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CASE REPORT

A report on omphalocele and associated congenital deformity with intrauterine death: need for early diagnosis

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ABSTRACT

Omphalocele, a type of anterior abdominal wall defect, is a rare congenital deformity that is infrequently reported, which leads to the emergence of interventions that could be carried out for a better holistic management, for the baby, mother, and the family. We report a case of single dead fetus of 24 weeks gestational age delivered by a 25-year-old primigravida. On examination, it was observed and diagnosed that the baby presented with extrusion of multiple abdominal organs through a midline breach in the continuity of anterior abdominal wall suggesting it to be a case of omphalocele. It had other associated congenital deformities. Of all the investigations, ultrasonographic assessment of fetal structure and well-being is most reliable along with other investigations like genetic screening. The lack in the proper antenatal examination and infrequent routine checkups leads to a large proportion of undiagnosed cases. Proper diagnostic measures and awareness will bring a marked reduction in perinatal morbidity and mortality and avoidance of stigma faced by the patients.

Key words: Awareness, omphalocele, ultrasound

Introduction

Anterior wall defects are a group of rare congenital deformities that are infrequently reported. The incidence of omphalocele, gastroschisis, and exomphalos are worth noting as they frequently turn out to be fatal for the fetus or the neonate [1]. Special emphasis needs to be made for omphalocele as they are more associated with other lethal congenital defects leading to poor prognosis [2].

Omphalocele has been reported to have an incidence of 2–3 in 10,000 pregnancies [2,3]. It is considered to be a defect in regression of the normal physiological umbilical herniation of abdominal content during 6th to 10th week of gestation [4]. The three major diagnostic investigations are targeted antenatal ultrasound, echocardiography to detect associated lethal cardiac anomalies and finally karyotyping as these are very often associated with chromosomal abnormalities. Targeted ultrasound has been the primary imaging modality for the diagnosis and can be used for early identification of the cases by 10th to 12th week of gestation [5].

In developing country like ours, where there is profound lack of awareness for routine antenatal checkups among gravid women and incompetent management of such cases, we have to come up with ideas to increase the efficiency of primary health centers and other health organizations. Proper ultrasound examination, family history, routine examination for well-being of the fetus, awareness programs, and possible genetic screening are the important measures to be achieved. A few viable fetuses may be considered, after proper assessment, for the continuation of pregnancy with proper surgical and medical follow-ups to limit the complications in such cases. Achieving this will bring a marked reduction in perinatal morbidity and mortality and avoidance of stigma faced by the patients. We herein report a case of a single dead fetus with omphalocele and associated congenital abnormalities.

Case Report

A 25-year-old primigravida in labor was admitted in the Department of Obstetrics and Gynaecology in our hospital, referred from a private clinic in the suburbs with ultrasonographic evidence of Intra-Uterine Death of a single fetus of 24 weeks gestational age. The mother belonged to the rural environment and was educationally qualified only till matriculation in the Indian system. Awareness regarding antenatal check-up was very poor. She was unaware of her 1st day of last menstrual period and expected date of delivery. She was unbooked, as only 2 Anganwadi (provider of basic health care in Indian villages) visit were documented and no proper routine check-ups were performed. Later in the 4th month of pregnancy, she visited a local doctor and was given Tetanus Toxoid (TT) and supplement of iron and calcium. She was immunized with the 2nd dose of TT the next month. The first trimester was uneventful. The first documented ultrasound was performed peripherally 1 day before admission in Rajendra Institute of Medical Sciences after mother's concern for bleeding per vaginum 5 days back. It revealed a single dead

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fetus in the transverse lie of about 24 weeks. No congenital abnormalities were reported in this investigation. Mother suffered from malaria 1 year back. There was no relevant family history on both maternal and paternal sides. The mother was addicted to tobacco for the past 7–8 years and there was no history of allergy.

At the time of admission, her vitals were stable; pulse was 82/min and with a blood pressure of 130/84 mm of Hg. On abdominal examination, uterus was about 24 weeks in size with uterine contractions. On pelvic examination, she was found to be in early labor. She delivered a dead fetus vaginally. Following delivery and complete removal of placenta, proper examination of the dead baby was carried out. On examination, it was observed and diagnosed that the baby presented with extrusion of multiple abdominal organs through a midline breach in the continuity of anterior abdominal wall [Figure 1]. This defect was located centrally corresponding to the connection of umbilicus. The organs were covered partially by an approximately 1–2 mm thick membrane which was similar to the outer membrane of umbilical cord. The herniated contents comprised liver and dark gangrenous loop of intestine [Figure 2]. The breach was about 8 cm in length and stretched to a width of about 6 cm. The continuity of umbilical cord and the covering membrane was disrupted during the process of delivery. The baby had other associated congenital defects including congenital talipes equino varus, webbed neck, improper development of genitilia, and imperforate anus [Figure 2]. The parents did not give consent for autopsy due to their religious and cultural belief. The midline defect corresponding to the region of umbilicus along with other congenital deformities without any relevant family history indicated it to be a case of sporadic, nonisolated, central omphalocele which can be differentiated from similarly presenting right paramedian deformity known as gastroschisis.

Discussion

Omphalocele results from improper closure or absence of midline anterior abdominal wall which may lead to herniation of abdominal contents. They are usually subdivided into epigastric, central, and infraumbilical omphalocele according to the position of the defect, central being the most common [1]. Studies have shown strong co-relation of the central type of defect with an abnormal karyotype (69%) of which trisomy 18 was most frequently noted [2]. Very sparse numbers of patients with such defect were associated with good prognosis (8%) [1].

The etiopathogenesis of omphalocele and gastroschisis are debatable and have different theories for origin. Gastroschisis is believed to be formed due to defect in the closure of lateral body wall folds comprised parietal layer of lateral plate mesoderm and overlying ectoderm during the 4th week of gestation [4]. Omphalocele (1 in 4000) is rarer than gastroschisis (1 in 2000) [6]. Other rare defects worth being differentiated are bladder exstrophy, ectopia cordsis, and cloacal exstrophy. Exomphalos is a weakness of anterior abdominal wall, which leads to protrusion of abdominal content but within a sac contrary to the case we are reporting.

After studying 827 cases of omphalocele from the year 1996 to 2006, Deng et. al. established that 52.4% of pregnancies terminated with late fetal death with upward trend in successive years and 37.4% resulted in early neonatal death [2]. The incidence of nonisolated omphalocele (27.9%) was very less compared to isolated cases (72.1%) evidencing the lesser incidence of syndromic feature of the defect supporting the rarity of our case [2]. The mortality rate for patients in the gestational age of 28–36 weeks was 2.42 times higher than 37–42 weeks and hence earlier diagnosis will give us increased chances toward effective management of more cases well within time [2].

Associated abnormality that usually occurs with omphalocele is cleft palate and spinal deformity, but other less common abnormalities have also been reported. Findings of a rare syndrome of caudal regression introduced by Duhamel characterized by continuum of anorectal, urogenital and skeletal congenital abnormalities, imperforate anus and presence of congenital idiopathic clubfoot (talipes equino-varus) was somewhat parallel to our case [3].

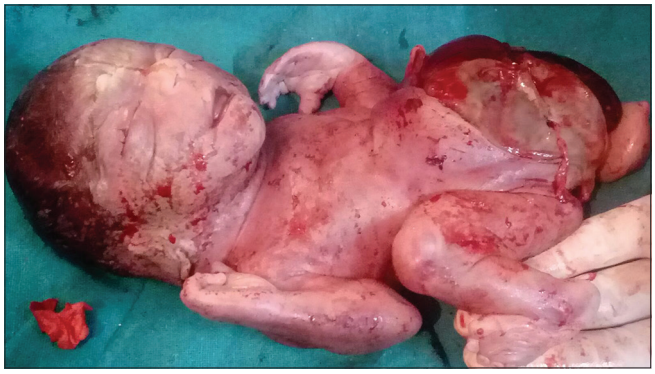

Figure 1 Omphalocele

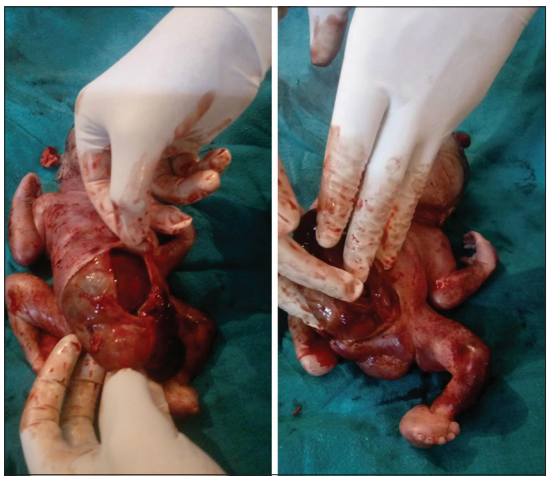

Figure 2 Features of omphalocele and associated abnormalities

Of all the investigations, ultrasonographic assessment of fetal structure and well-being is most reliable but the lack in the proper examination and infrequent routine checkups leads to a large proportion of undiagnosed cases. Only about 39.3% of cases are being diagnosed in the health centers and the rest 60.6% are confirmed by physical examination after birth similar to our report [2]. This evidence supported our concern toward the lack of awareness and proper diagnosis as projected in our report.

Conclusion

Omphalocele is a rare congenital anterior abdominal wall abnormality effecting infant mortality and quality of life. The diagnosis of such defect is necessary to be made well within time. Approach with better strategies to either electively terminate the pregnancy or assess the viability of fetus should be made followed by implementation of the optimal treatment protocol. The fact about lacking awareness and proper diagnosis yearns for more efficient training of health practitioner, proper reporting, and increased availability of diagnostic tools such as advanced ultrasonographic examination and availability of genetic screening in several neglected regions of developing country such as India.

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Authors' Contributions

Case identification, defining of intellectual content, manuscript preparation and editing, literature review and collection of media were done by the authors Utkarsh Gupta, Priyamvada Tiwari and Ridhi Kumari. Archana Kumari was the guarantor to our report.

Consent

The authors certify that a written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editor-in-chief of this journal.

Competing Interests

The authors declare that they have no competing interests

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