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Case of Omphalocele Diagnosed Prenatally: A Case Report

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Abstract

Omphalocele is congenital malformation resulted from abnormal closure of anterior abdominal wall. Omphalocele may exist in isolation or a part of syndromic congenital malformations. Several etiologies of omphalocele are known, including genetic, teratogenic, nutritional, or idiopathic causes. We reported a case of isolated omphalocele with no known risk factor. Female, 29 years old, G2P1A0, 21 weeks pregnant with no remarkable history referred to our institution for evaluation and management of suspected pregnancy complicated with omphalocele. Upon ultrasonographic examination, we discovered live singleton pregnancy with clear evidence of omphalocele. Patient was then planned to undergo elective cesarean section at 37th week of pregnancy. Subsequently, neonate was managed by the team of neonatologist and pediatric surgeon and the defect was repaired successfully. Prenatal diagnosis of omphalocele lead into improved outcome due to preparedness of the team, particularly in case of isolated case of omphalocele with no other congenital anomalies present.

Keywords: Omphalocele, Prenatal Diagnosis, Case Report

1. Introduction

Omphalocele is a type of congenital malformations signified with existence of the content of abdominal viscera outside the anterior wall, covered with peritoneum and Wharton's jelly, and results from the lack of reduction of physiological hernia during the first trimester of pregnancies.[1,2] Omphalocele occurs in approximately 2-3 per 10 thousand live births and tend to be related with other types of midline defects, although isolated cases are known to exist.[3,4] Several factors have been implicated in the occurrence of omphalocele, including genetic and chromosomal, exposure to teratogens, maternal nutritional factors, and idiopathic causes.[2,5,6] Exposed abdominal contents result in higher risk of mortality, thus planned delivery and surgical consultation is important when omphalocele is able to be diagnosed prenatal.[4,7,8] Isolated omphalocele case is slightly rarer (occurring in approximately 37% of all cases) than a part of multiple anomalies (42%).[3] In this case report, we present a case of giant omphalocele occuring in male fetus diagnosed prenatally since the second trimester. Due to the relative scarcity of reports regarding isolated omphalocele cases originating from Bangka and the elective cesarean section, we believe this case is able to contribute into the existing literature of omphalocele management

2. Summary of Case

Female, 29 years old, G2P1A0, 21 weeks pregnant, presented to our outpatient clinic after being referred due to suspicion of omphalocele. Obstetric history of the patient was unremarkable with single healthy female offspring was observed delivered via cesarean section in the last pregnancy. Previous ultrasonographic screening on 16th weeks of pregnancy revealed the existence of anterior abdominal wall defect with suspicion of omphalocele. The previous medical history of the patient was otherwise unremarkable with no known consumption of potentially teratogenic medications or environmental exposure, although the patient complained the allergy of several kinds of food.

Upon presentation, the general condition of the patient was unremarkable. The patient then underwent ultrasonographic examination and revealed the existence of singleton live pregnancy, EFW 351 grams, AGA, with anterior abdominal wall defect approximately 2.51 cm. No other congenital anomalies were observed. We monitored the patient closely and the patient was planned to undergo elective cesarean section in 37th week of pregnancy due to history of previous cesarean section.

Apart from the obvious omphalocele, screening for fetal anomalies (including fetal echocardiography, amniotic fluid measurements, and serial EFW evaluation) was within normal limits. Unfortunately, we did not follow the condition with prenatal genetic or chromosomal evaluation.

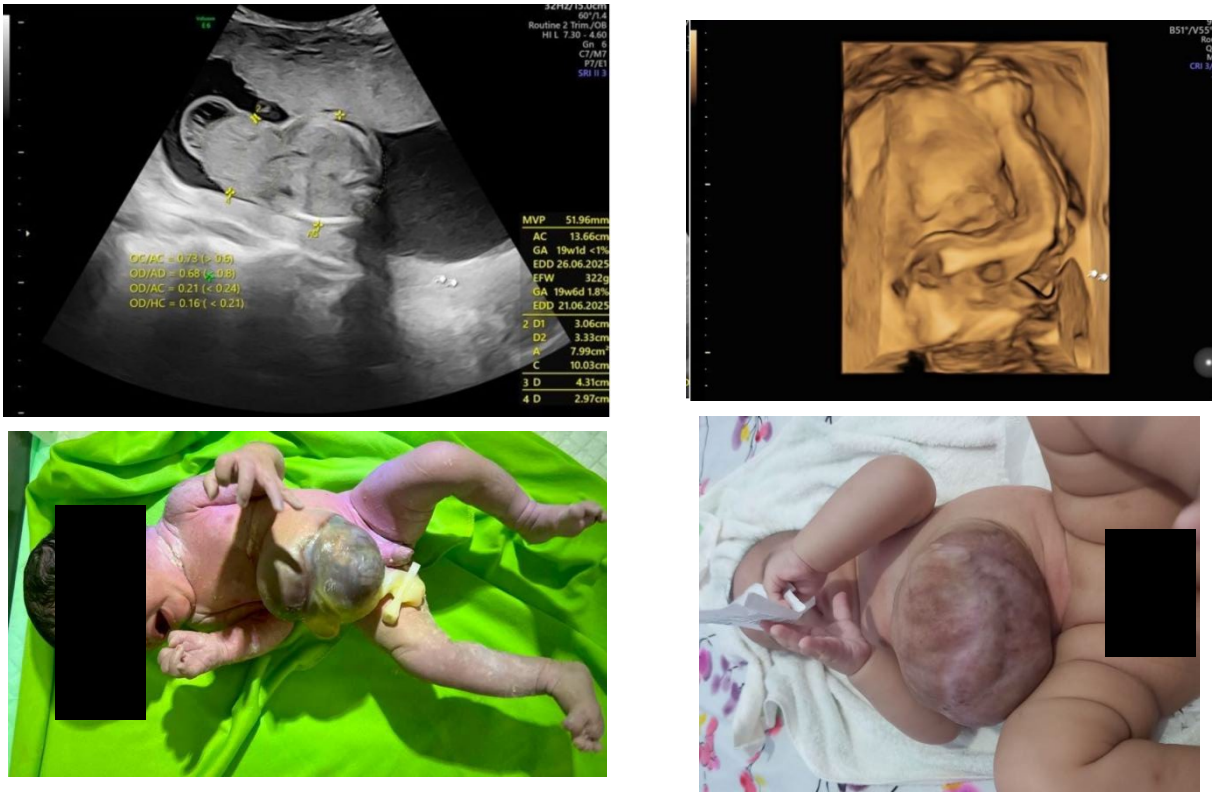

Figure 1. Case presentation. Upper left: omphalocele as seen on ultrasonographic screening; upper right: three-dimensional reconstruction of anterior abdominal wall defect; lower left: neonate showing large omphalocele; lower right: neonate approximately one month after successful repair of omphalocele

The last routine ultrasonography follow-up was conducted 36th week of pregnancy. During the ultrasonography, we discovered singleton live pregnancy, EFW 2236 grams, with anterior omphalocele measured approximately 6.92 cm accompanied with gastric herniation. Due to the history of previous cesarean section and large (6.92 cm defect with gastric herniation), the patient was planned to undergo cesarean section.

At 37th week of pregnancy, the patient underwent cesarean section in a standard Pfannenstiel fashion. We discovered the existence of live singleton male fetus, 2380 grams, APGAR 8/9 with visible omphalocele containing abdominal organs covered with peritoneum. The patient was subsequently handed over to neonatal team for intensive care and planned repair by the team of pediatric surgeon at approximately 8 months of age due to the extent of the defect with the defect was covered with sterile dressings. The patient and the neonate were discharged uneventfully. At the point of this case report was written, the infant was approximately 6 months of age and thus anterior abdominal wall defect repair has not been conducted.

3. Discussion

Approximately 50-70% of all omphalocele exist as a part of other congenital defects, particularly syndromes involving midline defects. In our case however, we discovered no other congenital anomalies, suggesting the existence of isolated case of omphalocele in our case. Omphalocele occurs in approximately 2.6 per 10 thousand live births and slightly more frequent in male offsprings. Nevertheless, approximately 25% of all omphalocele in a study in Indonesia was related to other congenital anomalies.[2,7,8]

As a type of midline defect, several factors have been implicated in the occurrence of omphalocele, including exposure to teratogenic agents, genetic and chromosomal disorders, folic acid deficiency, salicylic acid exposure, and idiopathic causes.[2,5,6] Unremarkable history of our patient nevertheless suggested that this case was purely idiopathic, with no known exposure to risk factor. Approximately one-third of all omphalocele occurs in isolation, while the rest occurs in conjunction with other anomalies. The outcome of omphalocele tends to be good when no other congenital anomalies are present, although approximately 32.1% of all omphalocele cases resulted in the neonatal demise when omphalocele is a part of syndromic anomalies. In a study conducted in Indonesia, approximately 50% of all omphalocele cases resulted in the neonatal death.[7–9]

Several embryological factors have been implicated in the occurrence of omphalocele. Particularly significant however, disorder in the cellular migration and fusion of embryonic folds during the 4th-7th week of pregnancy was implicated in the pathogenesis of the omphalocele. In the 6th week of pregnancy, due to growth of intestinal structures outpacing the development of abdominal wall, physiologic herniation occurs. Nevertheless, at approximately 10th week of pregnancy, recession of intestinal structures occurs and the intestine is covered by the newly formed abdominal wall. this highly concerted migration involves several types of cellular signaling processes.[5,10,11]

Thus, omphalocele is thus highly related to other midline defects, including intestinal stenosis and atresia, Beckwith-Wiedemann syndrome, and increased risk of several types of germinal cell tumors due to chromosomal or genetic abnormalities in variety of cases of syndromic omphaloceles. Unfortunately, the exact embryological sequence leading to development of omphalocele is far from fully elucidated and further research are still required.[2,5]

Due to the high risk of neonatal comorbidities, including infection, immediate repair of omphalocele thus becoming a major aim. In our case however, feasibility of primary repair of giant omphalocele are limited. In those cases, omphalocele repairs may be conducted in stages or delayed with similarly excellent outcomes.[12–14] Nevertheless, with the risk of serious adverse events, it is imperative to manage omphalocele cases in conjunction with pediatric intensivist and pediatric surgeon to provide best possible outcomes.

This study is limited with the unavailability of genetic or karyotyping testing due to the cost constraints. Nevertheless, we conducted serial prenatal ultrasonographic screening to rule out any gross congenital anomalies. Unfortunately, chromosomal or genetic anomalies cannot be ruled out entirely. In addition, at the point of this article is written, definitive surgical repair has not been conducted thus we are unable to report the final outcome of this interesting case.

4. Conclusion

Omphalocele is a type of rare congenital anomaly due to defect of anterior abdominal wall closure. Early detection resulted in planned delivery and subsequent prompt evaluation and treatment by the neonatologist and pediatric surgeon. Planned repair for omphalocele defect will be conducted at approximately 8 months of age in accordance with consultation with pediatric surgeon.

5. Informed consent

Written consent for publication of the journal and any accompanying image has been obtained from the patient and provided to the editor.

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