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A RETROSPECTIVE STUDY OF THE GASTROINTESTINAL CONGENITAL MALFORMATIONS

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Abstract: *A variety of congenital anomalies may occur during the embryological development of the gastrointestinal tract. These malformations include stenosis and atresia, duplications, abnormal rotation or fixation and abdominal defects. The purpose of the study is to evaluate the prevalence of the embryo-fetal gastrointestinal malformations diagnosed at the Clinical Hospital of Obstetrics and Gynecology Dr. I. A. Sbarcea Brasov. Anomalies have varied from minor malformations to severe ones, incompatible with life. Out of the 46 cases of digestive congenital malformations, the most frequent identified have been duodenal stenosis or atresia. Prenatal care for pregnant women, including screening tests and ultrasound, is crucial for diagnosing gastrointestinal malformations. A contributing factor to the rise in gastrointestinal malformations is also the level education of the pregnant women, as many of them do not request prenatal consultations.*

Key words: *congenital malformations, duodenal stenosis, screening.*

1. Introduction

A variety of congenital anomalies may occur during the embryological development of the gastrointestinal tract. These malformations are mainly represented by abnormal lumenisation that include stenosis and atresia, duplications, abnormal rotation or fixation and abdominal defects. Widely encountered are also the malformations associated with the persistence of embryonic structures such as the Meckel's diverticulum or abnormal formation of specific regions of the gastrointestinal tract such as microgastria [6], [19]. The gastrointestinal malformations are situated at the limit between embryology and pathology, such that they differ from postnatal lesions both through the way in which they have developed and the moment at which they occur [2], [13], [18]. The majority of these malformations are accompanied at birth by a variety of symptoms such as vomiting, lack or delayed meconium, difficulty in feeding. The high occurrence of gastrointestinal malformations suggests

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that a careful evaluation is required as to not omit any associated anomalies. [3]

2. Objective of the Study

The purpose of the current study was to evaluate the incidence of the embryo-fetal gastrointestinal malformations diagnosed at the Clinical Hospital of Obstetrics and Gynecology Dr. I. A. Sbarcea Brasov.

3. Material and Method

The retrospective study has been carried out at the Clinical Hospital of Obstetrics and Gynecology "Dr. I. A. Sbarcea" Brasov. The study includes 46 cases diagnosed with digestive tract malformations during the 1st of January 2005 to the 31st of December 2014.Data has been obtained from the birth logs and from the patient charts and compiled into a data base to be used for this study. Statistical results and graphic representations have been completed with the use of Microsoft Excel. The examinations were performed initially by more obstetricians but when an anomaly was identified, the women was referred and scanned by an experienced examiner in ultrasound from our hospital.

4. Results

During the period of the study, in the Clinical Hospital of Obstetrics and Gynecology "Dr. I. A. Sbarcea"" Brasov, there have been registered 44969 infants, of which 3432 (7.63%) have presented varied types of congenital malformations. The incidence of malformations has dropped from 482 (9.82%) in 2006 to 194 (4.55 %) in 2014.

Regarding the gastrointestinal malformations, a downtrend can be observed, as the maximum incidence is in 2005 (0.15%) and 2006 (0.14%). In figure 1 it can be observed the incidence of congenital malformations and gastrointestinal tract malformations.

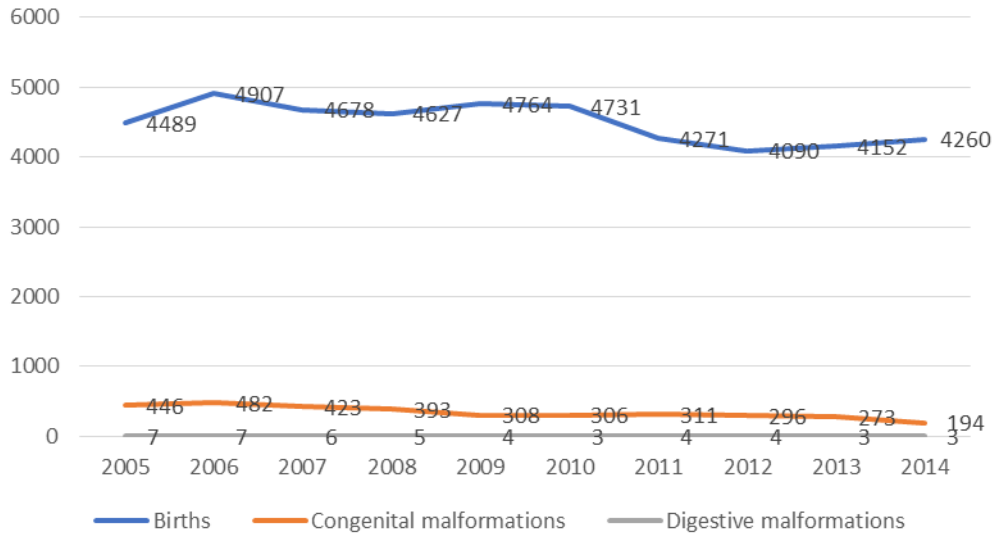

Fig. 1. *Incidence of congenital malformations and gastrointestinal tract malformations*

Anomalies have varied from minor malformations to more severe ones, incompatible with life. Out of the 46 cases of digestive tract malformations, the most frequent registered have been the duodenal stenosis or atresia. From the total number of born babies with digestive tract malformations we identified 8 (17.39%) cases of duodenal stenosis, 6 (13.04%) cases of duodenal atresia, 4 (8.70%) cases of esophageal atresia without transesophageal fistula, 4 (8.70%) cases of diaphragmatic hernia, 3 (6.52%) cases of imperforate anus and 3 (6.52%) cases of Hirschsprung disease, 5 cases of gastroschisis (10.85%) and 3 cases of omphalocele (6.52%).

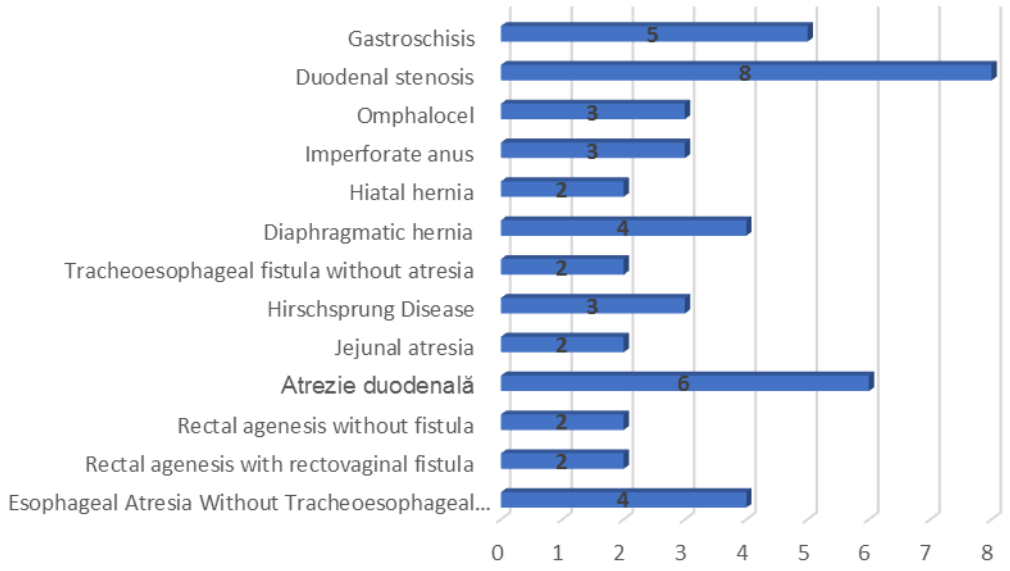

Fig. 2. *Distribution of gastrointestinal congenital anomalies*

The most frequent malformations have been located in the duodenum. Two of the cases with duodenal atresia have been found to also be associated with Down Syndrome.

Patients from the studied group mostly came from urban environment (60.87% - 28 patients), whereas from the rural environment only 39.13% patients were registered (18 patients), as it can be seen in the figure 3.

It is to be noted that a high number of cases come from urban environment. This indicates a possible correlation between the malformations and frequent exposure to trigger factors.

A significant number of pregnant women with malformed fetuses have not been registered by any medical caregiver during their pregnancy 34 (73.91%) patients.

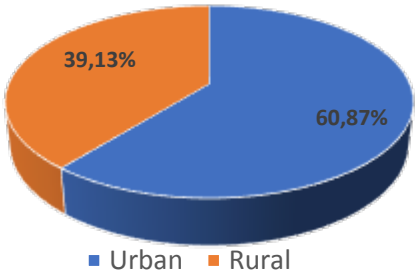

Fig.3. *Distribution of cases according to the residence area*

During the study, an attempt was to establish a causality relationship between various maternal factors (obstetrical case history, blood group, Rh, smoking, the consumption of vitamins) and fetal factors (sex, weight at birth, Apgar score) with the incidence of gastrointestinal tract malformations. The analysis revealed that the age of the patients was between 15 and 42, and the majority of the fetuses with digestive embryo-fetal malformations had young mothers with the age between 24-33, the average age being 27.5.

Most cases of digestive malformations have been found in women that were having their first or second pregnancy, representing a 60.8% percentage of the total born babies with digestive tract malformation.

Reported to the gestational age, 26 (56.52%) of the infants were premature, between 32 and 37 weeks of gestation. Of the total diagnosed cases with gastrointestinal tract malformations, a number of 9 cases (19.57%) had weight above 3000 g, 11 (23.91%) of cases had weight between 2500 and 3000 g, 18 cases (39.11%) between 1500-2500 gr , and 8 (17.39%) had weight under 1500 g , as it can be seen in Figure 4.

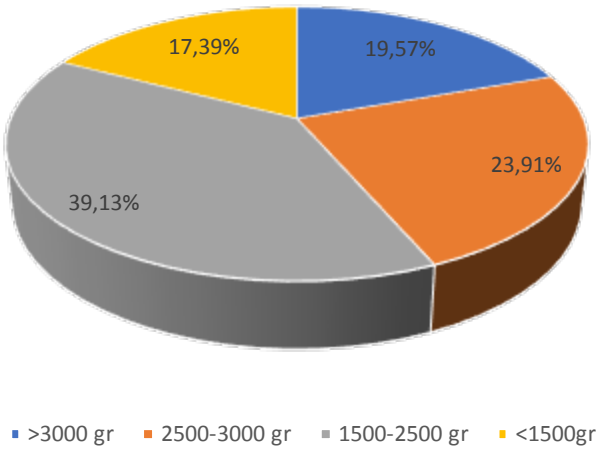

Fig. 4. *Distribution of the cases with digestive congenital malformation according to the birth weight*

The study group has also been evaluated from the perspective of the Apgar score, and it has been established that 80.4% of the babies had a score between 8 and 9. There have also been registered several cases of babies with a score between 7 and 6 (19.56%) due to severe malformations such as esophageal atresia with tracheo-esophageal fistula, trachea-esophageal fistula without esophageal atresia or hiatal hernia. The study also concluded that the gastrointestinal malformations have a higher occurrence in males (62.81%).

As per environmental risk factors, smoking and vitamin consumption has been also studied. A percentage of 36.96% have been reported to smoke. The consumption of vitamins during pregnancy was identified in 65.22% of the women.

5. Discussions

This subject is widely discussed in a variety of scientific journals, due to the high importance of diagnosis and treatment as soon and as efficiently as possible, thus lowering morbidity and mortality in infants with digestive tract malformations [16]. For this purpose, in 1980, in Europe, an epidemiological surveillance system has been developed – EUROCAT [5,7]. The incidence of these anomalies varies across countries, but has constantly maintained itself around the values of 5% to 10% in most of the reports [1,11].

A similar study has been performed by the University of Pecs in Hungary. It has the subject of the prevalence of gastrointestinal malformations during a period of 14 years (1987-2000). The results of the study concluded that out of 4241 infants cared for in the Pediatrics Department, 304 have presented gastrointestinal malformations (of which 133 cases had associated anomalies). The most common malformation found was the esophageal atresia with 18.75% compared to 10.77% registered in the Clinical Hospital of Obstetrics and Gynecology Dr. I. A. Sbarcea Brasov. Regarding the weight of the fetuses born with GI tract malformations, both studies have shown that more than half of them have had weights of less than 2500 g, which show an intrauterine growth restriction [17]. *Khoury et al* investigate the relationship between congenital malformations and intrauterine growth restriction, proving that 22.3% of infants have presented both diagnostics at birth, with the mention that pyloric stenosis has not been associated with excessive intrauterine growth restriction. The frequency of IUGR rises with the number of defects in infants with multiple malformations [10].

Another study, made in two development areas of Romania, in a period of 5 years (2003-2007), has monitored the prevalence of various types of embryo-fetal malformations. Data regarding the type of digestive tract malformations in infants has been registered for both regions and it was compared to the number of born infants registered in the database of the National Statistics Institute [8]. Thus, it has been concluded that from the entire lot of 1460 cases, the vast majority (35% meaning 511 cases) have been identified as malformations of the digestive tract out of which: biliary atresia – 15 cases, esophageal atresia - 58 cases, Meckel diverticulum – 23 cases, congenital defects of the abdominal wall – 120 cases, diaphragmatic hernia -18 cases, anorectal malformation – 57 cases, megacolon – 34 cases, duodenal stenosis – 94 cases [20]. The study concludes that the most common pathology is the congenital defect of the abdominal wall, followed closely by the duodenal malformation. The same conclusion arises from the study made in the Clinical Hospital of Obstetrics and Gynecology „Dr. I. A. Sbarcea" Brasov.

As mentioned, the study of the incidence of gastrointestinal tract anomalies has represented a study subject for a large number of clinical reviews. It is important though to study the incidence of these malformations in dynamics. The Department of Pediatric Surgery in Keimyung University, Korea has performed a clinical review on 343 infants and children diagnosed and treated surgically in their facility for congenital gastrointestinal anomalies. The period of the study ranged from January 1988 to December 1991. The conclusions of the study were that the largest number of cases had anomalies pertaining to the stomach (87 cases -25.4%). A high number of cases were reported also in the case of anomalies of the anus (80 cases -23.3%) followed by the colon (63 cases -18.4%) and biliary tract (38 cases -11.1%). Congenital hypertrophic pyloric stenosis was found to be the most common congenital anomaly (87 cases -25.4%). Also common was the imperforate anus (70 cases -20.4%), followed by congenital megacolon (63 cases – 18.4%). The ages of the infants were between 2 weeks and 3 months. It is important to mention that this review also concludes that the incidence of gastrointestinal malformations is higher in males than in females [15].

The differences that appear between the digestive tract anomalies can be observed in various locations across the globe, or at regional level, in the same country, which sheds a light upon geographic, cultural, educational and environmental factors that might influence the development of such anomalies.

Another aspect of major importance studied in the literature is represented by the factors that cause or influence the development of digestive tract malformations [9]. From all the external factors analyzed, smoking has had the higher impact, as it has registered in a increase proportion among the mothers of the infants with gastrointestinal tract malformations. This finding is correlated with conclusions from other studies [12], [14].

After a gastrointestinal malformation has been discovered through ultrasound tests, and confirmed through clinical and paraclinical data, advising the couple so that they can take the best decision is necessary [4]. When taking a therapeutic decision, a medical multidisciplinary team must contribute, regardless of the gravity of the malformation. This team should be composed of an obstetrician, a neonatologist, a geneticist and a psychologist, and the decision of the parents should be respected.

6. Conclusions

The frequency of the gastrointestinal embryo-fetal malformations identified in this study, from 2005-2014 has been 0.10%. The incidence of these cases has been constant during the 10-year period of the study, except for the year 2010, when a rise in the number of cases has been registered. Prenatal care for pregnant women, including screening tests and ultrasound, is crucial for diagnosing gastrointestinal malformations. Concerning environmental origin, it is to be noted that a high number of cases come from urban environment. This indicates a possible correlation between the malformations and frequent exposure to trigger factors. A contributing factor to the rise in gastrointestinal malformations is also the level education of the pregnant women, as many of them do not request prenatal consultations.

The study limitation was the lack of information regarding the postnatal outcomes of the diagnosed newborn with gastrointestinal pathologies, mostly because they followed surgical treatment in hospitals in other area then Brasov.

The future directions of the study include: analysis and prevention of the risk factors for gastrointestinal congenital anomalies and identification of the risk factors that influences the outcomes.

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