Non-immune hydrops fetalis due to αlpha(α)-thalassemia: Ethical dilemmas and grief following perinatal loss

Flecia Kundayis, Dr.Fam.Med (Fam Med)

Klinik Kesihatan Lohan, Ranau, Sabah, Malaysia

SUMMARY

Hydrops fetalis is a serious condition with a poor prognosis for the affected fetus. While the incidence of immune hydrops fetalis has significantly decreased, cases of nonimmune hydrops fetalis are becoming more common. Nonimmune hydrops can result from hemoglobinopathies, such as α-thalassemia. This case report discusses the diagnosis and management of nonimmune hydrops fetalis due to α-thalassemia. Given the high prevalence of thalassemia in Sabah, Malaysia, it is recommended that all cases of hydrops fetalis in this region be investigated for thalassemia. The case underscores the importance of early screening and highlights the ethical and psychological challenges associated with decisions regarding pregnancy termination. Counselling was provided to support the patient through the decision-making process, addressing both ethical dilemmas and emotional challenges. Postpartum, the patient experienced grief over the loss but was able to achieve emotional resolution with the support of her family and healthcare providers.

INTRODUCTION

Hydrops fetalis is a serious fetal condition characterized by the accumulation of excess fluid in two or more body compartments. It can be classified into immune hydrops fetalis, caused by maternal hemolytic antibodies, and nonimmune hydrops fetalis, which results from various other causes. Before the introduction of prophylactic anti-D immunoglobulin, immune hydrops was the most common type. However, immunologic causes now account for less than 20% of cases. In Southeast Asia, nonimmune hydrops fetalis is more prevalent, with an incidence ranging from 1 in 500 to 1 in 1500 cases.[1] α-thalassemia is a significant genetic blood disorder in this region, with a prevalence of 17.3% in Malaysia and a notably higher prevalence of 33.6% among the Kadazandusun ethnic group in Sabah.[2] Severe forms of α-thalassemia, such as Hb Bart's hydrops fetalis, cause fetal anemia due to impaired α-globin production, leading to high-output cardiac failure and fluid accumulation in multiple fetal compartments. Today, early diagnosis of hydrops fetalis and other congenital anomalies is possible through ultrasound, karyotyping, and molecular genetic testing. Due to the poor prognosis associated with hydrops fetalis, pregnancy termination is often considered. This report presents a case of nonimmune hydrops fetalis due to homozygous α-thalassemia, diagnosed late in pregnancy, and discusses the challenges faced by the medical team and the family in managing this condition.

CASE PRESENTATION

A 32-year-old Dusun woman, primigravida, was referred from a private clinic at 23 weeks of gestation due to suspected hydrops fetalis. She reported no symptoms of anaemia, fever, or bleeding. She had no history of blood transfusions or menorrhagia and had not been previously diagnosed with thalassemia. She was unsure of any family history of thalassemia. Concerned for her first child's health, she sought further evaluation.

On physical examination, the patient's height was 150 cm, weight 55 kg, and BMI 24.4 (normal). Vital signs were within normal limits. Although her conjunctivae appeared pale, she displayed no physical features typical of thalassemia. The uterus was appropriately sized for gestational age, with a symphysis-fundal height of 23 cm. There was no hepatosplenomegaly, and other systemic examinations were unremarkable.

Blood tests revealed the following key findings:

The DNA analysis revealed that both the patient and her husband were α-thalassemia carriers (Table I). Peripheral blood film (PBF), iron studies, and stool examination results were normal. Ultrasound findings showed fetal ascites in multiple compartments, including the abdomen, lungs, and heart. Doppler studies, including middle cerebral artery peak systolic velocity (MCA-PSV), were not performed in this case.

Table I: Patient's and husband's blood test panel

Test	Patient	Husband	Normal range
Hemoglobin (Hb)	9.9 g/dL	14.1 g/dL	12.0–16.0 g/dL (female) 14.0-18.0 g/dL (male)
Mean Corpuscular Volume (MCV)	69.2 fL	70.0 fL	80–100 fL
Mean Corpuscular Hemoglobin (MCH)	22.1 pg	21.4 pg	27–32 pg
DNA Analysis	Carrier of α-thalassemia	Carrier of α-thalassemia	Not Applicable

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Corresponding Author: Flecia Kundayis
Email: fleciakundayis@gmail.com

Breaking the news of the fetal abnormalities was emotionally challenging for both the couple and the clinician. The diagnosis, its causes, and the poor prognosis of hydrops fetalis were communicated to the couple in a clear and empathetic manner. Termination of pregnancy was presented as an option, and the steps and potential outcomes were explained in simple terms. The couple was given time to consider their decision and encouraged to ask questions. Clear communication and empathy were crucial in supporting the patient. After careful consideration, the patient chose to continue the pregnancy, and her decision was respected. Acknowledging the complexity of the case, the primary care team referred the patient to a maternal-fetal medicine (MFM) specialist for co-management and guidance on fetal monitoring and potential interventions.

At 24 weeks, the MFM specialist counselled the patient regarding the option of pregnancy termination. The couple participated in the discussion and received information on the fetus's condition and prognosis. Although termination was offered, the patient decided to continue the pregnancy. Her autonomy was respected, and the ongoing support from her husband helped her cope with her worries. She occasionally felt guilt, but she was reassured that the fetal anomaly was beyond her control. She did not exhibit symptoms of depression or anxiety during the visit.

By the 30-week follow-up at the MFM clinic, the patient had developed severe preeclampsia and became edematous. She presented with headache, blurred vision, elevated blood pressure at 163/109 mm Hg, and proteinuria (2+). She was admitted to stabilize her blood pressure and underwent an emergency caesarean section due to severe preeclampsia. Unfortunately, the fetus was born prematurely and, despite supportive care, died shortly after birth due to non-viability.

During her postpartum visit, her blood pressure remained stable with antihypertensive medication (Tablet Labetalol 200 mg TDS). She expressed grief over the pregnancy loss, often blaming herself for not recognizing the pregnancy earlier, though no depressive or anxiety symptoms were present. Her emotions were acknowledged, and she was reassured that the outcome was beyond her control. She received strong support from her husband, which was crucial in preventing complicated grief or depression. A referral to a psychologist was offered, and she was scheduled for a follow-up in two weeks, with instructions to return sooner if depressive symptoms emerged.

At subsequent follow-ups, her grief had resolved. She opted for long-acting reversible contraception and was advised to pursue early booking and referral to an MFM specialist clinic for prenatal testing if she becomes pregnant in the future.

DISCUSSION

α-thalassemia can lead to severe fetal conditions, such as hydrops fetalis, which often results in poor outcomes. In this case, the patient was informed of the fetus's prognosis and the option of pregnancy termination. Despite the severity, the patient chose to continue the pregnancy, influenced by personal and cultural factors. In Southeast Asian cultures, particularly in the Kadazandusun community, beliefs surrounding the sacredness of life and family values significantly impact such decisions. The patient's decision was supported by her husband, and cultural norms that emphasize continuing pregnancy regardless of fetal health likely played a role. The healthcare team respected the patient's autonomy while offering emotional support and counselling.

This case highlights the importance of cultural sensitivity in decision-making and the need for healthcare providers to balance ethical principles such as respect for autonomy, beneficence, and non-maleficence while considering the patient's values and preferences. Additionally, the parents were unaware of their α-thalassemia carrier status until the 23rd week of gestation, underscoring the need for early screening and genetic counselling before conception or during the first trimester. Early detection can reduce psychological distress and support timely decision-making regarding prenatal diagnosis and reproductive options.

While Malaysia has a national screening program targeting adolescents, there are gaps in the system, especially in high-risk regions like Sabah, where higher thalassemia carrier rates exist. Comprehensive screening strategies in these areas would allow for earlier identification of at-risk couples. Currently, most screening occurs during adolescence and early pregnancy, but many couples in high-risk areas may not receive adequate prenatal counselling or testing until later in pregnancy. This delay limits options for timely interventions and decisions regarding pregnancy outcomes. To address these gaps, earlier prenatal testing for couples with known risk factors is crucial.

Screening should be considered for couples with red cell abnormalities like microcytosis and hypochromia in the absence of iron deficiency.[3] In Sabah, where α-thalassemia is prevalent, investigating nonimmune hydrops fetalis for potential α-thalassemia is particularly important. Early identification of carrier status through expanded prenatal screening by the end of the first trimester could facilitate better decision-making and reduce emotional distress. Integrating genetic counselling into routine antenatal care for high-risk populations would help ensure families are fully informed, allowing for informed decisions when complications arise.

Improving accessibility to screening programs and raising awareness in high-risk communities through educational campaigns could encourage early diagnosis, reducing the incidence of late-stage diagnoses and improving clinical outcomes and patient well-being. The emotional and psychological impact of pregnancy loss, particularly after the diagnosis of severe fetal anomalies like hydrops fetalis, is significant. Grief counselling and support are vital in helping parents cope and preventing complicated grief. Research has shown that early intervention in grief counselling can significantly reduce the risk of complicated grief.[4]

Timely emotional support, active listening, and reassurance that the loss was beyond their control are essential. Referring parents to mental health professionals, such as psychologists,

can aid in managing deep emotional distress. Support groups for parents who have experienced similar losses provide validation and a network for shared experiences. Follow-up care is essential to monitor the development of complicated grief, and research shows that early grief counselling can significantly reduce the risk of prolonged mourning, anxiety, or depression. Offering emotional support, facilitating open discussions, and respecting cultural beliefs surrounding grief can aid in the recovery process. Structured grief counselling can prevent escalation into clinical conditions like depression and anxiety, promoting better long-term psychological outcomes.[5] Additionally, acknowledging the role of partners in the grieving process and encouraging shared support within the family unit is crucial.

CONCLUSION

Early screening for α-thalassemia is vital to identify at-risk couples early, allowing for timely counselling and decision-making, particularly in high-prevalence regions like Sabah. Empathetic and ethical communication is essential in managing complex cases, ensuring that patients are fully informed, supported, and respected in their decisions, ultimately improving both clinical and emotional outcomes.

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CONFLICT OF INTEREST

There is no conflict of interest to declare.

CONSENT FOR PUBLICATION

Written informed consent was obtained from the patient for publication of this case report.

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