Greener Journal of Medical Sciences
Vol. 11(2), pp. 246-248, 2021
ISSN: 2276-7797

https://gjournals.org/GJMS

Giant Omphalocele With Multiple Major Structural Anomalies In A Primipara At 25 Weeks Gestation: A Rare Case Report.

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ARTICLE INFO

Article No.: *112121129*
Type**:** *Research*
Full Text: *HTML, EPUB*

Accepted: *23/11/2021*
Published: *30/11/2021*

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Keywords: *Giant Omphalocoele, congenital anomaly, folic acid, prenatal diagnosis.*

ABSTRACT

Background: Omphalocele is a major congenital anterior abdominal wall defect. It is often associated with other congenital anomalies that contribute to an increase in perinatal morbidity and mortality. The diagnosis could be made earlier in pregnancy with routine antenatal ultrasonography or prenatal diagnosis. Following this, termination may be offered.
Objective: To report a case of omphalocele with major structural abnormalities in a primipara at 25 weeks gestation.
Methods: The case note of the patient and how she was managed were reviewed. A relevant review of the literature on the subject was also done.
Case report: Mrs OJ was a 25year-old primipara who presented for booking at 25 weeks of gestation. she had an ultrasound scan at booking and the foetus was incidentally diagnosed as a case of omphalocele with associated major structural anomalies. She had not received folic acid in the index pregnancy and had no other risk factors. The pregnancy was terminated with misoprostol tablets following counselling. The baby was a fresh stillborn with giant omphalocele associated with gastrointestinal, renal and limb abnormalities.
Conclusion: The incidence of omphalocele could be reduced by routine use of folic acid around the time of conception and in the first trimester. However, most women in developing countries book late in pregnancy when prevention is not possible. There is a need to educate women on early booking, improve access to antenatal care and supplement food with folic acid for women in the reproductive age group.

1. INTRODUCTION:

An omphalocele is a defect of the umbilical ring resulting in herniation of the abdominal viscera covered by a membrane. It is a rare and serious major congenital defect of the anterior abdominal wall[1]. However, it is one of the most common anterior wall defects. The covering membrane consists of peritoneum on the inner surface, amnion on the outer surface and Wharton's jelly between the layers[1,2]. The umbilical vessels insert into the membrane[1,2]. Giant Omphalocole occurs when the anterior abdominal wall defect is greater than 5cm with the sac containing the liver [3]. Omphalocele occurs in 1-3 per 10,000 births[4] and is often associated with other congenital anomalies which contribute to a significant increase in perinatal morbidity and mortality[2,5]. Early

diagnosis is possible with routine antenatal ultrasonography, magnetic resonance imaging, or prenatal screening for elevated maternal serum alpha-fetoprotein and testing for acetylcholinesterase in the amniotic fluid [4]. We report a rare case of giant omphalocele with major structural anomalies diagnosed at the booking visit in a primipara at 25 weeks gestation.

2. CASE REPORT:

Mrs OJ was a 25year old G2P1 who presented for booking at the antenatal clinic of Rivers State University Teaching Hospital (RSUTH) on 20th October 2021 at 25weeks of gestation. She had no complaints though was not on routine folic acid and ferrous supplements. She neither smoked nor took alcoholic beverages. She was not a known diabetic, hypertensive or epileptic and had no family history of a congenital anomaly; her first baby was normal.

An obstetric ultrasound scan done at booking showed anterior abdominal wall defect in the foetus with floating abdominal viscera, absent kidneys, left club foot, severe oligohydramnios and growth restriction. Other investigations done were normal.

She was counselled on the diagnosis and management options. Consent was obtained for termination of pregnancy. She was admitted into the ward and the pregnancy was terminated using 200ug of misoprostol tablets inserted 6hourly into the posterior vaginal wall. She received three doses and subsequently delivered a fresh stillborn baby weighing 0.92kg 14 hours following admission. The foetus was born with omphalocele, multiple congenital anomalies and indeterminate sex. The visible congenital malformations were huge anterior abdominal wall defect, herniation of the liver and intestine with covering membranes, absent right lower limb, clubbed left lower limb and multiple digital abnormalities.

After delivery, the mother was counselled to book early for antenatal care in a subsequent pregnancy and to take folic acid from three months before conception and in pregnancy. She was also counselled to avoid taking tobacco products and unprescribed medications in a subsequent pregnancy. She was discharged home on haematinics in good clinical condition.

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Figure 1:Images of the baby with giant omphalocele and associated anomalies

3. DISCUSSION:

Omphalocele is a major congenital anomaly characterised by midline anterior abdominal wall defect and herniation of the abdominal viscera covered with membranes [1,2]. The prevalence is 1-3 in 10,000 births. The risk factors include extremes of reproductive age, obesity, folic acid deficiency, smoking, alcohol, and first-trimester use of medications like sodium valproate, ibuprofen, selective serotonin reuptake inhibitors, marijuana and cocaine [1,4,6]. Mrs OJ was not on folic acid supplements before her booking visit to the antenatal clinic. Genetic analysis for aetiological research is recommended especially karyotype and chromosomal microarray [7-9]. This foetus had giant omphalocele with herniation of the liver and intestines covered with membranes. The eviscerated organs in omphalocele are predominantly intestines, liver or spleen and pancreas, rarely colon, ovaries and stomach are seen[9].

Omphalocele is often associated with structural, genetic or chromosomal anomalies though it can also occur as an isolated defect[4,5]. This case had associated structural anomalies which include absence of both kidneys, absent right lower limb, clubbed left lower limb and multiple digital abnormalities. The diagnosis could be with ultrasonography from the late first trimester or early second trimester [4]. Ultrasound scan as was done in this case can pick up omphalocele in 67.2% of cases [10-12]. Associated anomalies may be identified with ultrasonography or prenatal diagnosis as was discovered in the reported case [2,4,13].

Management of omphalocele is usually done in a tertiary facility and is modified by the presence of associated anomalies and complications[3,6]. Isolated omphalocele is usually managed conservatively till

delivery and the repair is done in the neonatal period either primarily, staged or by delayed repair [3,6]. In cases with associated major anomalies, termination of pregnancy may be offered [3,6]. Mrs O.J consented to termination of pregnancy due to multiple structural abnormalities.

Isolated Omphalocoele has a good prognosis. The presence of associated anomalies as in this case increases the perinatal morbidity and mortality [4,7-10]. The use of periconceptional folic acid has been reported to reduce the incidence of omphalocele [8]. Mrs OJ booked late for antenatal care and did not receive folic acid in the first trimester.

4. CONCLUSION:

Omphalocele is often associated with other congenital anomalies and has increased perinatal morbidity and mortality. The use of periconceptional folic acid and early diagnosis improves the outcome. There is a need for public awareness of this condition, early antenatal booking and nutritional supplementation with folic acid for women in the reproductive age group, especially in developing countries.

Disclosure of conflict of interest:

The authors did not have any conflict of interest in this case report

Acknowledgements:

Our profound gratitude goes to everybody who participated in the management of this patient.

Statement of informed consent:

As per university standard guidelines, participant consent and ethical approval have been collected and preserved by the authors.

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e this Article: Jumbo, AI; Nonye-Enyidah, EI (2021). Giant Omphalocele With Multiple Major Structural Anomalies In A Primipara At 25 Weeks Gestation: A Rare Case Report. *Greener Journal of Medical Sciences*, 11(2): 246-248**.**