CASE REPORT

Beyond the Norm: A Case Report and Review on Second Trimester Cystic Hygroma with Hydrops Fetalis.

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Submitted: 19/02/2024. Revised edition: 01/04/2024. Accepted: 09/04/2024. Published online: 01/06/2024.

Abstract

A 33-year-old woman spontaneously conceived a foetus with cystic hygroma (CH) and underwent medical termination. The anomaly was detected early via routine ultrasound at a primary health clinic and subsequently referred to tertiary care. Due to financial constraints and the unacceptance of conceiving an abnormal foetus, the parents were not keen on genetic testing via amniocentesis or maternal serum, despite comprehensive genetic counselling by a Fetomaternal specialist. The prognosis of CH is poor and often related to chromosomal and major structural abnormalities. Undiagnosed CH before birth can lead to complications for the mother and foetus. Therefore, early ultrasonographic assessments, conducted by skilled personnel facilitate timely diagnosis. Clinicians must approach counselling with vigilance and take a holistic approach to addressing the patient's factors in declining prenatal genetic screening. Given the possibility of a CH recurrence, offering prenatal genetic screening in future pregnancies is necessary. Therefore, ensuring the nationwide availability of these prenatal genetic screening tests at reduced costs is essential to make them accessible and affordable for all expectant mothers.

Keywords: *cystic hygroma; prenatal genetic screening; ultrasonographic investigation.*

Introduction

A cystic hygroma (CH) is a rare condition of congenital malformation caused by a vascular anomaly related to lymphatic malformations, also known as lymphangiomas. It is characterised by an abnormal accumulation of fluid in the first trimester at the beginning of embryogenesis. CH is primarily detected in the neck region and potentially extends throughout the entire length of the foetus. They can occur in other areas, such as the axilla, mediastinum, abdominal wall, inguinal, and retroperitoneal regions. [1, 2]. Prenatal diagnosis of CH via ultrasound shows bilateral, symmetrical cystic structures, which can be either septated or non-septated. In the first trimester, CH was identified as an increase in nuchal translucency (NT) [1, 3]. The incidence of CH is estimated to be 1 in 6000 to 16000 live births [2]. Foetuses affected by this condition are at higher risk of chromosomal abnormalities and major structural abnormalities. [1]. The overall survival rate for foetal CH is (2-3 %) when diagnosed in utero. [4]. This case study highlights the incidental finding of CH detected during routine antenatal follow-up, where the pregnancy was then terminated.

Case report

A 33-year-old woman, gravida 3, para 2, presented for routine antenatal follow-up at 19 weeks of gestation. She has no significant underlying medical conditions and has had two healthy children. In her previous pregnancy, in 2019, she had a lower caesarean section due to a failed vacuum-assisted delivery. Otherwise, she is not in a consanguineous marriage and has no family history of congenital anomalies.

Her first antenatal booking at 14 weeks of gestation included a dating transabdominal scan, which revealed a normal foetus with parameters corresponding to 13 weeks and 5 days, along with visible cardiac activity. Her blood investigations, vital signs, and clinical examinations were all normal.

She was advised to return for a routine antenatal follow-up scan at 19 weeks of gestation. The ultrasonographic examination revealed a singleton-lived foetus with a large CH located at the posterior of the neck. She was referred to a Fetomaternal specialist in a tertiary hospital immediately. The ultrasound findings showed a singleton foetus having cardiac activity [heart rate (HR)] of 165 beats per minute (bpm). The foetal biometry was compatible with 21 weeks. The foetus exhibited generalised subcutaneous oedema and presented with a large anechoic, thin-walled cyst, multiloculated with internal septations situated predominantly on the posterior aspect of the neck, measuring 16 x 5 cm (Figure 1), with no obvious internal flow. (Figure 3). Additionally, bilateral pleural effusions and ascites were observed. (Figure 2). The thalamus was fused, while the heart, spine, and kidneys appeared normal, with a single gastric bubble seen. The deepest vertical pocket of amniotic fluid measured 3.9 cm, and the placenta appeared normal. The diagnosis of CH with hydrops fetalis was established by the Fetomaternal specialist, who informed the parents of the foetus's overall findings, suggesting potential genetic issues and the very poor foetal prognosis of this abnormality. Genetic counselling, recommendations for amniocentesis, and screening maternal blood tests for genetic confirmation were advised by the Fetomaternal specialist. However, due to financial constraints and their unacceptance of conceiving an abnormal foetus, the parents were not inclined to proceed. After further comprehensive counselling, the parents ultimately decided on termination of the pregnancy. She was admitted to the ward at 19 weeks and 3 days and induced with a gemeprost pessary with a total dose of 1000mg. She subsequently aborted the foetus at 19 weeks and 5 days without any surgical intervention. A general examination of the aborted foetus revealed a cystic structure measuring 10 x 12 cm located at the posterior part of the neck, extending to the entire length of the foetus, associated with generalised oedema. Placental histopathological examinations and other blood investigations for

genetic studies were not sent as parents were not keen.

Discussion

Cystic hygromas are slow-growing benign tumours caused by malformation of the lymphatic system. Nuchal CH is characterised by increased NT of ≥ 3mm at the neck region, with or without septation or multilocular structures in other regions. NT measurements greater than 6.5 mm have a higher prevalence (74.2%) of abnormal karyotypes, while (25 %) miscarried and (41.7 %) had electively terminated the pregnancy. [1-3].

CH has a poor prognosis, with an unfavourable outcome observed in 77.8 %, It is often associated with chromosomal abnormalities, occurring in 51% of cases, with non-septated CH being more common at 61.4% and primarily linked to Down syndrome. Conversely, septated lesions are frequently related to Turner's syndrome (21%) and tend to have a poorer prognosis. Additionally, other genetic conditions, such as Noonan syndrome, multiple-pterygium syndrome, Fryns syndrome, and Neu-Laxova syndrome, are frequently reported in association with CH. Furthermore, CH is linked to non-chromosomal genetic factors, including major structural abnormalities such as ascites, pleural effusions, cardiac abnormalities, non-immune hydrops fetalis and intrauterine foetal demise. [1, 2, 5].

The recurrence of CH in future pregnancies is very rare. However, CH with a normal karyotype can be inherited as an autosomal recessive trait. [6] Undiagnosed CH before birth can lead to complications for both the mother and foetus, including shoulder dystocia and uterine rupture, especially when lymphangiomas are situated at the trunk. Meanwhile, lymphangiomas located at the anterior of the neck may compress the airway of the foetus, necessitating neonatal resuscitation. [1] A CH case involving a patient with spontaneous triploid conception that progressed into second trimester and accompanied with maternal complications of preeclampsia and HELLP syndrome has been reported. [7] This case was managed with medical termination of pregnancy due to foetal abnormalities and severe maternal preeclampsia. [7]

All pregnant patients should undergo a second-trimester detailed ultrasound, ideally at 18–22 weeks of gestation, to detect foetal structural defects. Additionally, prenatal genetic screening should be offered to all expectant mothers, regardless of maternal age. In cases where screening tests indicate foetal aneuploidy, patients should receive genetic counselling, detailed ultrasound evaluation, and invasive diagnostic testing for confirmation. The non-invasive screening test obtained from maternal serum includes: 1) evaluation of cell-free DNA to screen for trisomy 21,18,13 and sex chromosome aneuploidies. 2) evaluation for specific biomarkers associated with trisomy 21 and 18 (with or without assessment of specific ultrasound markers). Diagnostic invasive testing, such as chorionic villus sampling (CVS) or amniocentesis, is offered to patients with a structural foetal abnormality and positive genetic screening. [8, 9]

Malaysia lacks a nationwide maternal serum screening programme, making genetic testing financially inaccessible for some patients. This lack of access to genetic testing challenges the early detection and management of foetal anomalies during pregnancy. Moral and religious concerns may cause stigmatisation, and positive screening outcomes may impact emotional well-being [10]. Addressing these factors requires education, counselling, and support services. Early antenatal diagnosis and proper management are crucial for better maternal outcomes.

What distinguishes this case report from prior literature?

In our case we have emphasised the importance of early detection, and performing anomaly ultrasound in primary care is crucial to providing timely interventions.

2) The importance of clinicians educating and counseling patients on genetic testing options to prevent stigmatisation.

How does this benefit the patient?

Cystic hygroma with hydrops fetalis has a poor prognosis. Considering termination of pregnancy as an option for this pregnancy may help prevent complications for the mother. Additionally, in future pregnancies, serial ultrasound scans, detailed anomaly screening, and genetic testing can be conducted to screen for any potential recurrence. This can guide decision-making and management of the patient's condition, ensuring the possible outcome.

Acknowledgement

The authors wish to express their gratitude to the patient for granting permission and participating in the development of this case report.

Conflict of interest

There is nothing to declare.

Patients' consent for the use of images and content for publication

The patient gave verbal permission for the images and case to be used for publication.

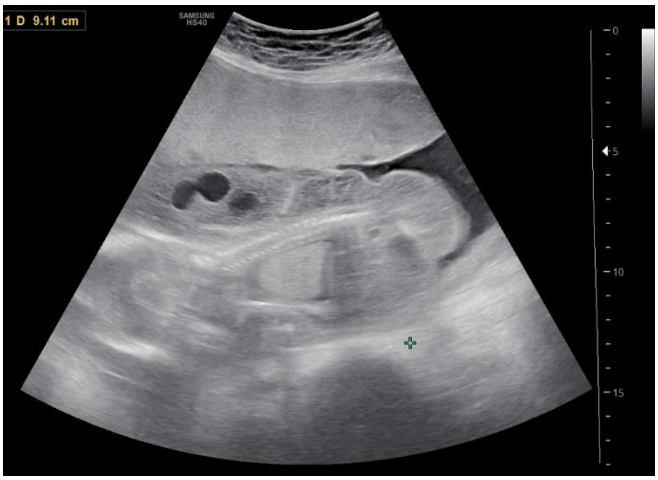

Figure 1. Sagittal view: Posterior multiloculated subcutaneous cystic neck mass measured (10 x12) cm extending the entire length of the foetus with generalised subcutaneous oedema

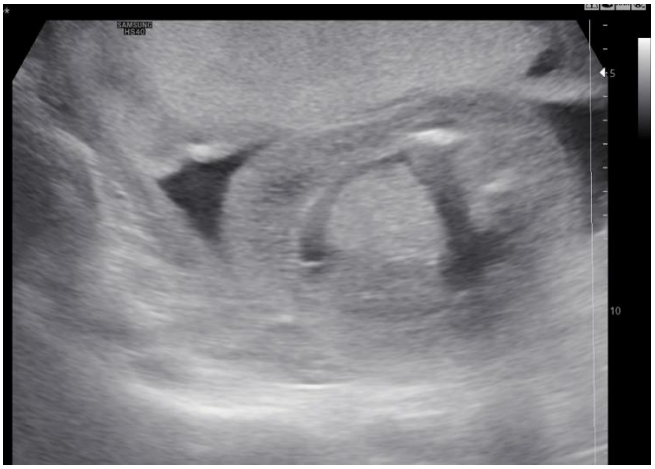

Figure 2. Axial thoracic view: Bilateral pleural effusion of foetus

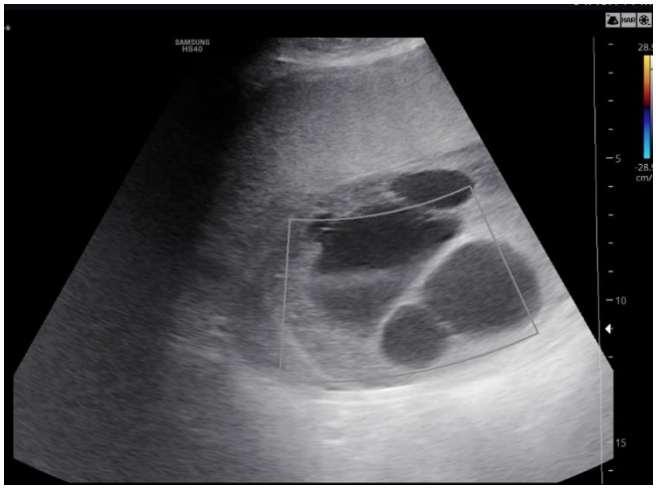

Figure 3. Sagittal view of the cystic mass at the neck: Colour Doppler ultrasound revealed a multiloculated cyst with internal septations and showed no internal flow.

foetal heart rate with no episodes of foetal bradycardia observed.

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