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Early gestational diagnosis of alobar holoprosencephaly with omphalocele: A case report

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Abstract

Background: Holoprosencephaly (HPE) is the most frequent congenital brain malformation, with alobar HPE representing the most severe form. It is often accompanied by other structural anomalies such as omphalocele, significantly complicating fetal prognosis and perinatal management.

Purpose: To present the early gestational diagnosis of alobar holoprosencephaly with omphalocele.

Method: A descriptive case report of a 40-year-old gravida 2 para 1 woman whose fetus was diagnosed via ultrasound at 17–18 weeks' gestation with alobar HPE and omphalocele. The case was managed through routine antenatal care and serial ultrasound monitoring.

Results: Serial imaging revealed progressive ventriculomegaly and worsening abdominal wall defect. Despite the fatal nature of the anomalies, legal constraints in Indonesia prevented pregnancy termination. The fetus survived until intrauterine demise at 37–38 weeks, after which labor was induced and a stillborn delivered vaginally.

Conclusion: This case underscores the importance of early gestational diagnosis and highlights the ethical and legal challenges in managing lethal fetal anomalies in restrictive settings. Comprehensive parental counseling and genetic evaluation are crucial components in delivering informed care.

Keywords: Congenital Anomaly; Fetal Malformation; Intrauterine Fetal Demise; Prenatal Diagnosis.

INTRODUCTION

Holoprosencephaly (HPE) is the most frequently occurring developmental anomaly of the forebrain, arising due to the partial or complete failure of the prosencephalon to divide properly (Abubakar, Ibrahim, Ahidjo, & Tahir, 2014). Animal studies, particularly in mice, indicate that the critical period for the development of HPE in humans is between the third- and fourth-weeks post-conception. This condition is commonly associated with midline facial abnormalities and has a high rate of fetal mortality (Conner, Vejde, & Burgos, 2018). HPE is estimated to affect around 1 in 16,000 live births and approximately 1 in 250 spontaneous miscarriages. The condition is classified into three types alobar, semilobar, and lobar based on the extent of brain malformation, with alobar HPE representing the most severe form (Crider, Qi,

Yeung, Mai, Zauche, Wang, Daniels, & Williams, 2022). Omphalocele, which involves the protrusion of abdominal contents into the umbilical cord base, occurs in roughly 1 in 4,000 live births and is frequently found alongside other congenital defects (Ionescu, Călin, Navolan, Matei, Dimitriu, Herghelegiu, & Pleș, 2018). The co-occurrence of alobar HPE and omphalocele is exceptionally uncommon. Typically, fetuses diagnosed with alobar HPE do not survive until term, making the near-term gestation in this particular case both rare and clinically noteworthy (Tonni & Centini, 2006; Bogart, 1986; Waghmare, Sathe, Goel, & Kandalkar, 2016).

A 40-year-old woman, gravida 2 para 1, presented at 38–39 weeks of gestation reporting the absence of fetal movement for four hours. Her antenatal care had

been routine, and at 17–18 weeks' gestation, she had been informed of multiple fetal anomalies identified via ultrasound, including alobar holoprosencephaly and an omphalocele. She had no notable medical, familial, or psychosocial history, and there were no known congenital abnormalities in her family. The patient denied smoking, alcohol intake, radiation exposure, or chronic conditions such as hypertension, diabetes, or renal disease. She had not taken folic acid supplements during the pregnancy. Her obstetric history included a previous spontaneous vaginal delivery of a healthy 3000-gram female infant, assisted by a traditional birth attendant (Waikar & Singh, 2019; Suwardewa, Mulyana, & Setiawan, 2022). After that birth, she used injectable contraception for one year before becoming pregnant again. Upon physical examination, the patient appeared to be in moderate condition with stable vital signs. Abdominal assessment showed asymmetrical enlargement consistent with a term pregnancy. Due to polyhydramnios, fetal positioning through Leopold's maneuvers was difficult, and fetal heart tones were absent. A vaginal exam revealed a closed, posterior cervix with a Bishop score of 2. No labor activity, abnormal discharge, or vaginal bleeding was observed. Laboratory results were within normal limits, and serologic screening for HIV and hepatitis B was negative (Liang, Chen, Chang, & Wang, 2013; El-Dessouky, El-Khayat, El-Fiki, & El-Gharib, 2023).

RESEARCH METHOD

This study utilized a descriptive case report approach to present the clinical course of a pregnant woman diagnosed with alobar holoprosencephaly and omphalocele during early gestation. Data were collected through patient interviews, antenatal medical records, serial ultrasound findings, and delivery outcomes. Ethical considerations were observed, and written informed consent was obtained from the patient for publication and educational use.

RESEARCH RESULTS

This case report describes a 40-year-old gravida 2 para 1 woman whose fetus was diagnosed in early gestation (17–18 weeks) with alobar

holoprosencephaly and omphalocele through routine obstetric ultrasound. The diagnosis was based on findings of fused thalami, monoventricle, absent cerebral hemispheres, and a membranous abdominal wall defect. Serial ultrasonography revealed progressive worsening of both cranial and abdominal anomalies, along with persistent polyhydramnios (Bangma, Lunshof, Opstal, Galjaard, & Papatsonis, 2011). Despite the poor prognosis typically associated with alobar HPE, the fetus survived in utero until 37–38 weeks, when intrauterine fetal demise was confirmed. Labor was medically induced, resulting in the vaginal delivery of a stillborn male fetus with classic signs of alobar holoprosencephaly and a large omphalocele. This case highlights the clinical challenges of managing lethal congenital anomalies in environments with legal restrictions on pregnancy termination, emphasizing the importance of early prenatal diagnosis, ongoing parental counseling, and multidisciplinary care (Poenaru, Vilcea, & Marin, 2012).

Fetal development was monitored through serial ultrasounds during the pregnancy. At 17–18 weeks, ultrasonography revealed a singleton fetus with ventriculomegaly, fused thalami, monoventricle, and absence of the cerebral hemispheres—findings characteristic of alobar holoprosencephaly (Nyberg, Mack, Bronstein, Hirsch, Pagon, & Shepard, 1987; Mohamed, Ahmed, Elsayed Ahmed, & Ibrahim, 2024). Additionally, a membrane-covered abdominal wall defect measuring 2.71 cm × 2.23 cm with visible herniation of intestines and the umbilical vein indicated the presence of an omphalocele (Figure 1) (Taipale, Ammälä, Salonen, & Hiilesmaa, 2003). These anomalies persisted in subsequent scans, with a poor prognosis throughout. At 33–34 weeks, follow-up ultrasound showed enlargement of the abdominal wall defect to 6.29 cm × 4.23 cm, ongoing ventriculomegaly, and consistent signs of alobar HPE (Figure 2). At 37–38 weeks, intrauterine fetal demise was confirmed, along with continued polyhydramnios. No signs of maceration were noted, and fetal biometry corresponded to a gestational age of approximately 27–28 weeks (Figure 3).

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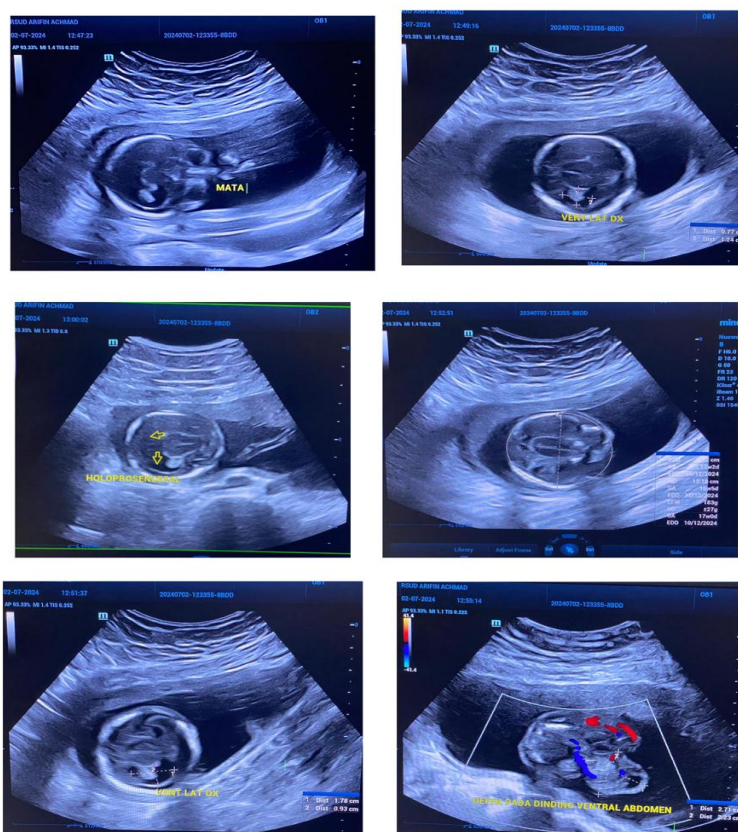


Figure 1. Ultrasound of the patient at 17-18 weeks of gestation, revealing several congenital anomalies

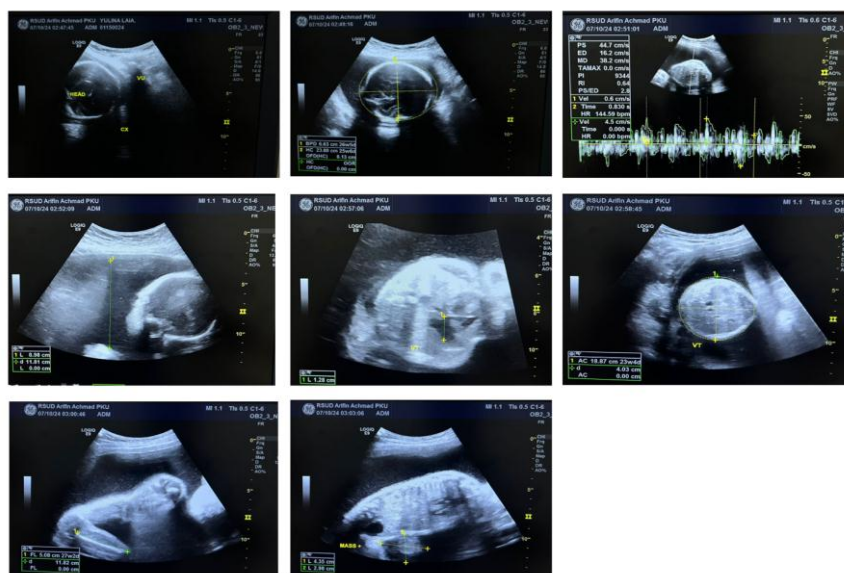


Figure 2. Ultrasound of the patient at 33-34 weeks of gestation, revealing persistent congenital anomalies

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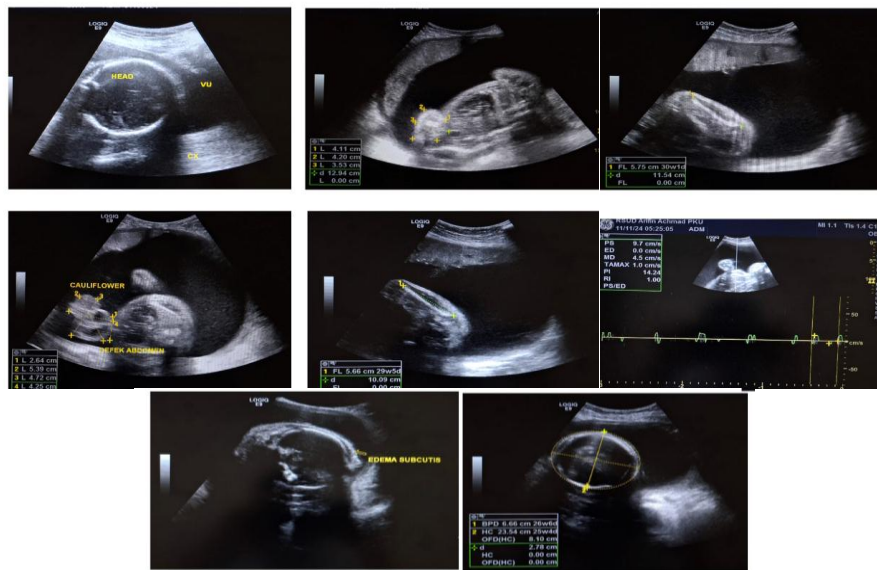


Figure 3. Ultrasound performed at 37–38 weeks of gestation demonstrating intrauterine fetal demise and persistent congenital anomalies: subcutis edema (+), omphalocele (+) alobar holoprosencephaly(+).

Following the diagnosis of intrauterine fetal demise, labor was induced using 50 mcg of misoprostol administered vaginally every four hours. As cervical dilation progressed and the bishop score surpassed six, an intravenous infusion of 10 IU oxytocin was initiated and gradually titrated to stimulate effective uterine contractions. The patient delivered a stillborn male infant via spontaneous vaginal delivery on November 12, 2024, at 11:05 AM. The fetus weighed 1320 grams and measured 34 cm in length. The umbilical cord was clamped and cut, and active management of the third stage of labor was conducted, including the administration of 10 IU intramuscular oxytocin and uterine massage.

When the placenta failed to expel spontaneously within 15 minutes, an additional dose of oxytocin was given. Persistent retention necessitated manual extraction, which was performed with informed consent. The placenta was delivered intact, weighing 220 grams, with a diameter of 16 cm and an umbilical cord measuring 35 cm in length (Figure 4).



Figure 4. Stillborn male infant with alobar holoprosencephaly and omphalocele, shown alongside the placenta.

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After delivery, the patient was closely monitored for uterine contraction, vaginal bleeding, and overall recovery. Her vital signs remained stable, and there were no signs of excessive postpartum hemorrhage, with an estimated blood loss of approximately 100 mL. She received intravenous fluids and was prescribed cefadroxil 500 mg twice daily, mefenamic acid 500 mg three times daily, and iron supplements. As part of postpartum family planning, an intrauterine contraceptive device (IUD) was inserted before discharge. The patient experienced no complications and was observed for 24 hours prior to being discharged in good condition with follow-up guidance. Emotionally, the patient reported profound sadness upon learning of the fetal abnormalities early in the pregnancy. While she initially held onto hope for a favorable outcome, she gradually came to terms with the poor prognosis following repeated consultations and ultrasound evaluations. She willingly provided consent to share her case for medical documentation and educational purposes, with assurance of confidentiality.

DISCUSSION

Alobar holoprosencephaly represents the most critical variant of holoprosencephaly, marked by the complete failure of the forebrain to divide, leading to the absence of structures such as the interhemispheric fissure and the corpus callosum (Kauvar & Muenke, 2010; Levey, Stashinko, Clegg, & Delgado, 2010). This condition is frequently accompanied by profound midline facial deformities and multiple systemic anomalies, including omphalocele (Waghmare et al., 2016). The case described in this report involves a fetus diagnosed early in pregnancy with both alobar holoprosencephaly and omphalocele, emphasizing the diagnostic and management complexities faced in prenatal care. What makes this case particularly notable is the fetus's survival until near-term gestation, despite the severity of its conditions. Typically, fetuses with alobar holoprosencephaly combined with multiple structural anomalies experience early fetal demise or are electively terminated. A neonate born with alobar HPE, meningomyelocele, and omphalocele, who passed away shortly after birth despite receiving intensive support (Tokmak, Timur, Dağlar, & Kara, 2014).

Several contributing factors are known to influence the development of holoprosencephaly, including advanced maternal age, chromosomal anomalies, and environmental exposures (Crider et al., 2022; Essa, Asrade Feleke, & Ahmed, 2018). Chromosomal abnormalities especially trisomy 13 and 18 are most frequently associated with HPE. However, in this case, no genetic testing was conducted to explore such possibilities. The patient had no history of alcohol intake, chronic illness, or infections, and she did not suffer from diabetes all known risk factors for HPE. Notably, she also did not take folic acid supplements during pregnancy, which may have contributed to neural tube defects and possibly this presentation.

The management of this case offered both benefits and limitations. One of the key advantages was the early identification of congenital anomalies, which allowed clinicians to offer counseling regarding the fetus's prognosis and care plan. Serial ultrasounds helped track the worsening condition. However, the absence of chromosomal analysis limited understanding of potential genetic causes. Although the patient received appropriate counseling, viable interventions were restricted by the poor prognosis (Tonni & Centini, 2006). Additionally, in Indonesia, termination of pregnancy due to fetal anomalies is legally constrained and requires both spousal approval and strong medical grounds, increasing the psychological stress for families. Cultural and religious sensitivities further complicate these decisions, as pregnancy termination is often regarded as morally or spiritually objectionable. Ultimately, this case illustrates the multifaceted challenges in managing severe congenital anomalies. Even when early diagnosis is achieved, clinical outcomes are generally poor and options for intervention remain limited. Furthermore, the legal and ethical frameworks in Indonesia add complexity to reproductive decisions, especially in cases of lethal anomalies. This case reinforces the urgent need for better access to genetic testing, enhanced prenatal counseling, and ethical guidance. As medical technologies evolve, expanding awareness and support systems will be crucial in empowering families to make informed decisions and receive compassionate care throughout the prenatal period (Suwardewa et al., 2022).

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CONCLUSION

Early gestational diagnosis of alobar holoprosencephaly with omphalocele presents significant clinical, ethical, and emotional challenges, particularly in settings with limited access to genetic testing and restrictive legal frameworks regarding pregnancy termination. This case underscores the importance of early prenatal screening, comprehensive parental counseling, and a multidisciplinary approach to managing lethal fetal anomalies. Enhancing access to diagnostic tools and supportive care can improve decision-making and provide more holistic care for affected families.

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