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**CASE REPORT**

# Omphalocele

Muhammad Johar Nafis[1], Yusrawati[2]

***Affiliations** : 1. Resident of Obstetrics and Gynecology, Faculty of Medicine, Andalas University, Dr. M. Djamil Central General Hospital Padang; 2. Sub Division of Maternal Fetal Medicine, Obstetrics and Gynecology Department, Faculty of Medicine, Andalas University, Dr. M. Djamil Central General Hospital Padang*

***Correspondence:** Muhammad Johar Nafis, email : johar.nafis88@gmail.com, Hp: 085357198828*

***Abstract***

***Objective**: Reporting a case of omphalocele.*

***Method**: Case report.*

***Result**: A case of a 24 years-old woman, diagnosed with G1P0A0H0 37-38 weeks of term pregnancy + omphalocele. According to ultrasonography, biometric measurements of the fetus were: BPD: 8.4 cm, FL: 6.7 cm, HL: 5.9 cm, AC: 30.5 cm, AFI: 8.7 cm, there were no image of abdominal wall, but a membrane surrounding the abdomen was visible. From the USG examination, the conclusion was Omphalocele. Chromosomal analysis was done using G-Comparative technic, with result 46, XY, which means the amount of the chromosomes was 46, and sex chromosomes were XY. There was no major structural defect. Mostly, the disorders causing these conditions are 13, 18 and 21 trisomy, and Beckwith-Wiedemann Syndrome.*

***Conclusion**: Omphalocele is a genetic disorder marked by failure of the abdominal organs to return into the abdominal cavity. It requires thorough and accurate prenatal examinations to establish a diagnosis.*

***Keywords:** Omphalocele, prenatal diagnosis*

## INTRODUCTION

Omphalocele is a defect of the abdominal wall in the insertion of the umbilical cord with intestinal herniation or other contents of the abdominal cavity wrapped in a membrane / membrane. One other abdominal wall defect is gastroschysis, which is if the herniated organ is not covered by any membrane. Initially intestinal and other abdominal organs develop outside the abdomen and will return to the abdominal cavity at 10-12 weeks. But in patients with omphalocele the process of return to the abdominal cavity does not occur perfectly. Organs that do not return to the abdominal cavity are covered by a membrane / membrane.[1,2]

Omphalocele is a defect abdominal wall is most often found, which is about 1.92 per 10,000 live births. Newborns with omphalocele more male sex and born to mothers aged 35 years and over or 20 years and under. Infants with omphalocele are usually accompanied by other congenital abnormalities such as cardiac abnormalities (32%). In addition, omphalocele is also associated with chromosomal abnormalities, especially trisomy 13, 18, and 21, which are found in about 20-50% of cases, and often cause fetal death. The survival rate of isolated omphalocele (without comorbid or chromosomal abnormalities) is 96%, and this number



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drops dramatically when accompanied by other anomalies or abnormal karyotypes. Infant mortality (28.7%) which usually occurs within the first 28 days of life.[1,2]

The exact mechanism of the occurrence of omphalocele is still not fully explained. The abdominal wall is formed through the fusion of four plates, namely cephalic plates that form the thoracic wall and the epigastric part of the abdomen; caudal plates that form the perineum, bladder and hypogastric region; and two side plates that form the lateral sides of the abdomen.

The four meet in the middle and fused to form the umbilic ring in the 4th week. The primitive intestine undergoes rapid enlargement in the 6th week, which causes herniation of the umbilical ring and then goes through a process of rotation and reintegration in the 10th week. Abdominal wall defects are thought to be due to early disruption in the process of mesenchymal differentiation due to something not yet known.

Omphalocele can be diagnosed prenatally by ultrasound examination. Return of the intestine after physiological herniation at 10 weeks can be confirmed by ultrasonography in the 11th to 14th week together with nuchal translucency screening for Down's Syndrome.[4]

## CASE REPORT

A 24-year-old female patient, with a diagnosis of gravid preterm G1P0A0H0 37-38 weeks + omphalocele + IUGR. On ultrasound examination at 34 weeks gestation, fetal biometry was found; BPD: 84 mm, FL: 67 mm, HL: 59 mm, AC: 305 mm, AFI: 8.8 cm, protruding mass appears on the anterior abdominal wall.

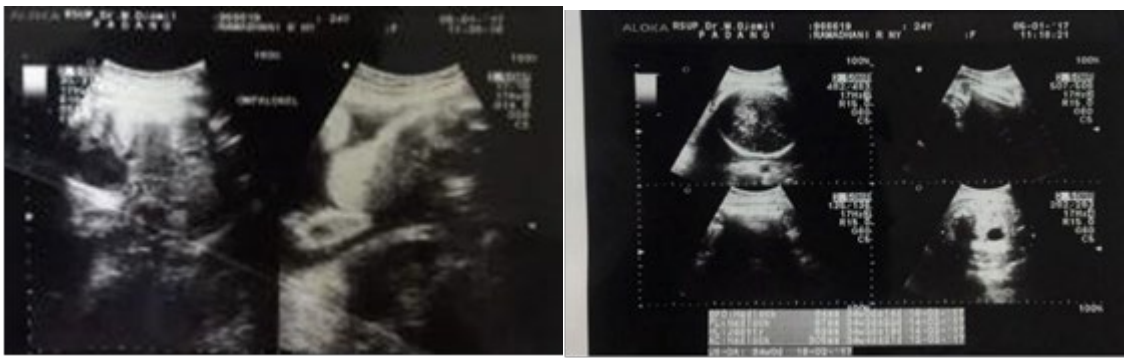

**Figure 1**. (a ) Omphalocele mass, (b) Fetal biometry

Chromosome Analysis Test carried out by the G-Banding technique has been studied chromosomes from 20 cells and get the number of chromosomes in each cell studied is 46, XY. Which means the number of chromosomes is 46 units with the patient's sex chromosome is XY. No major structural abnormalities appear.



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Figure 2. Chromosome 46, XY

Born baby boy, body weight 2200 gr, body length 39 cm, AS 8/9, visible contents of the abdominal cavity wrapped in protruding membranes from the abdomen (omphalocele).

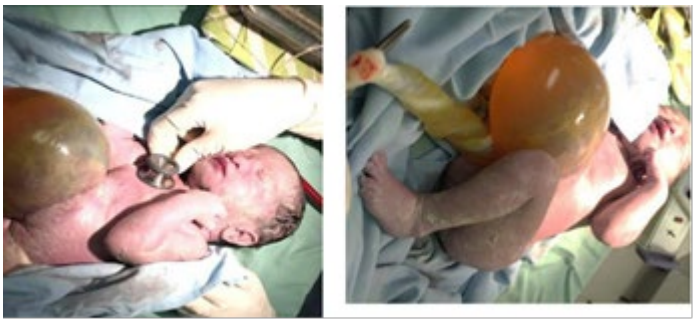

Figure 3. Omphalocele

## DISCUSSION

An Omphalocele case was reported. The diagnosis is made by careful and repeated antenatal ultrasound examination. Based on this examination fetal biometry results were obtained; BPD: 84 mm, FL: 67 mm, HL: 59 mm, AC: 305 mm, AFI: 8.8 cm, visible mass protruding out of the anterior abdominal wall.

In some of the literature mentioned that prenatal diagnosis can be done by ultrasound examination. On this examination it can be found: the appearance of a mass with a sharp border protruding out of the anterior wall abdomen and sometimes accompanied by polyhydramnios.[2,5]

In this case, the antenatal ultrasound results were consistent with the image of an omphalocele patient in which a well-defined mass protruded from the surface of the anterior abdominal wall. The basis of the defect usually comes from the insertion of the umbilical cord. Herniation of intra-abdominal organs usually consists of intestines sometimes accompanied by other organs.

The chromosome analysis test was performed using the G-Banding technique. The chromosomes of 20 cells were studied and the chromosome number in each cell studied was



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46, XY. Which means the number of chromosomes is 46 with the patient's sex chromosome is XY. No major structural abnormalities were seen.

However, there are several chromosomal abnormalities that usually accompany omphalocele events, the most common being trisomy 13, 18 and 21 and with Beckwith-Wiedemann Syndrome. In addition there are several other chromosomal abnormalities that are rare, namely, Turner syndrome, Klinefelter syndrome and Pallister-killian syndrome.[5]

Another examination that can be done while the patient is antenatal control is MRI. In contrast to ultrasound which is highly operator dependent, MRI is not. The field of view obtained from MRI is also broader than that obtained through ultrasound examination. A wide field of view will provide a good anatomical orientation. MRI was also unaffected by maternal obesity and oligohydramnios. However, fetal motion can affect MRI.[5]

If a case of omphalocele was diagnosed before delivery, carry out a thorough examination to check for other congenital congenital abnormalities. The choice for termination of pregnancy in this patient is cesarean section. The cesarean section was chosen to avoid trauma during the delivery process of the omphalocele mass, because if the delivery is done vaginally, when the baby is born where there is an omphalocele mass, labor will be obstructed and if it is forced it can result in injury or trauma to the omphalocele mass itself.[5]

After the baby is born, the possibility of other congenital abnormalities such as heart defects (occurs in 50% of cases), central nervous system disorders, urogenitalia (bladder exstrophy, cloacal exstrophy) and skeletal abnormalities is not found. other comorbidities in infants. Furthermore, this baby has also been performed three times by the pediatric surgery department, namely, two operations for the treatment of hernias in January and February 2018 and one operation to close the omphalocele defect in April 2018.

## CONCLUSION

Omphalocele is a defect of the abdominal wall in the insertion of the umbilical cord with herniation of the intestine or other contents of the abdominal cavity that are covered with a membrane / membrane. The prenatal diagnosis of omphalocele can be confirmed by ultrasound examination. It takes collaboration from multidisciplinary science for handling the baby.

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