International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Kamble V et al. Int J Reprod Contracept Obstet Gynecol. 2014 Sep;3(3):847-850
www.ijrcog.org

pISSN 2320-1770 | eISSN 2320-1789

DOI: 10.5455/2320-1770.ijrcog20140971

Case Report

Cystic hygroma with hydrops fetalis: a rare case report

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Received: 2 August 2014
Accepted: 9 August 2014

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ABSTRACT

Cystic hygromas are malformations of the lymphatics system that appear as fluid-filled, membranous cysts, lined by true epithelium in the anterolateral or occipito-cervical area. They result from the jugular lymphatic obstruction sequence, in which the normal communication between the jugular veins and the jugular lymphatic sacs fail to develop by 40th day of gestation. Most of the cystic hygromas are associated with chromosomal anomalies. When diagnosed in-utero, the survival rate of foetuses affected with cystic hygroma is only 2-6%.When hydrops is present alongwith cystic hygroma, the mortality rate is near 100%. The incidence of cystic hygroma is estimated to be 1 case per 6000-16000 live births. We here present a case of 25 years old primigravida with 16 weeks 5 days of gestation was diagnosed prenatally during ultrasonography for the foetal well-being with a large cystic hygroma with septation extending in the entire length, associated with bilateral pleural effusion and ascites with a variable heart rate. The pregnancy was terminated with the consent of the parents. Foetuses with cystic hygroma are at high risk for adverse outcomes. Due to its extremely poor prognosis, termination should be considered when the diagnosis is made before viability and the chromosomes are abnormal.

Keywords: Cystic hygroma, Hydrops fetalis, Chromosomal abnormalities

INTRODUCTION

Cystic hygromas are malformations of the lymphatics system that appear as fluid-filled, membranous cysts, lined by true epithelium in the anterolateral or occipito-cervical area. The hygroma may be small, simple and transient or large, multiseptated and persistent. They result from the jugular lymphatic obstruction sequence, in which the normal communication between the jugular veins and the jugular lymphatic sacs fail to develop by 40th day of gestation. If a connection between the lymphatic and the venous system does not occur at this point, a progressive peripheral lymphoedema and hydrops develops, leading to early intrauterine death.[1] Cystic hygroma is synonymous with cystic lymphangioma, which is also known as a macrocystic lymphatic malformation and was first described in 1828 by Redenbacker. Most of the cystic hygromas are associated with chromosomal anomalies. When diagnosed in-utero, the survival rate of foetuses affected with cystic hygroma is only 2-6%.[2] The incidence of cystic hygroma is estimated to be 1 case per 6000-16000 live births.

CASE REPORT

A 25 years old primigravida with 16 weeks 5 days of gestation was diagnosed prenatally during ultrasonography for the foetal well-being with a large cystic hygroma with septation extending in the entire length, associated with bilateral pleural effusion and ascites with a variable heart rate. Family history revealed no specific disease or congenital malformation. There was no history of consanguinous marriage or Rh negative pregnancy. The pregnancy was terminated with the consent of the parents after explaining about the

condition and poor outcome of pregnancy. The pregnancy was terminated after induction with misoprostol tablets with a maximum dose of 1200 mcg. Examination of aborted foetus revealed a cystic structure extending posterolaterally along the neck; it was also associated with ascites and foetal oedema.

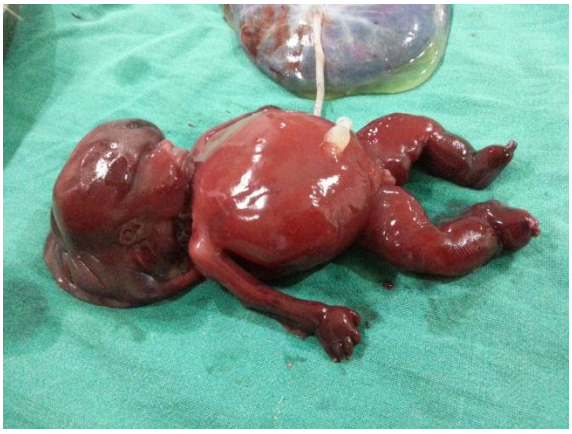

Figure 1: Aborted foetus with cystic hygroma with hydrops fetalis.

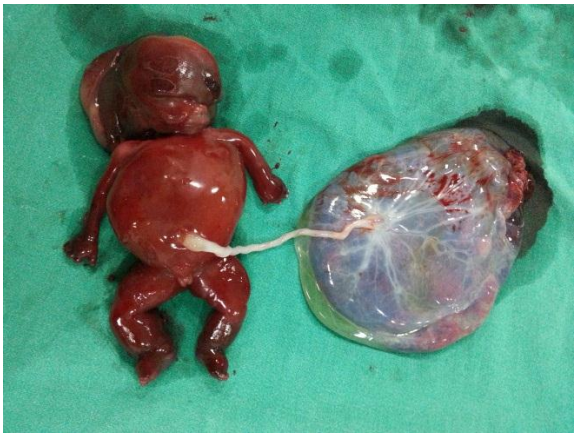

Figure 2: Aborted foetus with cystic hygroma with hydrops fetalis with oedamatous placenta.

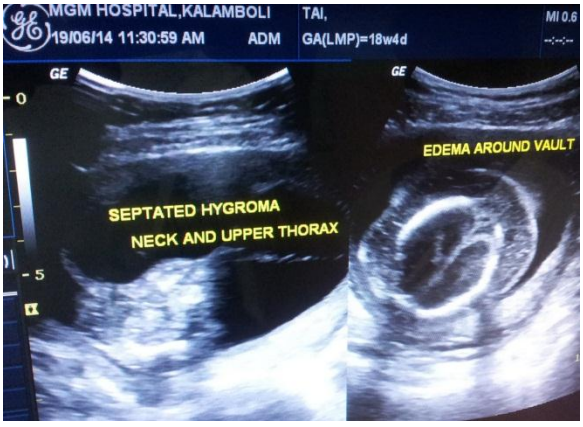

Figure 3: USG showing septated hygroma, edema around the vault.

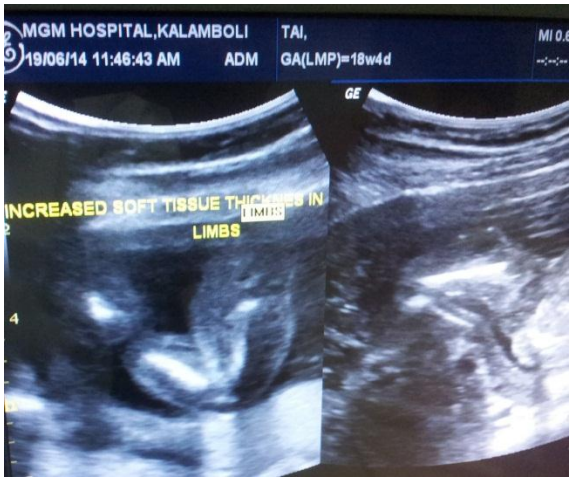

Figure 4: USG showing increased soft tissue thickness in the limb.

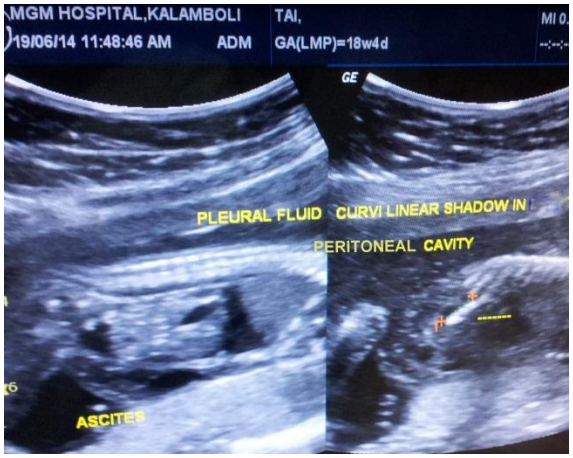

Figure 5: USG showing pleural fluid and ascites.

DISCUSSION

A cystic hygroma can present as congenital or develop at any time during a person's life. A cystic hygroma in a developing baby can progress to hydrops and eventually foetal death. Some cases of congenital cystic hygromas resolve leading to webbed neck, edema, low posterior hair-line. In other instances the hygroma can progress in size to become larger than the fetus.

Pathophysiology

Lymphangiomas are thought to arise from a combination of the following: a failure of lymphatics to connect to the venous system, abnormal budding of lymphatic tissue, and sequestered lymphatic rests that retain their embryonic growth potential. These lymphatic rests can penetrate adjacent structures or dissect along fascial planes and eventually become canalized. These spaces retain their secretions and develop cystic components because of the lack of a venous outflow tract.[3] The nature of the surrounding tissue determines whether the lymphangioma is capillary, cavernous, or cystic.

Location

The most common site of cystic hygroma is neck (nuchal cystic hygroma-80%).[4] Other sites includes axilla with only 10% of cases extending to the mediastinum[5,6] and only 1% confined to the chest.[7]

Associations

Cystic hygromas are associated with chromosomal anomalies in 60-75% of the cases. The most common chromosomal anomaly associated with cystic hyroma is Turner's syndrome.[8] Others include Down's syndrome, Klinefelter's syndrome, and Trisomy 18 and 13. Non-chromosomal disorder, including Noonan syndrome, multiple pterygium syndrome, Pentalogy of cantrell,[9] Fryns syndrome,[10] Apert syndrome, Pena Shoiker syndrome[10] and achondroplasia, are associated with an increased incidence of CH. Intrauterine alcohol exposure has been associated with the development of cystic hygroma. *In utero* exposure to aminopterin and trimethadione has also been reported to be associated with cystic hygroma. Maternal viral infections, such as Parvovirus of Fifth's disease can also be a cause of cystic hygroma.

Cystic hygromas are also sometimes associated with cardiac malformations, most commonly coarctation of the aorta and a hypoplastic left heart. They may also be associated with abdominal, pleural and pericardial effusion.

Staging

de Serres et al.[11] have proposed the following system for staging of CH of the head and neck:

Stage I - Unilateral infrahyoid (17% complication rate)

Stage II - Unilateral suprahyoid (41% complication rate)

Stage III - Unilateral and both infrahyoid and suprahyoid (67% complication rate)

Stage IV - Bilateral suprahyoid (80% complication rate)

Stage V - Bilateral infrahyoid and suprahyoid (100% complication rate)

Diagnosis

- Cystic hygroma can be visualized using abdominal ultrasonography by 10 weeks' gestation, although transvaginal ultrasonography provides superior detail. Ultrasound of cystic hygroma shows a thin walled cystic mass on the posterolateral aspect of the neck. Identification of the nuchal ligament within the cyst constitutes the most specific sign for diagnosis of cystic hygroma. A detailed ultrasound is performed, including foetal echocardiogram, to look for other anomalies that may indicate the cause for the hygroma.[12]
- Fast-spin MRI can also be used to determine the extent of foetal CH.
- Elevated alpha fetoprotein levels in amniocentesis fluid have been reported in pregnancies with CH.[13]
- Amniocentesis or CVS to look for chromosome abnormalities or a specific genetic syndrome may be performed.
- Fluorescent *In Situ* Hybridization (FISH) can be used to evaluate for cystic hygroma in prenatal chromosomal analysis. Chromosomes 13, 18, 21, X, and Y are specifically mentioned.

Management

Successful intrauterine treatment of cystic hygroma colli using OK-432 has been in cases where the cystic hygroma is not associated with any chromosomal abnormality.[14]

Prognosis

Cystic hygroma with a normal chromosome and no other associated anomaly have a spontaneous resolution before 20 weeks of gestation while those associated with chromosomal anomalies have a poor prognosis. Foetal death usually occurs shortly after the diagnosis. Foetal death has been related to chronic foetal hypoxemia secondary to compression of the thoracic structures by the generalised oedema. When hydrops is present alongwith cystic hygroma, the mortality rate is near 100%.[15] The risk of recurrence of cystic hygroma associated with chromosomal abnormalities is around 1%.

Differential diagnosis

1) Encephalocoele.
2) Occipito-cervical meningomyocoele.
3) Nuchal oedema.
4) Rarely posterior teratoma.

CONCLUSION

Foetuses with cystic hygroma are at high risk for adverse outcomes. Due to its extremely poor prognosis, termination should be considered when the diagnosis is made before viability and the chromosomes are abnormal.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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DOI: 10.5455/2320-1770.ijrcog20140971
Cite this article as: Kamble V, Bhatia T, Patil S. Cystic hygroma with hydrops fetalis: a rare case report. Int J Reprod Contracept Obstet Gynecol 2014;3:847-50.