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Bilateral Cystic Hygroma with Gross Polyhydramnios and Increased Nuchal Thickness - A Rare Mid-Trimester Ultrasonographic Presentation

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Abstract

Cystic hygromas, referred to as cysts filled with fluid that develop due to abnormalities in the lymphatic system, often appear in the occipito-cervical or anterolateral area. The presence of these variations is attributed to the jugular lymphatic obstruction sequence, it occurs when the normal connect within the jugular lymphatic sacs jugular veins and does not form by the 40th day of pregnancy. Cystic hygromas, often linked to chromosomal disorders, provide a difficult prognosis. The survival rate for patients identified by prenatal testing is around 2-6%. However, if hydrops interacts with cystic hygroma, the possibility of fatality exceeds 100%. The prevalence of cystic hygroma is around 1 case per (6000-16000) live births. The study highlights a particular instance of a 20-year-old woman who became pregnant for the first time without any medical intervention. She worked as a sewing worker and did not use folic acid before or throughout the first three months of her pregnancy. Ultrasonography conducted at 16 weeks and 4 days detected a viable foetus inside the uterus with cystic hygroma, polyhydramnios, and other abnormalities. Although they received counselling about amniocentesis, the parents chose to terminate the pregnancy for medical reasons. This provided information on foetal anomalies such as cystic hygroma, occipital encephalocele, and spina bifida occulta. After the termination, karyotyping revealed Turner syndrome (45X).

Keywords: Cystic Hygromas, Lymphatic System Malformations, Fetal Anomalies, Medical Termination of Pregnancy

Introduction

Cystic hygromas are characterised by the presence of fluid-filled cysts in the lymphatic system. These cysts could be observed in the front-lateral or back-neck area and exhibit different forms.[1] The anomalies exhibit a range of sizes, ranging from small and transient forms to large and persistent ones, with several compartments.[2] Jugular lymphatic blockage sequence impedes the formation of jugular veins and lymphatic sacs, resulting in the inability to establish normal connection between them through the 40th day of gestation.[3] Absence of the lymphatic-venous connection could result in the emergence of advancing peripheral lymphedema and hydrops, potentially leading to premature intrauterine demise.[4] These malformations, also known as lymphangioma,[5] are often linked to chromosomal abnormalities, resulting in an inadequate survival rate of 2-6% when detected during pregnancy.[6]

Lymphangioma is generally recognised as a paediatric condition characterised by the proliferation of lymphatic vessels.[7] Cystic hygromas are fluid-filled sacs that are often seen in the neck area. A cystic hygroma may manifest as a congenital birth abnormality or arise at any point in an individual's lifespan.[8] The frequency of expression in adults is exceptional and its precise cause is unidentified,[9] however trauma and upper "respiratory tract" infection have both been implicated as potential precipitants for

its initiation.[10]

Diagnosing adults is often more difficult than diagnosing children, and the ultimate diagnosis is typically determined by examining the tissue after surgery.

The diagnosis technique for cystic hygromas typically includes a blend of clinical assessment, imaging examinations, and, often, postoperative histology.[11] While imaging tools like ultrasound and MRI may help diagnose cystic hygromas in children, adults may need surgery to confirm the diagnosis.[12] Postoperative histology involves the analysis of tissue samples collected following surgery to verify the existence of cystic hygromas and gain more understanding of the characteristics of the disorder.[13]

The prevalence of cystic hygroma is quite low, with an estimated incidence of roughly 1 case per 6000-16000 live births.[14] The phrase "live births" denotes the count of babies that are delivered and survive outside the womb. It is important to mention that cystic hygromas are also linked to a higher incidence of spontaneous abortions, indicating a larger occurrence in pregnancies that result in miscarriage.[15]

These sacs stored with fluid usually have a thickness of 3 mm or less. Cystic hygromas may exhibit variations in size and location, however they are mostly seen in the neck and axillary areas.[16] Nuchal translucency is the measurement of the thickness of a particular region at the posterior of the foetal neck. An elevated nuchal translucency is one of the indicators that may be detected during ultrasound scans in the first trimester.[17]

Cystic hygromas could occur in any population, however their occurrence during pregnancy is rather rare.[18] Identifying cystic hygroma during pregnancy may lead to further diagnostic assessments and counselling to evaluate any related illnesses or problems.[19] The management and treatment choices are contingent upon many aspects, such as the severity of the ailment and its effect on the individual's health. Healthcare practitioners must offer thorough information and assistance to people and families impacted by cystic hygroma.[20]

Case Study

A 20-year-old primigravida, spontaneously conceived, presented at 16 weeks and 4 days gestation to the obstetrics outpatient department at HIMS, Varanasi. Her last menstrual period was on 20-11-2022, with an expected due date of 27-08-2023. Employed as a sewing worker, she had no history of folic acid intake before or during the first trimester. Medical, surgical, and family history were unremarkable, with no addiction history. Basic antenatal care investigations were normal. On examination, her abdomen was distended, and the uterus was enlarged to 24-26 weeks, relaxed, with a present foetal heart sound. Ultrasonography revealed a single live intrauterine fetus of 16 weeks and 4 days with cephalic presentation, gross polyhydramnios (AFI=32.31), Increased Nuchal fold Thickness(2.22cm) and bilateral cystic swelling in the neck (2.11*2.32cm and 2.13*1.84cm) with internal echoes (**Figure 1**). The patient was counselled and offered amniocentesis for fetal karyotyping, but she refused. Due to the associated risks, the parents opted for medical termination of pregnancy, which was completed without complications. Gross examination of the fetus revealed bilateral neck swelling (cystic hygroma), a swelling over the posterior occiput (occipital encephalocele), and spina bifida occulta (**Figure 2**). Fetal tissue karyotyping post-termination revealed 45X (Turner syndrome).

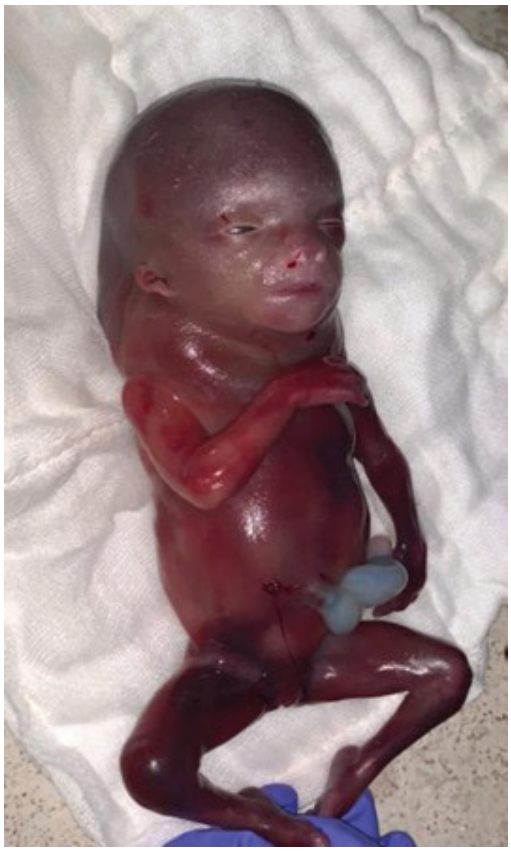

Figure 1

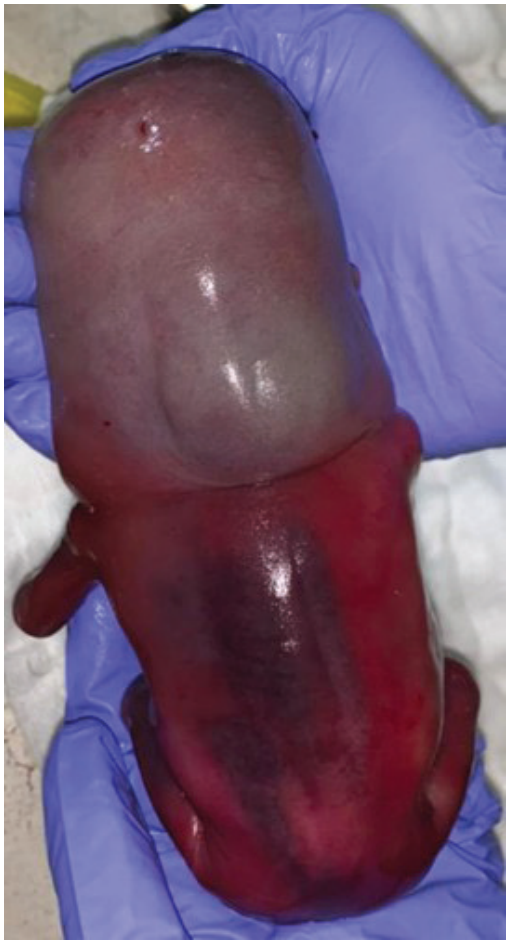

Figure 2

Discussions

Cystic hygromas are abnormalities in the development of the lymphatic system, which is an intricate network of delicate vessels responsible for transporting tissue fluid to the venous system.[21] These lesions are identified by the presence of one or more fluid-filled sacs that develop at locations where the lymphatic and venous systems link, often seen at the back of the neck.[22] Cystic hygromas often arise during the latter part of the first trimester or early in the second trimester,[23] and their occurrence decreases as pregnancy advances. Lymphedema is caused by the buildup of fluid in enlarged lymphatic vessels and the surrounding tissue, resulting in swelling. Nonimmune hydrops often accompanies this condition.[24] In the present report, a case study of a 20-year-old woman in her first pregnancy, exhibiting an intrauterine foetus with cephalic presentation, significant polyhydramnios, and bilateral cystic neck swellings. Following pregnancy termination, gross examination revealed bilateral neck swelling (cystic hygroma), occipital encephalocele, and spina bifida occulta. Foetal tissue karyotyping confirmed Turner syndrome (45X), emphasizing the need for thorough evaluation and counselling in complex prenatal situations. It underscores the challenging decisions faced by both healthcare providers and parents, highlighting the importance of a comprehensive and compassionate approach in managing reproductive health challenges.

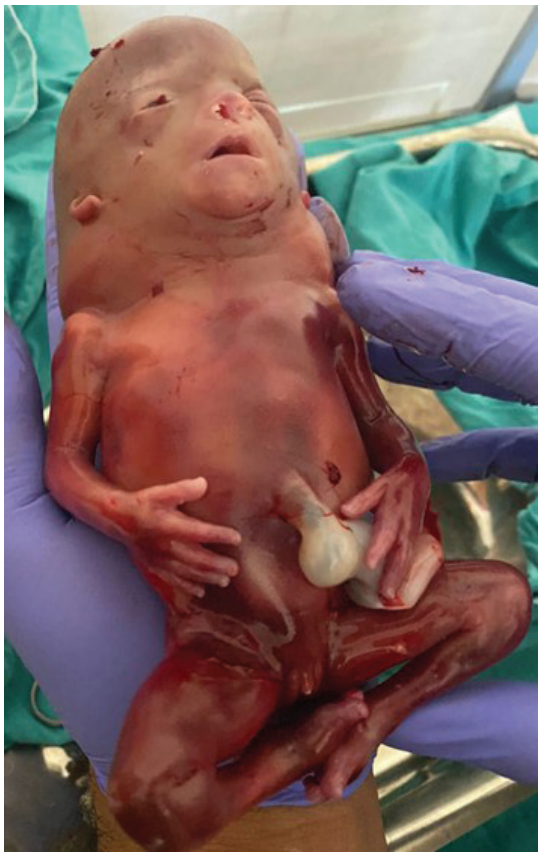

Figure 3

In a previous research Chen et al., (2017) examined the specific characteristics of nuchal cystic hygroma. The findings indicated that this condition presents as a translucent area in the soft tissue of the posterior part of the head, comprising of two identical cavities that are entirely divided by a central partition. These cysts may or may not have interior trabeculae, appearing as cysts with several compartments. The research defines nuchal translucency (NT) as the existence of a single-chambered accumulation of fluid in the nuchal region that measures 3 cubic millimetres or more.[25]

In a similar way, Guiglia and Rosati[26] revealed that 63.2% of non-septated cystic hygromas and 28.6% of septated cystic hygromas reduced during pregnancy, compared to nonseptated cystic hygromas, those with septated cysts are more probable to have aneuploidy. Turner syndrome was exclusively detected in cases with septated hygromas, accounting for 30 out of 39 cases.[27] On the other hand, Trisomy 21 was the most often seen aberrant chromosomal arrangement in cases with nonseptated hygromas, representing 5 out of 16 cases. The prevalence of hydrops fetalis was higher in cases with "septated cystic hygromas" compared to "nonseptated cystic hygromas", with rates of 60% and 19% respectively. Additionally, the survival rates were greater in "nonseptated cystic hygromas" (27%) compared to "septated cystic hygromas" (2%).[28]

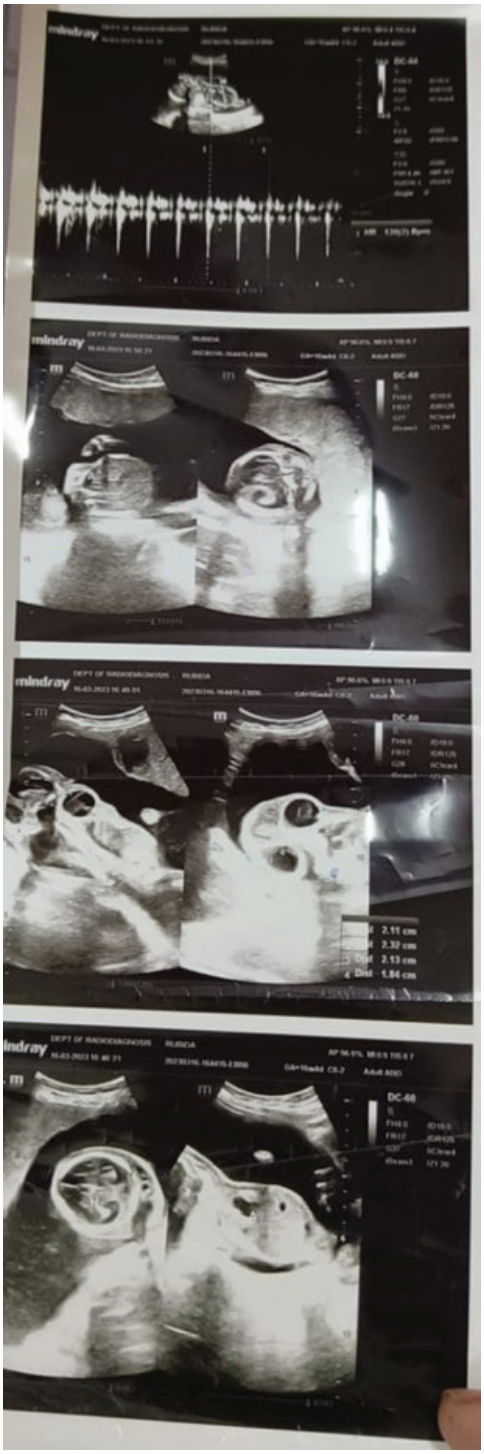

Figure 4

This case highlights the significance of thorough prenatal screening and counselling. The involvement of a diverse healthcare team, consisting of obstetricians, genetic counsellors, and maternal-foetal medicine experts, is essential for managing the intricate choices and offering assistance to the parents. Moreover, it emphasises the need of continuous study and medical progress in order to expand our comprehension of these illnesses and provide suitable and prompt therapies.

Conclusion

In conclusion, the existence of bilateral cystic hygroma, significant polyhydramnios, and heightened nuchal translucency at 16 weeks of gestation gives rise to apprehensions over possible foetal abnormalities, necessitating the need for genetic assessment of both the foetus and parents. The correlation between cystic hygromas and aneuploidy underscores the need of comprehensive diagnostic methods, such as amniocentesis, for foetal karyotyping. Close monitoring is necessary because structural problems may become apparent at a later stage of pregnancy, necessitating careful observation. In circumstances when successive scans show no further abnormalities, selective continuation of pregnancy may be explored as an alternative to medical termination of pregnancy. This highlights the intricacy of handling such instances, requiring a careful equilibrium between diagnostic treatments and continuous assistance for the parents throughout the process of making alternatives.

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