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CONGENITAL DEFECTS OF THE ANTERIOR ABDOMINAL WALL AND DIAPHRAGM

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Abstract: *Congenital defects of the anterior abdominal wall and diaphragm are malformations occurring during embryogenesis and include gastroschisis, omphalocele and diaphragmatic hernia. Diaphragmatic hernia is often accompanied by pulmonary hypoplasia that causes fetal asphyxia with varying degrees of severity. The purpose of this study was to evaluate the prevalence of congenital malformations of the anterior abdominal wall and diaphragm in the Clinical Hospital of Obstetrics and Gynecology Brasov between the years 2011 and 2017. The 29 cases included in the study range in severity from minor malformations, easily corrected through surgical procedures to severe malformations associated with other congenital anomalies that are incompatible with life. The most common congenital defect encountered in our study is diaphragmatic hernia, which can be diagnosed through correct follow up of the pregnancy, monthly ultrasounds and screening tests.*

Key words: *omphalocele, gastroschisis, diaphragmatic hernia, abdominal wall defect.*

1. Introduction

Congenital defects of the anterior abdominal wall and diaphragm are malformations occurring during embryogenesis. Omphalocele and gastroschisis are the most frequent defects of the anterior abdominal wall, with a prevalence of 3 in 10,000 cases [7], [20]. Congenital diaphragmatic hernia is a congenital malformation of the diaphragm. This is uncommon and frequently associated with pulmonary hypoplasia and persistent pulmonary hypertension [14]. Eighty-five percent are left sided hernias, but the commonest form of congenital diaphragmatic hernia is the classic posterolateral or Bochdalek hernia [5]. Omphalocele is a large defect of the abdominal wall, in which the viscera coming out of the abdomen always has a sac and occurs because the rectus muscles are inserted laterally on the costal margins. It often has other associated abnormalities and the main cause of producing this malformation is a failure of closure of a primary body fold [11]. Gastroschisis is a defect of the anterior abdominal wall in which the viscera never has a sac. Also, the rectus muscles are inserted at the xiphoid.

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One hypothesis about the error produced during embryogenesis is that gastroschisis develops because the umbilical coelom fails to form. As a result, it forces the elongating midgut to rupture into the amniotic cavity [11]. Nowadays, the closure of this abdominal wall defects is possible even in multiple stages, as well as the management of most of the associated malformations [7]. Also, the management of these congenital defects is a great challenge in case of large defect, ruptured sac or if it is associated with other congenital abnormalities [1].

2. Objectives

The objective of this study was to evaluate the incidence of newborns with congenital defects of the anterior abdominal wall and diaphragm recorded in the Clinical Hospital of Obstetrics and Gynecology Brasov.

3. Material and Methods

This retrospective study has been carried out in the Clinical Hospital of Obstetrics and Gynecology Brasov and included 29 cases of newborns diagnosed with congenital defects of the anterior abdominal wall and diaphragm during the 1st of January 2011 to the 30th September 2017. The database used in this study has been composed of data obtained from the birth logs and from the patient charts, including mothers and newborns charts. Statistical results and graphic representation have been completed using Microsoft Office Excel.

4. Results

During the period of the study, there have been registered 27755 newborns, of which 29 (0.1%) presented varied types of congenital defects: omphalocele, gastroschisis and congenital diaphragmatic hernia.

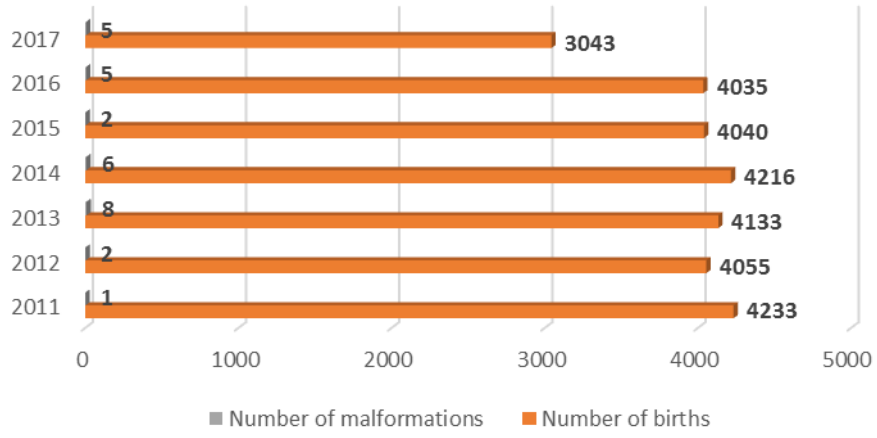

Fig. 1. *Incidence of congenital malformation (gastroschisis, omphalocele and diaphragmatic hernia)*

The incidence of these malformations varied from 2011 to 2017: in 2011 it have been registered 1 malformation (0.02%) from 4,233 newborns, in 2012- 2 cases (0.04%) from 4,055 newborns, in 2013- 8 cases (0.19%) from 4,133 infants, in 2014- 6 cases (0.14%) from 4,216, in 2015- 2 cases (0.04%) from 4,040 infants, in 2016-

5 cases (0.12%) from 4,035 newborns and in 2017- 5 cases (0.16%) from 3,043 infants until 30th of September. The highest incidence of infants with abdominal wall malformations was recorded in 2013 (0.19%) and the lowest in 2011 (0.02%). In figure 1 can be observed the incidence of congenital malformations.

From the total number (29 cases) of newborns with congenital malformations of the abdominal wall, 15 (51.72%) presented congenital diaphragmatic hernia, 9 (31.03%) gastroschisis, 3 (10.34%) omphalocele and 2 (6.89%) other unspecified abdominal wall malformations.

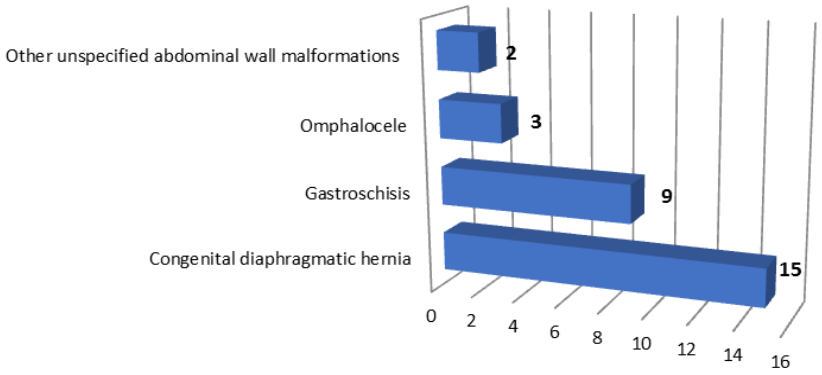

Fig. 2. *Distribution of congenital malformations of the abdominal wall*

Patients from the studied group mostly came from rural environment (16 cases- 55.17%) whereas from the urban environment, only 13 pregnant women (44.83%) were registered (figure 3).

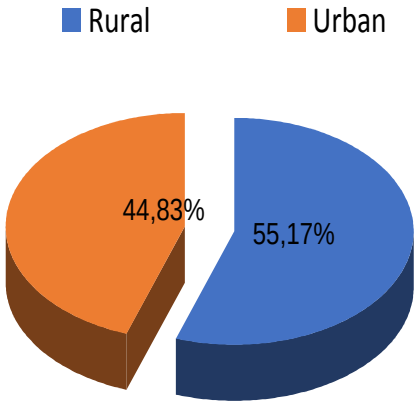

Fig. 3. *Distribution of cases according to the residence area*

It is to be noted that a higher number of cases came from rural area. This fact can indicate a possible causality relationship between the presence of malformations because of the lack of appropriate follow up of the pregnancies occurring in patients with a low socio-economic status that have not been registered by any medical caregiver during the pregnancy.

Another attempt of this study was to establish a causality relationship between the maternal age and the congenital abdominal wall defects. The analysis revealed that the maternal age varied between 13 years old and 44 years old. The majority of the fetuses with congenital abdominal wall defects had young mothers aged between 20-25 years old, the average age being 21.27 (Figure 4).

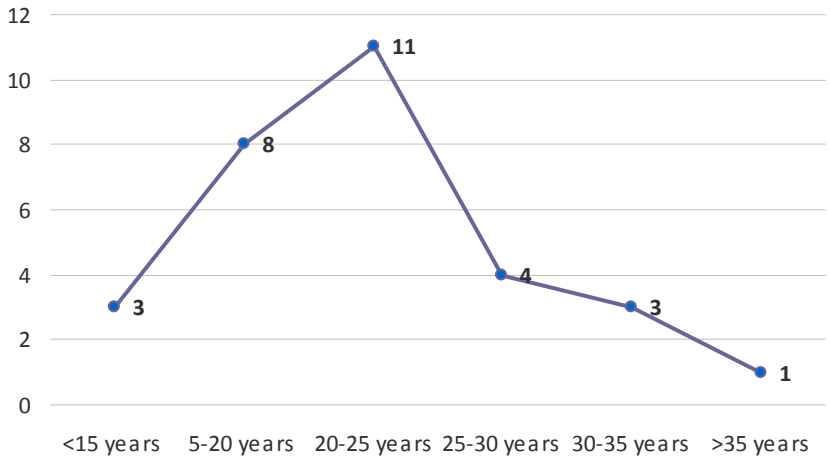

Fig. 4. *Distribution of the congenital malformations regarding the maternal age*

Educational level is another factor used in our study. Of the 29 pregnant women included in the cohort, 13 (44.82%) followed primary education and only 3 of them had higher education (10.34%). This fact may indicate that lack of education is a factor influencing the late discovery of pregnancies with malformed infants.

Most cases of congenital defects of abdominal wall and diaphragm have been found in women that were having their first or second pregnancy. 17 women (58.62%) were at their first pregnancy, 6 women (20.68%) were at second pregnancy, 4 of them (13.79) at third pregnancy and only one (3.44%) was multiparous with antecedents of malformed newborns (Fig. 5).

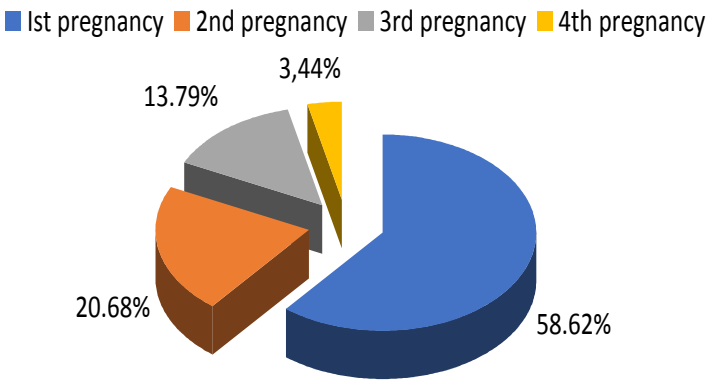

Fig. 5. *Distribution of maternal parity*

The way of delivery has also been studied. Of the 29 women in the cohort, most of them 19 (65.51%) underwent caesarian surgery and the remaining 10 (34.48%) gave birth via the vaginal route. The most common indications of caesarian surgery were: pelvic presentation at the primiparous (4 cases - 21.05%), uterine scar and fetal malformation previously diagnosed with oligoamnios or acute fetal distress.

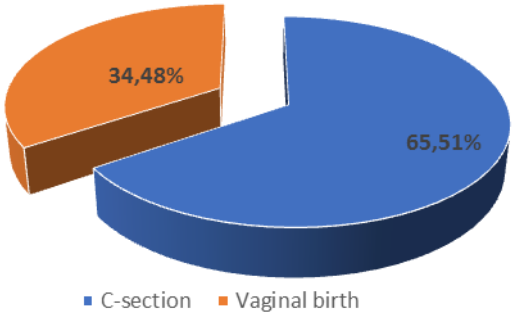

Fig. 6. *Distribution of the ways of delivery*

Regarding the gestational age, 16 of the infants were premature (55.17%), between 28 and 37 weeks of gestation. 6 of them were born between 37 and 39 weeks of gestation and 7 infants after 39 gestational weeks. Of the total number of malformed newborns, a number of 2 cases (6.89%) had weight between 1,000-1,499 g, 6 cases (20.68%) had weight between 1,500-1,999g, 8 infants (27.58%) had weight 2,000-2,499 g, 10 cases (34.48%) between 2,500-2,999 g, 2 newborns had weight between 3,000-3,499 g and only one weighted 3,800 g. The last one infant, who had weight 3,800 g, required at birth the use of vidextractor for the head recess. The distribution of the birth weight is illustrated in figure 7.

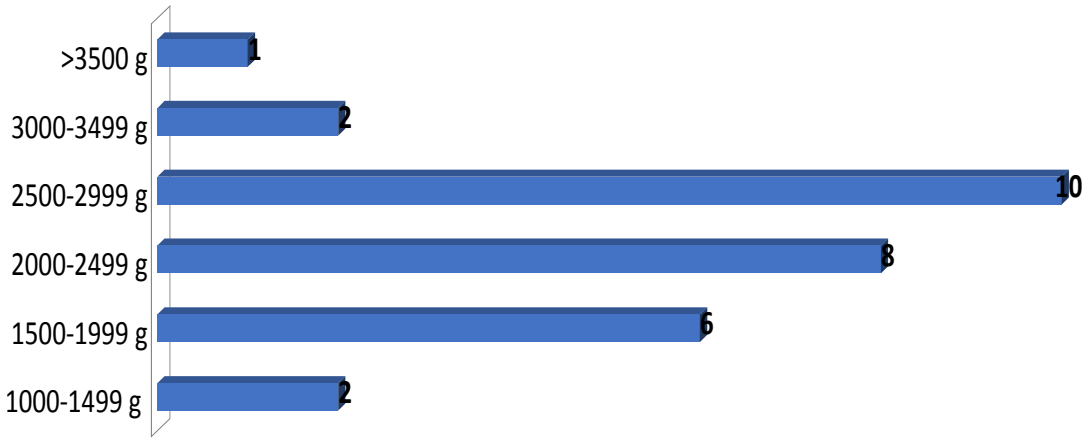

Fig.7. *Distribution of the birth weight*

Fetal gender has also been studied. From the total number of newborns with congenital defects of anterior abdominal wall and congenital diaphragmatic hernia, 16 infants were girls (55.17%) and 13 (44.82%) boys. Considering the

approximately equal number of the genders born with congenital abdominal wall defects with no statistically significant difference, fetal gender seems not to be an important factor in the study of these types of congenital abnormalities.

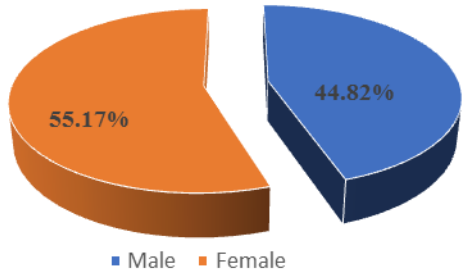

Fig. 8. *Distribution of fetal gender*

This cohort of 29 cases has also been evaluated from the perspective of the Apgar score. 31.03% (9 cases) of the babies had an Apgar score of 7. Apgar score 1 obtained 4 newborns and Apgar 9 obtained 4 infants.

5. Discussions

The congenital defects of anterior abdominal wall and the congenital diaphragmatic hernia is a widely discussed subject in scientific journals, because those malformations are significant causes of neonatal death [13]. Early diagnosis and treatment are of real importance as they can decrease fetal mortality and morbidity.

The incidence of these abnormalities varies depending on the region and maternal factors such as age, residence or race [16,17]. The incidence of gastroschisis, omphalocele and congenital diaphragmatic hernia in our study was 0.1 %. Mohamed M.A. and Aly H. performed a study based on the datasets from the Healthcare Cost and Utilization Project for the years 1997-2004. The incidence of congenital diaphragmatic hernia was 0.031% of the newborns compared to 0.051% registered in our study [17]. A study conducted in United States between 1995 and 2002 on 32 million births mentions a incidence of congenital diaphragmatic hernia of 1.93/10 000 births. This anomaly is induced by risk factors such as fetal male gender, maternal age beyond 40, smoking and consumption of alcohol use during pregnancy [3].

In the Czech Republic during the period of 1961-2000, a retrospective demographic epidemiological study was conducted in the Institute for the Care of Mother and Child from Prague. A total number of 2,293 cases of abdominal wall defects were registered including 1,450 cases of omphalocele and 843 cases of gastroschisis. The authors also found a significantly higher risk for omphalocele in women older than 39 years, and a higher risk for gastroschisis in women under 18 years and above 39 years [22]. The study of the Clinical Hospital of Obstetrics and Gynecology revealed a higher risk for congenital abdominal wall defects in women aged between 20-25 years old. Werler M.M. et al. sustained in their study the hypothesize that the defects are resulting from vascular disruption, produced by maternal vasoactive exposure, in the early embryo. But the vascular disruption was not supported by their findings [26].

For the total number of 29 cases, most of them were born prematurely (55.17%) under 37 gestational weeks. A similar retrospective study, based on 63 patients with congenital gastroschisis was conducted by Barseghyan K. et al. of these, 32 newborns (51%) was born prematurely [4].

Another study based on collected data from the Congenital Diaphragmatic Hernia Registry from January 1995 to July 2009, aimed to determine the impact of prematurity on survival of the fetuses with congenital diaphragmatic hernia. The total number of malformed infants was 5,022: 3,895 term infants (77.6%) and 1,127 preterm infants (22.4%). Preterm infants had also chromosomal abnormalities (4% term vs 8.1% preterm) and major cardiac anomalies (6.1% term vs 11.8% preterm). Survival rate decreased with lower gestational age (73.1% term vs 53.5% preterm) and a higher percentage of term infants were able to support a surgical intervention to repair the hernia (86.3% term vs 69.4% preterm).

The study concluded that more than 50% of preterm infants survived [25]. In Clinical Hospital of Obstetrics and Gynecology from 29 infants from the cohort, it has been registered 5 newborn deaths. Of these, only 2 were born prematurely at 35 respectively 36 weeks of gestation.

The complications associated with congenital defects of anterior abdominal wall and diaphragm varies. In our study, severe fetal asphyxia, pulmonary dysplasia, acute respiratory distress and severe intrauterine hypoxia are the main causes of fetal death. Within the cohort, along with omphalocele, gastroschisis and diaphragmatic hernia, congenital abnormalities such as dextrocardia, not descended testicle in the scrotum or malformations of the penis have been discovered. A study of Tarca E. and Aprodu S. was conducted over a period of 23 years and includes 105 children with omphalocele. Their conclusion was that omphalocele has a high frequency of associated congenital malformations (71.4%) and chromosomal abnormalities (27.6%) [24]. In literature it is also mentioned that major unrelated defects are associated with gastroschisis (10% of cases) whereas omphalocele is frequently associated with chromosomal or genetic abnormalities [20]. A study that included 88 cases of fetuses diagnosed prenatally with omphalocele identified 21 (24%) cases that were isolated and 67 cases with additional structural anomalies. Among newborns with structural abnormalities, 44 (50%) had chromosomal abnormalities and only 2 had omphalocele as solitary malformation [12]. Congenital diaphragmatic hernia consists in a congenital defect in the diaphragm, with herniation of abdominal contents into the fetal chest. The consequences of this herniation lead to varying degrees of pulmonary hypoplasia and pulmonary hypertension [2], [15,16]. The literature cites sources according to which antenatal administration of simvastatin improves pathological features of lung hypoplasia and persistent pulmonary hypertension of the newborn, in experimental nitrofen-induced congenital diaphragmatic hernia [16].

The way of birth chosen by the obstetrician for mothers with fetuses with congenital abdominal wall and diaphragm malformations is a subject discussed both in our study and in the literature. Of the 29 cases registered at the Clinical Hospital of Obstetrics and Gynecology Brasov, 19 pregnant women underwent a C-section and 10 of them vaginal birth. A meta-analyze including 15 studies based on the birth way in women carrying a fetus with abdominal wall defect was conducted to determine the most appropriate way of giving birth. The conclusion was that the available data do not provide sufficient

evidence to support the cesarean section in those cases [21]. Another study of Sipes S.L, Weiner C.P et al. concluded that the way of delivery did not affect the outcome for either omphalocele or gastroschisis [10], [18], [23]. A descriptive study from 1990 included 125 infants with abdominal wall defects: 56 cases of gastroschisis and 69 cases of omphalocele. Regarding infant mortality, frequency of associated anomalies and long-term infant outcome, it has not been demonstrated a significant difference between the two routes of delivery. Therefore, vaginal delivery of women carrying a malformed fetus does not adversely affect the infant outcome, although the C-section is preferred [18], [22].

The treatment of congenital defects of the anterior abdominal wall and diaphragm is a controversial topic, discussed in many studies in the literature [1]. In our study, discussions about the surgical treatment of these conditions were limited by the transfer of children to other hospital. A prospective intraoperative management protocol was developed to determine the successful primary closure of congenital abdominal wall defects (omphalocele/gastroschisis) in newborn infants, depending on the intragastrical pressure and central venous pressure. According to this protocol, infants who had an intragastrical pressure < 20 mm Hg or an increase central venous pressure of less than 4 mm Hg were primarily closed. If intragastrical pressure was greater than 20 mm Hg or if central venous pressure increased by more than 4 mmHg, the temporary closure of the abdomen was reopened and a prosthetic silo was placed [27]. A retrospective study that included a total of 94 children treated for omphalocele mentioned two ways to repair this type of congenital abdominal wall defect: the use of a silon chimney to reduce gradually the contents of the sac and a nonoperative management consisting in epithelialization of the omphalocele followed by repair of the residual ventral hernia. Fifty-five babies were treated by primary closure of the abdominal wall: silon chimney was used in 15 cases and 7 newborns had associated abnormalities that made the surgical intervention impossible [19]. Regarding the repair of congenital defects of diaphragm, several studies have been made on this topic. During 1995-1996, Congenital Diaphragmatic Hernia group included 62 centers and created a cohort with 461 babies with this congenital defect. The diaphragmatic defect was left-sided in 78% of cases, right-sided in 21% of cases and bilateral in 1%. 91% of patients were treated using a subcostal approach and pleural drainage was used in 76%. A patch of polytetrafluoroethylene was used in 51% of cases [5,6], [8]. Regarding to the material of the patch used to repair the diaphragmatic defect, it was demonstrated that the rate of recurrence of prosthetic patch repaired hernias was similar regardless of patch material (Surgisis or Gore-Tex) [8]. Fetal tracheal occlusion is a new procedure used in pregnant women carrying fetuses with congenital diaphragmatic hernia. According to *Michael R. Harrison et al*., tracheal occlusion did not improve survival rates in newborns with congenital diaphragmatic hernia [9].

6. Conclusions

The incidence of congenital defects of anterior abdominal wall and diaphragm identified in this study was 0.1%. The incidence of cases varied over the years, but the highest incidence was recorded in 2013 (8 cases - 27.58% of the total number of studied cases).

Correct follow up, screening tests and monthly ultrasounds are the key to early detection of these malformations and may decrease the perinatal mortality and

morbidity caused by the studied abnormalities. Regarding the environmental origin, the higher frequency of abdominal wall defects was registered among pregnant women in rural areas. This may suggest a possible relationship between low socio-economic status and lack of appropriate follow up of pregnancy. The low educational level of the pregnant women is also a contributing factor to the increased incidence of omphalocele, gastroschisis or congenital diaphragmatic hernia.

The main limitation of our study consists in the lack of information regarding the long term evolution of the infants with this anomalies diagnosed in Clinical Hospital of Obstetrics and Gynecology Brasov, because they followed surgical treatment in hospitals from other areas.

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