International Journal of Academic Health and Medical Research (IJAHMR)
ISSN: 2643-9824
Vol. 5 Issue 4, April - 2021, Pages: 75-77

Antenatal Diagnosis Of Omphalocele: A Case Report And Review Of The Literature

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Abstract: *Omphalocele (exomphalos) is one of the most common abdominal wall defects. The size of the defect and the severity of the associated anomalies determine the overall morbidity and mortality. Routine prenatal screening and diagnosis of the abdominal wall defect and concurrent anomalies is important as it allows for effective prenatal counseling and optimal perinatal management. The purpose of this article is to discuss the approach to prenatal diagnosis and management of omphalocele.*

Keywords—omphalocele, exomphalos, prenatal diagnosis, prenatal management

1. INTRODUCTION

Omphalocele (exomphalos) is one of the most common congenital anterior abdominal wall defects. This midline defect which occurs at the umbilical ring often contains the midgut and other abdominal organs such as the liver ,spleen and gonads.(1)

The herniated abdominal contents are encased in a three layer covering: the outer amniotic layer, the middle Wharton's jelly layer and the inner peritoneal layer(2)

An omphalocele can range in size frome 2to 10 cm and about 40-80% of all cases will have at least one concurrent anomaly(1)

Associated anomalies include cardiac (7-47%), gastrointestinal (3-20%), genitourinary (6-20%), chromosomal (3-20%), musculoskeletal (4-25%) and central nervous system (4-30%)(1-3)

The size of the defect and the severity of the associated anomalies impact the pre- and postnatal morbidity and mortality(4-5)

Early detection allows for prenatal counseling and safe delivery at a tertiary care center with a multidisciplinary team involving neonatologists, obstetricians and pediatric surgeons. The purpose of this article is to discuss the approach to prenatal diagnosis and management of omphalocele.

2. CASE REPORT :

This is a 28-year-old patient, G2 P1, with no notable pathological antecedents, without consanguineous marriage admitted to the maternity hospital emergency room of the Hassan II Hospital of FES for the management of a fetal malformation discovered during a screening ultrasound done for the first time in the second trimester since the pregnancy was not followed.

The obstetrical ultrasound showed the presence of an omphalocele in a pregnancy of 24 weeks of amenorrhea

the pregnancy was followed up without any particularity with a scheduled delivery at 39SA after consultation with the pediatric surgeons and neonatologists

image 1: showed the presence of an omphalocele in a pregnancy of 24 weeks of amenorrhea

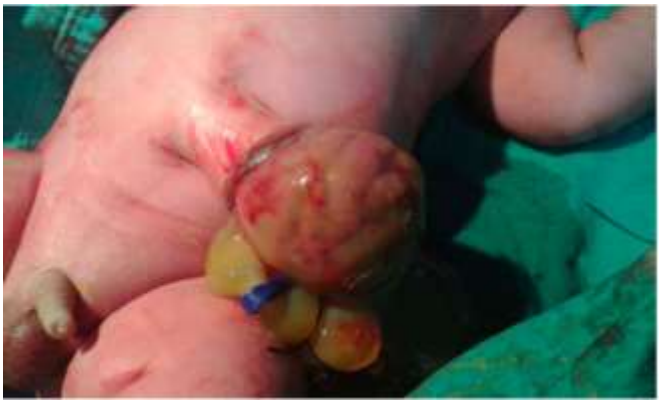

image 2: showed newborn male with omphalocele

3. DISCUSSION

omphalocele is one of the most common congenital anomalies of the abdominal wall.

The incidence rate of omphalocele is 1 to 2 per 10 000 live births (2-6-7).

Early antenatal diagnosis of omphalocele allows to reduce the number of polymalformed newborns by therapeutic interruption of pregnancy and to improve their management in the perinatal period.

In recent years, real progress has been made with the improvement of the different diagnostic methods.

The use of endo-vaginal ultrasound allows for an authentic fetal morphological examination at the end of the first trimester of pregnancy. The optimal period is between the 12th and 14th week of amenorrhea (SA). The main fetal structures are then in place and are accessible to ultrasound examination. A large number of pathologies can be detected at this time. First, the normal first trimester examination and its conduct are described. Secondly, the fetal pathologies accessible to an early diagnosis are evoked system by system, and the performances of ultrasound are discussed. It is important to note that the technical possibility of a diagnosis in the first trimester does not imply that this diagnosis is recognized as one of the objectives of screening ultrasound. (8)

Indeed, it is sometimes only possible to make a diagnosis under optimal technical conditions, as the overall sensitivity of ultrasound for the anomaly in question remains limited. Moreover, the identification of an anomaly in the first trimester does not always lead to a clear prognostic evaluation and may raise difficult questions about the course of action. (8)

Examination of the abdominal wall is possible as early as the 12th week of intrauterine life by ultrasound (9).The presence of an umbilical hernia before complete reintegration of the primary bowel loop is physiological before 12 weeks of amenorrhea (9), but its diameter is always less than 10 millimeters (9).

Ultrasound can be performed abdominally, but the endo-vaginal approach is often more accurate.

The ultrasound diagnosis of an omphalocele combines the following elements:

-the presence of a hyperechoic mass, at the base of the umbilical cord median rounded anterior, with extra abdominal development, connected to the abdominal wall by a collar. This mass is limited by a membrane that is sometimes difficult to detect, but its contours are clear. Its size and content are variable (mixture of fluid and echogenic images depending on the organs present in the
organs present in the sac) (10) ;

-The viscera contained in the omphalocele can be intestinal anses, liver and stomach
liver and stomach;

-The insertion of the umbilical cord is done at the lower part of the omphalocele sac.
of the omphalocele. The location of the vascular elements can be helped by examination in
color DOPPLER mode;

-The presence of an ascites layer is possible, indicating an in-utero rupture of the
of the omphalocele. It is located in the fetal abdomen and/or in the omphalocele itself (10).

Ultrasound can provide useful information in determining the prognosis of this malformation (8): the presence of hydramnios and associated malformations are factors of poor prognosis (11).

When the ultrasound diagnosis of omphalocele is made, it is necessary to look for associated malformations and chromosomal abnormalities which are frequent and will influence the fetal prognosis.

When the omphalocele is associated with other serious malformations, medical termination of the pregnancy will be proposed, or the omphalocele is isolated or associated with a minimal malformation, a fetal karyotype must be performed given the frequency of associated chromosomal anomalies. (12)

The size and content of the omphalocele should also be determined by ultrasound because large omphalocele require more difficult and sometimes multistage surgical closure.

In total, the antenatal diagnosis of omphalocele is currently based on ultrasound which guides :

- The positive diagnosis.
- The malformative assessment.
- Antepartum management.
- Delivery methods: In general, the obstetrical management of an isolated omphalocele is without particularity and the delivery is done by vaginal delivery.

omphalocele is a soft swelling and does not present an obstacle to the progression of the fetus in the genital tract, except for very large omphaloceles which sometimes require a caesarean section. (2)

Magnetic resonance imaging is a non-invasive and harmless technique. Thanks to its high resolution, it allows the diagnosis of fetal malformations, in particular those of the anterior abdominal wall and associated malformations, and contributes to the therapeutic decision.

However, it is an expensive exploration whose practice is limited to a few hospitals.

AMNIOCENTESIS: will search by puncture and analysis of the amniotic fluid, chromosomal abnormalities associated or not with the omphalocoele. This will give a prognostic idea.

Omphalocele is found in the context of certain genetic anomalies: trisomy 13-18-21, aneuploidy, triploidy (13).

As soon as the child is born, the omphalocele should be covered with a sterile small bag or, failing that, with a sterile bandage to protect against trauma, infection, necrosis and desiccation. The immediate management aims to prevent certain complications related to the omphalocele such as hypothermia, digestive distension, hydro-electrolyte imbalance and infection or to its reintegration such as hemodynamic and respiratory complications and to carry out a good preoperative assessment.

4. CONCLISION

The prenatal diagnosis of omphalocele remains significant. Additionally, early detection of other associated structural or chromosomal anomalies remains paramount as this has a direct correlation to the prenatal and postnatal prognosis. However, a significant number of omphalocele cases may

still be diagnosed with an additional anomaly after birth. Therefore, particularly for large omphaloceles, timely prenatal counseling and management by a team of neonatologists, pediatric surgeons and obstetricians at a tertiary referral center with a neonatal intensive care unit and immediate access to surgical services will minimize the morbidity and mortality associated with this devastating congenital defect.

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