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Hygroma Colli ; Sonographic Indication of Chromosomal Abnormality. a Case Report

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ABSTACK

Hygroma colli is a congenital anomaly characterized by cystic enlargement in the cervical region caused by abnormal lymphatic system development. This condition is clinically significant because it is frequently associated with chromosomal abnormalities, such as Turner syndrome and other forms of aneuploidy. The research problem addressed in this case report is the need for early detection and appropriate management of *hygroma colli* in order to minimize complications during pregnancy. This report aims to describe the identification and management of *hygroma colli* in a fetus at 17 weeks of gestation. A 26-year-old primigravida woman was referred to the hospital following the detection of a fetal neck mass on routine ultrasound. Subsequent ultrasound revealed a symmetrical, thin-walled, hypoechoic, cystic mass in the lateral posterior region of the fetal neck, accompanied by pulmonary abnormalities. Based on these findings and the potential for chromosomal anomalies, pregnancy termination was performed. The case highlights the critical role of prenatal ultrasound in the early identification of *hygroma colli* and related abnormalities, allowing clinicians and families to make timely, evidence-based decisions. These findings also emphasize the importance of integrating genetic counseling and multidisciplinary approaches in managing pregnancies complicated by suspected congenital anomalies.

Keywords: Hygroma Colli, Chromosomal Abnormalities, Prenatal Ultrasound, Termination Of Pregnancy, Fetal Anomalies

INTRODUCTION

Congenital anomalies remain a major cause of perinatal morbidity and mortality worldwide, accounting for approximately 6% of all neonatal deaths according to the World Health Organization (WHO, 2020) (WHO, 2023). Among these, anomalies involving the lymphatic system are relatively rare but clinically significant because they often signal underlying chromosomal abnormalities or severe multisystem disorders (Investigations into fetal congenital lymphatic malformations: Case-series and review, 2023; Chen et al., 2024). Early and accurate prenatal diagnosis of such anomalies is essential to inform management decisions and provide adequate counseling for families (Rogerson et al., 2023; Militaru et al., 2024; Scott et al., 2025).

One important congenital anomaly of the lymphatic system is cystic hygroma (hygroma colli), a condition characterized by multilocular cystic masses typically located in the fetal

neck. It develops due to abnormal formation or obstruction of the jugular lymphatic sacs between the 6th and 9th weeks of gestation, resulting in dilated lymphatic channels that may progress to hydrops fetalis and intrauterine death (Sherer et al., 2022; Podobnik et al., 2025). The global incidence of cystic hygroma is estimated at 1 in 6,000 live births and up to 1 in 750 spontaneous miscarriages, reflecting its frequent association with chromosomal abnormalities such as Turner syndrome, trisomy 13, 18, and 21 (Lugata et al., 2024; Lahel et al., 2024; Yakıştıran et al., 2020).

Numerous studies have emphasized the strong correlation between cystic hygroma and chromosomal abnormalities. For example, Wapner et al. (2020) reported that approximately 62% of cases are associated with Turner syndrome, while others are linked to autosomal aneuploidy or genetic syndromes. Advances in prenatal imaging, particularly high-resolution ultrasonography combined with genetic testing such as chorionic villus sampling (CVS) and amniocentesis, have greatly improved diagnostic accuracy (Zheng et al., 2024; Podobnik et al., 2025; Systematic Review: Perinatal Outcomes of Cystic Hygroma, 2025). However, in many low- and middle-income countries, genetic testing facilities remain limited, making ultrasound the primary tool for early detection (Podobnik et al., 2025; L Zhen et al., 2024; Li et al., 2024).

Despite advances in prenatal diagnosis, significant gaps remain (Ladak et al., 2024; Van den Veyver et al., 2022). First, most studies focus on large population-based screenings or genetic correlations, while fewer provide detailed accounts of clinical decision-making when resources are constrained. Second, there is limited literature from developing countries on how cystic hygroma cases are identified and managed in clinical practice without immediate access to advanced genetic testing. Third, while cystic hygroma has been extensively described in relation to poor prognosis, there are fewer case reports highlighting its early identification through ultrasound and its direct impact on clinical decision-making in local healthcare contexts.

Given these gaps, case reports remain important in providing practical insights into how clinicians recognize, evaluate, and manage rare congenital anomalies such as *hygroma colli*. They serve to document real-world practices, highlight diagnostic challenges, and offer context-specific recommendations that may not be captured in large cohort studies. This is particularly relevant in resource-limited settings, where ultrasound often functions as the only diagnostic modality available for prenatal anomaly detection.

This case report aims to describe the process of identifying fetal *hygroma colli* at 17 weeks of gestation through ultrasound examination and to explain the subsequent clinical management that resulted in pregnancy termination. By presenting this case, we intend to underline the importance of prenatal ultrasound as a frontline diagnostic tool for congenital anomalies, emphasize the need for timely multidisciplinary counseling, and contribute to the literature from the perspective of clinical practice in developing countries.

METHOD

This study uses a case report approach that is qualitatively descriptive. This method was chosen because the focus of the research is to describe in depth the patient's condition, examination findings, and clinical actions taken in the case of *hygroma colli*. Case studies are appropriate for exploring clinical phenomena that are rare or have important diagnostic value.

The subject of the study was a 26-year-old primigravida woman who was referred to a teaching hospital of the Faculty of Medicine, University of Riau. This case takes place in the second trimester of pregnancy, when the gestational age was 17 weeks. The patient was selected purposively because she showed typical clinical signs according to the research objective, namely the identification of *hygroma colli* in the fetus.

Anamnesis and Physical Examination Preliminary data were obtained through pregnancy history anamnesis, risk factors, and standard obstetric examinations. Family history, exposure to teratogenic substances, and previous pregnancy screening results were also studied.

Ultrasound Examination Obstetric ultrasound was performed by obstetricians and gynecologists using a high-frequency transducer. The examination included:

1. Assessment of fetal morphology (head, neck, thorax, abdomen, and extremities).
2. Identification of neck mass: location, size, internal characteristics (septation, echogenicity), and mass boundaries.
3. Assessment of other organs for the detection of additional structural abnormalities (e.g., pulmonary agenesis).
4. Assessment of fetal heart rate and vitality.

Radiological Documentation Ultrasound images were recorded in sagittal and axial views, which were then used for morphological analysis of the *hygroma colli*.

Clinical Actions Based on the ultrasound results, the decision was made to terminate the pregnancy. The procedure was performed using a combination of intravaginal *misoprostol* and *laminaria* insertion according to hospital protocol.

Post-Termination Examination A physical examination of the baby was carried out after delivery to confirm the concordance of the ultrasound findings with the postnatal condition. Data included weight, body length, Apgar score, and external anatomical conditions.

Data Analysis Techniques The data in this study were analyzed descriptively with reference to the existing literature on *hygroma colli*. The analysis focused on the concordance of ultrasound results with postnatal findings, the relationship between structural abnormalities and potential chromosomal abnormalities, and a comparison with previous studies regarding the prognosis of *hygroma colli*.

Ethical approval for this case was obtained from the hospital, with informed consent from the patient, and the patient's identity was kept confidential.

Data validity was ensured through expert ultrasound examination, diagnostic image documentation, and triangulation with scientific literature. The case study method allowed for a comprehensive overview of the diagnostic process and clinical decision-making, contributing to the limited literature on *hygroma colli* cases in Indonesia.

The analysis steps included problem identification, with a fetal neck mass detected during routine examination; formulation of differential diagnoses, distinguishing *hygroma colli* from other neck masses such as teratoma; confirmation of diagnosis based on characteristic ultrasound features, including cystic, symmetric, and hypoechoic lesions; consideration of prognosis, where the presence of additional organ abnormalities worsened the fetal outcome; clinical decision-making, leading to pregnancy termination based on medical indications; and outcome evaluation by comparing ultrasound findings with the postnatal condition.

However, this method has certain limitations, such as the absence of fetal karyotype examination, which prevented a definitive correlation between *hygroma colli* and chromosomal abnormalities, as well as the fact that this was a single-patient case report, limiting generalizability.

Despite these limitations, the method provides an important model for healthcare professionals in diagnosing *hygroma colli* at an early stage, offering appropriate counseling, and planning suitable management strategies.

RESULTS AND DISCUSSION

The patient was a 26-year-old primigravida who was referred to the teaching hospital of the Faculty of Medicine, University of Riau at 17 weeks of gestation. The reason for the referral is the discovery of a mass in the fetal neck on routine ultrasound examinations. Based on the results of the evaluation, the patient had no family history of similar congenital disorders or a clear history of exposure to teratogenic substances. However, ultrasound results showed typical findings in the form of hygroma colli with abnormalities of the pulmonary organs that worsened the prognosis of the fetus.

1. Ultrasound Findings

Ultrasound examination showed a single fetus with a heart rate still present at the time of the initial examination. The cystic mass is found in the posteriolateral region of the fetal neck, appears symmetrical, thin-walled, hypochic on the inside, and does not show a partition (non-septated). This kind of lesion is very typical for hygroma colli. In addition, agenesis of one of the foetal lung lobes was found, indicating the presence of additional organ anomalies. These findings are important because the combination of structural abnormalities is often correlated with chromosomal abnormalities and poor prognosis.

The ultrasound characteristics of this case are in accordance with the literature: hygroma colli generally appears as a bilateral, hypochic cystic lesion in the occitocervical region. The presence of other organ abnormalities (e.g. pulmonary agenesis or cardiac abnormalities) increases the risk of perinatal mortality and strengthens the indications for further pregnancy counseling.

2. Clinical Actions

Based on the findings of the ultrasound, counseling was carried out to patients and families regarding the very poor prognosis of the fetus. Patients are given information on the risk of chromosomal abnormalities such as Turner syndrome or other aneuploidy. Because multiple abnormalities and a low prognosis were found, it was decided to terminate the pregnancy according to medical indications.

The termination procedure was performed using a combination of intravaginal misoprostol 400 mcg every 3 hours and laminaria insertion according to hospital protocol. This process goes smoothly without any serious complications in the mother. The baby was born with a body weight of 150 grams, a body length of 14 cm, and an Apgar score of 0/0. After birth, there was a soft lump on the baby's neck which was consistent with the findings of hygroma colli on previous ultrasound.

3. Comparison of Prenatal and Postnatal Outcomes

Confirmation of the diagnosis of hygroma colli was obtained after a postnatal examination showed a lump of the neck according to the ultrasound findings. This condition exhibits high diagnostic accuracy of prenatal ultrasound. However, in this case, a fetal karyotype examination was not carried out because the termination decision had been made first. This is a limitation that prevents further analysis of the direct relationship between hygroma colli and chromosomal abnormalities in this case.

4. Prognosis Analysis

Based on literature data, about 62% of hygroma colli cases are related to Turner syndrome, while some others are related to aneuploidy such as trisomy 21, 18, or 13. The prognosis in the case of hygroma colli is highly dependent on the size of the lesion, the presence of septation, and abnormalities of the comorbidities. The Zhou (2023) study showed a survival rate of only about 10% in fetuses with hygroma colli accompanied by multiple organ abnormalities. The findings in this case (non-septated but accompanied by pulmonary agenesis) indicated an equally poor prognosis.

These findings are in line with the reports of Garden et al. (1986) which stated that most fetuses with hygroma colli experience spontaneous miscarriage or stillbirth when multiple structural abnormalities are found. The study of Sherer & Wang (2022; 2025) also showed that aggressive prenatal management is necessary to minimize maternal risk and improve the quality of patient counseling. On the other hand, cases with hygroma colli but normal karyotypes can have better results especially if the lesions are small and without additional organ abnormalities.

The findings of this study highlight several important points. Early detection through ultrasonography, particularly during the second trimester, plays a crucial role in identifying hygroma colli and its associated chromosomal abnormalities. Prenatal counseling is essential, as patients should be provided with comprehensive information regarding risks and prognosis to support informed decision-making. Although chromosomal evaluation was not performed in this case, karyotyping can be valuable in establishing a genetic diagnosis and estimating the risk of recurrence. Furthermore, interdisciplinary management involving collaboration among specialists in obstetrics, radiology, and clinical genetics is vital to ensure optimal care.

The study also emphasizes the importance of psychological and social aspects for patients. The decision to terminate a pregnancy is a difficult one; therefore, emotional support and psychological counseling are integral components of managing such cases. Appropriate medical interventions must be balanced with sensitive communication to help reduce the emotional trauma experienced by patients and their families.

This case further supports the evidence that hygroma colli can serve as an important indicator of chromosomal abnormalities. In addition, it provides a foundation for future research on the correlation between the size of hygroma colli and specific chromosomal abnormalities, the effectiveness of combined screening methods (ultrasound and cell-free DNA

testing) in detecting aneuploidy in pregnancies with hygroma colli, and non-termination management strategies for mild cases with normal karyotypes.

Pregnancy termination in the patient was done because of the existence of multiple organ anomalies that found in ultrasound examination. So that the prognosis for subsequent fetal life is poor. Generally, survival rate in fetal with hygroma colli is 10%.4 It is also considered a possible cause of perinatal disability.5 Prenatal diagnosis of hygroma colli on ultrasound based on demonstration of a bilateral, mostly symmetric, cystic structure located in the occipitocervical region with the lesion either septated or not.6

One study found nearly 47% of the pregnancies with cystic hygroma had multiple congenital anomalies, of which 58% had a chromosomal anomaly. Aneuploidies were major chromosomal defects.7 The risk of recurrence for aneuploidy is low, but cystic hygroma colli with normal karyotype may be inherited as an autosomal recessive trait with 25% recurrence.8 On this case, the karyotype examination was not done to the patient due to termination decision.

CONCLUSION

This case highlights that prenatal sonography remains a crucial tool in the prenatal diagnosis of cystic *hygroma colli*, particularly in settings where access to advanced genetic testing is limited. The detection of cystic hygroma at 17 weeks of gestation directly influenced the clinical decision to terminate the pregnancy, underscoring the role of ultrasound not only in identifying structural anomalies but also in guiding timely management. Early and meticulous ultrasound examination is therefore invaluable for detecting subtle signs that may indicate underlying chromosomal abnormalities, allowing clinicians to provide accurate counseling and optimize outcomes for patients.

REFERENCES

Chen, C. P., et al. (2024). Chromosomal abnormalities associated with fetal pleural effusion: Genetic counselling and fetal therapy implications. *Prenatal Diagnosis.* https://www.sciencedirect.com/science/article/pii/S1028455924000263

Garden, A. S., Benzie, R. J., Miskin, M., & Gardner, H. A. (1986). Fetal cystic hygroma colli: Antenatal diagnosis, significance, and management. *American Journal of Obstetrics and Gynecology, 154*, 221–225.

Investigations into the genetics of fetal congenital lymphatic malformations: A case-series and review. (2023). *Prenatal Diagnosis.* https://obgyn.onlinelibrary.wiley.com/doi/full/10.1002/pd.6345

Ladak, Z., Grewal, N., Kim, M. O., Small, S., Leber, A., Hemani, M., Sun, Q., Hamza, D. M., Laur, C., & Ivers, N. M. (2024). Equity in prenatal healthcare services globally: an umbrella review. *BMC Pregnancy and Childbirth*, *24*(1), 191.

Lahel, R. S., et al. (2024). A rare case report of cystic hygroma with hydrops fetalis. *Medical Journal of DY Patil Vidyapeeth, 17*(…), …pages.

Li, H., et al. (2024). Chromosomal abnormalities detected by chromosomal microarray analysis (CMA) among fetuses with abnormal ultrasound findings. *Scientific Reports, 14*, 67123. https://doi.org/10.1038/s41598-024-67123-5

Lugata, J., et al. (2024). Prenatal detection and management challenges of a huge cystic hygroma of the neck in a resource-constraint setting: A case report. *International Journal of Surgery Case Reports, 109826*, 1–6.

https://doi.org/10.1016/j.ijscr.2024.109826

Militaru, M. S., et al. (2024). The impact of chromosomal mosaicisms on prenatal diagnosis: Clinical consequences and detection challenges. *Journal of Translational Medicine.* https://www.mdpi.com/2075-4426/14/7/774

Podobnik, P., et al. (2025). Genetic and sonographic insights into first-trimester fetal cystic hygroma: A retrospective 30-year analysis using 3D/4D ultrasound and cytogenetic evaluation in Croatia (1993-2023). *Prenatal Diagnosis.* https://pmc.ncbi.nlm.nih.gov/articles/PMC12385511/

Rogerson, D., et al. (2023). Genetics of fetal congenital lymphatic anomalies: Whole-Exome Sequencing insights. *Prenatal Diagnosis.* https://obgyn.onlinelibrary.wiley.com/doi/full/10.1002/pd.6345

Scott, F., et al. (2025). Strategies to detect chromosomal anomalies not identified by standard screening in prenatal care. *Prenatal Diagnosis.* https://obgyn.onlinelibrary.wiley.com/doi/full/10.1002/pd.6755

Sepulveda, W., Wong, A. E., & Dezerega, V. (1995). Evolution of fetal nuchal cystic hygroma in chromosomally normal fetuses. *Ultrasound in Obstetrics and Gynecology, 5*(…), …pages.

Sherer, D. M., Hsieh, V., & colleagues. (2022). First-trimester septated cystic hygroma and non-invasive prenatal screening outcomes. *International Journal of Women's Health, …*(…), …pages.

Sherer, D. M., Hsieh, V., Hall, A., Gerren, A., Walters, E., & Dalloul, M. (2022). Current Perspectives of Prenatal Cell-free DNA Screening in Clinical Management of First-Trimester Septated Cystic Hygroma. *International Journal of Women's Health*, 1499–1518.

Systematic Review: Perinatal outcomes of cystic hygroma. (2025). *American Journal of Obstetrics & Gynecology Maternal-Fetal Medicine,* Article S2589933325001041. https://doi.org/10.1016/S2589-9333(25)00104-1

Tsegaye, M. A., et al. (2024). An early second-trimester cystic hygroma with hydrops fetalis: Case report and review. *Journal of Medical Case Reports, …*(…), …pages. https://pmc.ncbi.nlm.nih.gov/articles/PMC11585464/

Van den Veyver, I. B., Chandler, N., Wilkins-Haug, L. E., Wapner, R. J., Chitty, L. S., & Directors, I. B. of. (2022). International society for prenatal diagnosis updated position statement on the use of genome-wide sequencing for prenatal diagnosis. *Prenatal Diagnosis*, *42*(6), 796–803.

Wang, M. J., Bazan, M., Hsieh, T., Mita, C., Ferrés, M. A., & Oyelese, Y. (2025). Perinatal outcomes of cystic hygroma: a systematic review and meta-analysis. *American Journal of Obstetrics & Gynecology MFM*, *7*(7).

Wapner, R. J., et al. (2020). Genetic syndromes associated with cystic hygroma. *Prenatal Diagnosis, 40*(…), …pages.

WHO. (2023). *Birth defects.* World Health Organization. https://www.who.int/news-room/fact-sheets/detail/birth-defects

WHO. (2024). *Newborn mortality fact sheet.* World Health Organization. https://www.who.int/news-room/fact-sheets/detail/newborn-mortality

Yakıştıran, B., et al. (2020). Analysis of cystic hygroma diagnosed in the first trimester: Single-center experience. *Journal of Turkish German Obstetrics and Gynecology Association, …*(…), …pages. https://pmc.ncbi.nlm.nih.gov/articles/PMC7294829/

Zhen, L., et al. (2024). Recurrent first-trimester cystic hygroma with normal karyotype strongly indicates the existence of an autosomal recessive type of genetic disorder. *Case Reports in Obstetrics and Gynecology,* Article ID …

https://doi.org/10.1155/2024/…

Zheng, J., et al. (2024). Genetic correlation between fetal nuchal translucency and chromosomal anomalies detected by CMA in fetuses with cystic hygroma. *Scientific Reports*. https://www.nature.com/articles/s41598-024-76628-y

Zhou, Y., Lu, X., Zhang, Y., Ge, Y., Xu, Y., Wu, L., & Jiang, Y. (2023). Prenatal genetic diagnosis of fetal cystic hygroma: a retrospective single-center study from China. *Cytogenetic and Genome Research*, *162*(7), 354–364.