormalities.

An important finding in the present case was the presence of maternal syphilis, evidenced by a reactive VDRL test and an RPR titre of 1:16. Although chromosomal abnormalities remain the most common cause of cystic hygroma, congenital infections have also been implicated in the development of fetal hydrops and severe fetal abnormalities. Previous reports have suggested that maternal infections, including syphilis and parvovirus B19 infection, may contribute to abnormal fetal development and hydrops fetalis [11,12]. Therefore, maternal infectious etiologies should be considered during the evaluation of fetuses presenting with cystic hygroma and hydrops.

The recognition of multiple anomalies in a fetus should prompt evaluation for an underlying malformation syndrome, sequence, or association. Accurate prenatal diagnosis is essential for determining prognosis, planning management, assessing recurrence risk, and providing appropriate genetic counseling to the family [13]. This case highlights the importance of detailed first-trimester ultrasonographic screening, comprehensive fetal anomaly assessment, and multidisciplinary management involving obstetricians, fetal medicine specialists, radiologists, pathologists, and clinical geneticists.

The major limitation of this report was the inability to perform fetal karyotyping and postmortem genetic evaluation, which precluded definitive determination of the underlying chromosomal etiology.

CONCLUSION

Cystic hygroma is an important prenatal marker of chromosomal abnormalities, genetic syndromes, and major structural malformations. The presence of associated anomalies such as hydrops fetalis, facial dysmorphism, and suspected cardiac defects is indicative of a poor fetal prognosis. Early prenatal ultrasonographic diagnosis enables timely counseling, further evaluation, and informed decision-making regarding pregnancy management. A multidisciplinary approach involving obstetricians, fetal medicine specialists, radiologists, pathologists, and clinical geneticists is essential for accurate diagnosis and optimal counseling. Furthermore, appropriate evaluation of maternal risk factors, including infectious diseases such as syphilis, should be undertaken in all cases presenting with fetal hydrops and multiple congenital anomalies.

DECLARATIONS

Availability of Data and Materials: All relevant data supporting the findings of this case report are included within the article. Additional information can be made available by the corresponding author upon reasonable request.

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REFERENCES

1. Goetsch E. Hygroma colli cysticum and hygroma axillare: pathologic and clinical study and report of twelve cases. Arch Surg 1938; 36:394-479.
2. Potter EL, Craig JM. Pathology of the fetus and the infant. 3d ed. Chicago: Year Book, 1975.
3. Cullen MT, Gabrielli S, Green JJ, et al. Diagnosis and significance of cystic hygroma in the first trimester. PrenatDiagn 1990; 10:643-51.
4. Manikoth P, Mangalore GP, Megha V. Axillary Cystic Hygroma. J Postgrad Med. 2004; 50:215–216.
5. A. Gedikbasi, A. Gul, A. Sargin, and Y. Ceylan, Cystic hygroma and lymphangioma: associated findings,perinatal outcome and prognostic factors in live-borninfants, Arch Gynecol Obstet, 276 (2007), 491–498.
6. N. Roozbeh, M. Azizi, L. Darvish, Pregnancy outcome of abnormal nuchal translucency: a systematic review, J. Clin. Diagn. Res. 11 (3) (2017) QC12–QC16.
7. Anderson NG, Kennedy JC. Prognosis in Fetal CysticHygroma. Aust NZ J Obstet Gynaecol. 1992Feb;32(1):36-9.
8. Anteby EY, Yagel S. Route of delivery of fetuses with structural anomalies. Eur J Obstet Gynecol Reprod Biol. 2003 Jan 10;106(1):5-9. doi: 10.1016/s0301-2115(02)00033-7.
9. Hsieh YY, Lee CC, Chang CC, Tsai HD, Hsu TY,Tsai CH. Prenatal sonographic diagnosis of Cantrell's pentalogy with cystic hygroma in the first trimester. J Clin Ultrasound. 1998;26(8):409-12.
10. Entezami M, Albig M, Knoll U, Gasiorek-Wiens A.Chromosomal disorders. In: Entezami M, Albig M,Knoll U, Gasiorek-Wiens A, eds. Ultrasound Diagnosis of Fetal Anomalies. 1st ed. New York:Thieme; 2003: 178-231.
11. Tica OS, Gug C, Tica AA, Busuioc CJ, Amiri S, Tica I, Bică Brăiloiu G, Tica VI. A unique case ofrecurrent fetal cystic hygroma: First fetus with an inherited heteromorphism of chromosome 1(1qh+) and the second fetus with 69 XXX Triploidy. Rom J Morphol Embryol. 2021;61(3):935–40.https://doi.org/10.47162/rjme.61.3.34.
12. de Serres LM, Sie KC, Richardson MA. Lymphatic malformations of the head and neck. A proposal for staging. Arch Otolaryngol Head Neck Surg. 1995 May;121(5):577-82.
13. Puri, R.D. Fetal Dysmorphology. *J. Fetal Med.* **2**, 151–159 (2015). https://doi.org/10.1007/s40556-015-0057-8