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Case Report

Fetal Cystic Hygroma Accompanied by Oligohydramnios: Case Report and Clinical Insights

Shamima Haque Chowdhury[1*], Waheeda Nargis[2]

[1]Professor & Head, Department of Gynaecology & Obstetrics, Uttara Adhunik Medical College, Dhaka, Bangladesh
[2]Professor, Department of Biochemistry, Uttara Adhunik Medical College, Dhaka, Bangladesh

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***Corresponding author:** Shamima Haque Chowdhury
Professor & Head, Department of Gynaecology & Obstetrics, Uttara Adhunik Medical College, Dhaka, Bangladesh

Abstract

Background: Cystic hygroma is a congenital lymphatic malformation frequently associated with chromosomal abnormalities, hydrops fetalis, and high fetal mortality. Diagnosed predominantly in the first trimester, late-detected cases often present severe complications and poor prognosis, particularly in low-resource settings with limited access to advanced diagnostic modalities. ***Case Presentation****:* We report the case of a 30-year-old third gravida woman in Bangladesh who presented at over 24 weeks' gestation with amenorrhea and a prenatal ultrasound indicative of cystic hygroma, pleural effusion, and oligohydramnios. Initial ultrasound findings included a large cystic mass at the fetal neck, pleural effusion, and body hyperflexion, suggestive of fetal hydrops and cystic hygroma. Serial ultrasounds confirmed persistent cystic hygroma, increased pleural effusion, and ascites, alongside declining amniotic fluid levels. Limited access to genetic testing restricted comprehensive diagnostic evaluation. The case highlights the high-risk nature of late-diagnosed cystic hygroma, especially where resource constraints limit available interventions. ***Conclusion****:* This case underscores the poor prognosis of cystic hygroma in advanced gestation, emphasizing the importance of early screening, regular prenatal visits, and improved access to genetic counseling and non-invasive diagnostic technologies. Future research should prioritize affordable diagnostic tools for low-resource healthcare environments to support timely diagnosis and management.

Keywords: Cystic Hygroma, Fetal Hydrops, Oligohydramnios.

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INTRODUCTION

Cystic hygroma is a congenital lymphatic malformation characterized by fluid-filled cysts, often arising in the fetal neck due to abnormal development or blockage of the lymphatic system. These cystic structures, filled with lymphatic fluid, manifest as translucent, septated masses commonly detectable during routine ultrasound (USG) in the first or second trimester of pregnancy, allowing early identification of potential complications in the fetal development trajectory [1]. Globally, cystic hygroma has an estimated prevalence of approximately 1 in 1,000 to 1 in 6,000 live births, with some regional variations observed in specific genetic or ethnic populations [2]. Although it can present as an isolated anomaly, cystic hygroma is frequently associated with chromosomal abnormalities, notably Turner syndrome (45,X) and trisomy 21, both of which carry serious prognostic implications [3].

The clinical relevance of diagnosing cystic hygroma in utero cannot be understated due to its potential impact on fetal viability and developmental outcomes. Early prenatal identification facilitates timely decision-making, allowing clinicians to counsel families about the prognosis and, when possible, prepare for perinatal interventions. According to a study by Chen *et al*., (1996), fetal cystic hygroma detected in the second or third trimester often coincides with severe complications, such as hydrops fetalis, oligohydramnios, or intrauterine fetal death, and requires advanced cytogenetic evaluation to provide accurate prognostic information [4]. However, the prognosis for cystic hygroma is highly variable, with outcomes dependent on the presence of additional anomalies or genetic abnormalities and the timing of diagnosis. For instance, the prognosis is generally poorer when cystic hygroma is associated with fetal hydrops or oligohydramnios, conditions which heighten the risk of fetal growth

restriction, developmental anomalies, and, in many cases, perinatal mortality [5, 6]. This variability underscores the need for individualized case assessment and multidisciplinary management strategies.

In terms of diagnostic modalities, ultrasound remains the frontline tool for the prenatal detection of cystic hygroma, often supplemented by genetic testing via amniocentesis or chorionic villus sampling (CVS) to confirm chromosomal abnormalities [7]. Amniocentesis and fetal karyotyping are critical, as approximately 50-60% of cystic hygroma cases are linked to chromosomal abnormalities, with monosomy X and trisomy 21 being the most common associations [8]. Demir *et al*., (2022) recommend invasive diagnostic techniques in cases of cystic hygroma to verify chromosomal status, given the significant implications of aneuploidy for fetal health [3]. In cases complicated by oligohydramnios, however, diagnostic challenges arise, as low amniotic fluid levels can obstruct detailed imaging and restrict amniocentesis, thereby complicating the assessment. Floriani *et al*., (2016) addressed this diagnostic dilemma by successfully utilizing fluid from the cystic hygroma itself for fetal karyotyping when amniocentesis was not feasible, which revealed a diagnosis of Turner syndrome in one of the cases, suggesting an alternative approach for cytogenetic analysis in complex cases [4].

Epidemiological data reveal some notable trends regarding the prevalence of cystic hygroma. Studies have shown that regional and ethnic factors may influence the incidence rates, as observed in various demographic studies worldwide. For example, research by Forrester and Merz (2004) highlighted elevated incidence rates among specific ethnic groups, pointing to potential genetic predispositions that may affect fetal susceptibility to cystic hygroma [2]. In South Asia, however, there is limited access to advanced prenatal diagnostic technologies, particularly in rural or under-resourced regions. This disparity in access impacts the early detection and management of complex fetal anomalies, making cases like this particularly relevant to understanding regional healthcare challenges. Additionally, the association of cystic hygroma with environmental or genetic factors in the South Asian context, though not extensively documented, suggests that future research should focus on genetic and sociodemographic aspects influencing its prevalence [9].

Importantly, cystic hygroma's association with chromosomal abnormalities necessitates genetic counseling, a vital component of comprehensive prenatal care that aids families in understanding the prognosis and making informed decisions. Shimada *et al*., (2009) emphasized that cystic hygroma is a strong predictor of chromosomal abnormalities, warranting detailed genetic counseling to help families navigate complex prognostic information [6]. Such counseling is essential in cases where cystic hygroma is detected alongside other markers, such as increased nuchal translucency, polyhydramnios, or fetal growth restrictions, as these indicators heighten the risk of chromosomal anomalies and adverse outcomes [10]. Furthermore, Zhou *et al*., (2023) illustrated that integrating advanced genetic testing methods, such as chromosomal microarray analysis (CMA), with conventional karyotyping improves diagnostic precision, allowing for better-informed clinical decisions and personalized genetic counseling [5].

In summary, cystic hygroma is a significant fetal condition with varied prognostic implications, strongly influenced by genetic factors, gestational timing, and the presence of additional abnormalities like oligohydramnios. The complexity of diagnosing and managing cystic hygroma, especially in low-resource regions, underscores the importance of both early prenatal detection and comprehensive genetic evaluation to provide optimal care and support for affected families. This case report aims to add to the literature by examining the diagnostic and management challenges presented by a second-trimester fetal cystic hygroma with oligohydramnios, offering clinical insights relevant to the regional healthcare context in Bangladesh.

CASE PRESENTATION

A 30-year-old woman from Bangladesh presented at a tertiary hospital with a primary complaint of amenorrhea lasting over 24 weeks. She had recently confirmed her pregnancy using a strip test and sought further evaluation following an initial ultrasonography (USG) that revealed fetal abnormalities. Her obstetric history included being a third gravida with one prior live birth and one previous abortion approximately 10 years earlier. Her last menstrual period (LMP) estimated her due date (EDD) as November 9, 2021. She had no significant medical history, reporting no diabetes mellitus (DM), hypertension (HTN), bronchial asthma (BA), or notable drug usage.

At the first visit, the patient was clinically stable, with a pulse of 70 beats per minute, blood pressure of 100/70 mmHg, and a respiratory rate of 16 breaths per minute. General physical examination revealed no significant abnormalities, although her abdomen was soft and tender on palpation. Her body build was noted as average. A detailed ultrasound conducted during this visit revealed a floating fetal presentation and an estimated gestational age of 22 weeks and 5 days, based on biometric parameters with a biparietal diameter (BPD) of 52.1 mm and femur length (FL) of 37.2 mm. The estimated fetal weight was 575 ± 86 grams. Notably, the scan showed absent fetal cardiac pulsation and movement, along with significant abnormalities, including thickening of the abdominal wall, body hyperflexion, bilateral pleural effusion, and a large cystic mass posterior to the fetal neck, measuring approximately 12 x 10 cm. These findings suggested a diagnosis of missed abortion at about 23 weeks, accompanied by cystic hygroma. A provisional

diagnosis of missed abortion with cystic hygroma was made, and further follow-up and anomaly scans were recommended.

During the second visit, a follow-up USG revealed a single live fetus with an unstable presentation and an estimated gestational age of 16 weeks and 3 days (BPD 33 mm, FL 19 mm). Amniotic fluid levels were adequate, although a cystic structure with a volume of 34 ml was observed at the posterior neck, indicative of cystic hygroma. Further evaluation, including an anomaly scan, was recommended to assess the fetal condition more thoroughly. Laboratory investigations during this visit included hematology, biochemistry, serology, and urine analysis. The hematology report showed a hemoglobin level of 10.4 g/dL, total red blood cell (RBC) count of 4.32 million/uL, and platelet count of 299,000/uL. Biochemistry findings were within normal limits, with serum urea at 19 mg/dL, random plasma glucose at 5.02 mmol/L, and serum creatinine at 0.53 mg/dL. Serology tests were negative for HbsAg and VDRL. Urine examination showed no significant abnormalities.

At the third visit, a follow-up ultrasound revealed an estimated gestational age of 21 weeks and 5 days (BPD 51 mm, FL 31 mm). The fetus was in breech presentation with an anterior placenta positioned away from the os. Amniotic fluid was adequate, with an amniotic fluid index (AFI) of 13.0 cm. Findings included a large multilocular cystic mass, measuring 11.2 x 11.3 cm, located at the posterior neck and extending to the abdominal wall with diffuse edema. Additional findings included bilateral pleural effusion, though the fetal brain, spine, intracranial ventricles, and abdominal organs appeared normal. Laboratory results from this visit showed a further decrease in hemoglobin to 9.6 g/dL, with an RBC count of 3.61 million/uL and WBC count of 12,500/uL. Immunology tests revealed a TSH level of 1.01 µIU/mL, elevated alpha-fetoprotein at 68.4 ng/mL, and low vitamin D at 17.0 ng/mL. Biochemistry results showed random plasma glucose at 6.09 mmol/L, with no significant findings in the urine examination.

A final ultrasound revealed an estimated gestational age of 22 weeks and 2 days (BPD 53 mm, FL 32 mm). The fetus remained in breech presentation, and amniotic fluid levels were noted to be at the lower limit of normal (AFI 9.3 cm). The cystic abnormality had enlarged to a multilocular mass measuring 13.2 x 12.9 cm, surrounding the fetal neck and extending into the soft tissues of the abdominal wall, consistent with diffuse edema. Additional findings included bilateral pleural effusion, ascites, and non-visualization of the fetal face and extremities due to edema. The scan suggested a viable pregnancy of 22+ weeks complicated by cystic hygroma with reduced amniotic fluid levels.

During the final follow-up, the patient's hemoglobin level had further declined to 8.7 g/dL. This continued decrease in maternal hemoglobin, combined with the advanced gestational cystic hygroma, bilateral pleural effusion, ascites, and reduced amniotic fluid, indicated a poor prognosis for fetal viability. This case highlights the complexities and high-risk nature of advanced gestational cystic hygroma compounded by oligohydramnios and structural abnormalities.

DISCUSSION

In this case report of a 30-year-old woman presenting with fetal cystic hygroma at an advanced gestational age, we observe a complex interplay of clinical findings, diagnostic challenges, and prognostic factors that echo the poor outcomes associated with late-diagnosed cystic hygroma. Cystic hygroma, a congenital lymphatic malformation commonly presenting as a cystic mass in the fetal neck, is often linked with chromosomal abnormalities such as Turner syndrome and trisomy 21, and complications like hydrops fetalis and pleural effusion further exacerbate its poor prognosis when diagnosed beyond the first trimester [11]. Similar to findings by Behera *et al*., (2020), where late antenatal diagnoses led to compromised neonatal outcomes, our case also encountered persistent cystic hygroma, oligohydramnios, and pleural effusion, elements that severely limit management options and are typically associated with near-certain fetal demise [12].

The diagnosis and management challenges are particularly noteworthy in this case. Late presentation at over 24 weeks compounded the limited visualization due to oligohydramnios, a scenario corroborated by Budd *et al*., (2011), who emphasized the increased risk of poor outcomes due to restricted imaging options in late-diagnosed cystic hygroma [13]. Additionally, Zhou *et al*., (2023) highlighted the value of early genetic screening with chromosomal microarray and karyotyping, which are less accessible in low-resource settings, to improve diagnostic certainty and guide family counseling [5]. In the context of our case, the absence of early screening and the resulting limitations in performing advanced diagnostic tests like amniocentesis reflect the real-world challenges faced in resource-limited settings, where timely diagnosis is critical yet often delayed.

When comparing prognostic factors, it is evident that cases diagnosed in the first trimester with isolated cystic hygroma and normal karyotypes hold better outcomes than those diagnosed later, which aligns with Thomas (1992), who observed that cases detected in the first trimester have more favorable prognoses if structural abnormalities and aneuploidy are absent [14]. However, our case involved persistent cystic hygroma and hydrops fetalis in the second trimester, reflecting the findings by Ravikanth & Prasannan (2019), who reported that the presence of hydrops, pleural effusion, and associated structural abnormalities in cystic hygroma significantly reduce survival rates [15]. Further, the literature suggests that in settings with

delayed presentation, outcomes are notably worse due to the added diagnostic and management limitations. Floriani *et al*., (2016) proposed alternative diagnostic methods using cystic hygroma fluid for genetic testing as a viable approach when amniocentesis is challenging due to low amniotic fluid, a method that may be relevant for cases like ours to overcome such resource constraints [4].

Moreover, findings in the literature stress the importance of case reports like this one in understanding the course and challenges of managing cystic hygroma, particularly in under-resourced settings where late presentation and limited diagnostic facilities complicate management. Behera *et al*., (2020) and Fisher *et al*., (1996) highlighted how retrospective case reviews help to refine clinical approaches in regions with restricted healthcare access, advocating for multidisciplinary teams to support family counseling and management [12, 16]. This aligns with our observations, as the late detection limited genetic testing options, underscoring the need for regular prenatal screenings and multidisciplinary collaboration to support timely diagnosis and optimal care.

In conclusion, this case reflects the poor prognosis associated with advanced cystic hygroma complicated by oligohydramnios, underscoring the need for accessible early screening and comprehensive diagnostic approaches, as suggested by Budd *et al*., and Zhou *et al*., [5, 13]. Future research should focus on cost-effective, non-invasive screening methods and resource-appropriate diagnostic tools that facilitate timely interventions in low-resource settings. Through observational case reports, we can deepen our understanding of cystic hygroma's clinical progression and highlight the need for broader access to prenatal care resources in similar healthcare environments.

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CONCLUSION

In this case report of advanced gestational cystic hygroma with associated pleural effusion, ascites, and oligohydramnios, we underscore the challenges and poor prognosis linked to late-diagnosed fetal anomalies, particularly in low-resource settings. Diagnosed in the second trimester, this case reflects the significant morbidity associated with cystic hygroma when compounded by hydrops fetalis and limited diagnostic options. The case highlights the crucial role of early screening, regular prenatal visits, and the need for accessible diagnostic tools and genetic counseling to improve management and outcomes. Future efforts should focus on enhancing diagnostic capabilities and developing cost-effective screening alternatives, especially tailored for low-resource healthcare environments, to facilitate timely diagnosis and intervention for complex congenital anomalies like cystic hygroma. This report contributes valuable clinical insights into the management challenges and prognostic complexities of cystic hygroma in advanced gestation and limited-resource settings.

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REFERENCES

1. Chen, C. P., Liu, F. F., Jan, S. W., Lee, C. C., Town, D. D., & Lan, C. C. (1996). Cytogenetic evaluation of cystic hygroma associated with hydrops fetalis, oligohydramnios or intrauterine fetal death: the roles of amniocentesis, postmortem chorionic villus sampling and cystic hygroma paracentesis. *Acta obstetricia et gynecologica Scandinavica*, *75*(5), 454-458. Available from: https://onlinelibrary.wiley.com/doi/abs/10.3109/00016349609033353
2. Forrester, M. B., & Merz, R. D. (2004). Descriptive epidemiology of cystic hygroma: Hawaii, 1986 to 1999. *Southern medical journal*, *97*(7), 631-637. Available from: https://go.gale.com/ps/i.do?p=AONE&sw=w&issn=00384348&v=2.1&it=r&id=GALE%7CA120354581&sid=googleScholar&linkaccess=abs
3. Demir, S. S., Cagliyan, E., Öztürk, D., Özmen, S., Altunyurt, S., Çankaya, T., & Bora, E. (2022). Prenatal diagnosis of cystic hygroma cases in a tertiary centre and retrospective analysis of pregnancy results. *Journal of Obstetrics and Gynaecology*, *42*(7), 2899-2904. Available from: https://doi.org/10.1080/01443615.2022.2112023
4. Floriani, M. A., Dorfman, L. E., Koshiyama, D. B., Zen, T. D., Telles, J. A. B., Cunha, A. C., ... & Rosa, R. F. M. (2016). Uso do fluido obtido por punção do higroma cístico: método alternativo para cariotipagem fetal. *Jornal Brasileiro de Patologia e Medicina Laboratorial*, *52*, 391-392. Available from: https://www.scielo.br/j/jbpml/a/Zq3vv9CcSzY6Z8KYsczMCSS/abstract/?lang=pt
5. 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. Available from: https://doi.org/10.1159/000528600
6. Shimada, S., Yamada, H., Hoshi, N., Kobashi, G., Okuyama, K., Hanatani, K., & Fujimoto, S. (2009). Specific ultrasound findings associated with fetal chromosome abnormalities. *Congenital anomalies*, *49*(2), 61-65. Available from: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1741-4520.2009.00224.x
7. Bianca, S., Bartoloni, G., Boemi, G., Barrano, B., Barone, C., Cataliotti, A., ... & Ettore, G. (2010). Familial nuchal cystic hygroma without fetal effects: Genetic counselling and further evidence for an autosomal recessive subtype. *Congenital anomalies*, *50*(2), 139-140. Available from:

https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1741-4520.2010.00273.x

8. Dhillon, P. K., Jeemon, P., Arora, N. K., Mathur, P., Maskey, M., Sukirna, R. D., & Prabhakaran, D. (2012). Status of epidemiology in the WHO South-East Asia region: burden of disease, determinants of health and epidemiological research, workforce and training capacity. *International journal of epidemiology*, *41*(3), 847-860. Available from: https://doi.org/10.1093/ije/dys046
9. Golan, A., Lin, G., Evron, S., Arieli, S., Niv, D., & David, M. P. (1994). Oligohydramnios: maternal complications and fetal outcome in 145 cases. *Gynecologic and obstetric investigation*, *37*(2), 91-95. Available from: https://doi.org/10.1159/000292532
10. Langer, J. C., Fitzgerald, P. G., Desa, D., Filly, R. A., Golbus, M. S., Adzick, N. S., & Harrison, M. R. (1990). Cervical cystic hygroma in the fetus: clinical spectrum and outcome. *Journal of pediatric surgery*, *25*(1), 58-62. Available from: https://www.jpedsurg.org/article/S0022-3468(05)80164-2/abstract
11. Descamps, P. H., Jourdain, O., Paillet, C. H., Toutain, A., Guichet, A., Pourcelot, D., ... & Body, G. (1997). Etiology, prognosis and management of nuchal cystic hygroma: 25 new cases and literature review. *European Journal of Obstetrics & Gynecology and Reproductive Biology*, *71*(1), 3-10. Available from: https://www.ejog.org/article/S0301-2115(96)02590-0/abstract
12. Behera, S., Bawa, M., Saha, P. K., Singh, T., & Samujh, R. (2020). Outcome of antenatally diagnosed cystic hygroma-Lessons learnt. *International journal of pediatric otorhinolaryngology*, *138*, 110227. Available from: https://www.sciencedirect.com/science/article/pii/S0165587620303700
13. Budd, J. L. S., Craft, E., & Draper, E. S. (2011). Outcomes following prenatal diagnosis of cystic hygroma: east midlands & south yorkshire congenital anomalies register (EMSYCAR) 2000–2009. *Archives of Disease in Childhood-Fetal and Neonatal Edition*, *96*(Suppl 1), Fa73-Fa74. Available from: https://fn.bmj.com/content/96/Suppl_1/Fa73.3
14. Thomas, R. L. (1992). Prenatal diagnosis of giant cystic hygroma: prognosis, counselling, and management; case presentation and review of the recent literature. *Prenatal diagnosis*, *12*(11), 919-923. Available from: https://onlinelibrary.wiley.com/doi/abs/10.1002/pd.1970121111
15. Ravikanth, R., & Prasannan, R. (2019). Hydrops fetalis with septated cystic hygroma. *Apollo Medicine*, *16*(1), 47-49.
16. Fisher, R., Partington, A., & Dykes, E. (1996). Cystic hygroma: comparison between prenatal and postnatal diagnosis. *Journal of pediatric surgery*, *31*(4), 473-476. Available from: https://www.jpedsurg.org/article/S0022-3468(96)90477-7/abstract