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Original Research Article

Prenatal diagnosis of fetal gastroschisis – The experience of Maternal-Fetal Medicine Centre in Olsztyn

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ABSTRACT

Introduction: Gastroschisis represents a congenital malformation of the anterior abdominal wall. It is a defect characterized by protrusion of the abdominal viscera through the full-thickness abdominal defect (peritoneum, muscle, fascia and skin).

Diagnosis of gastroschisis can be made as early as the first trimester and routinely in the second trimester by fetal ultrasound. Prenatal diagnosis of gastroschisis is important because it enables rapid and appropriate management both during pregnancy and after delivery.

Aim: The aim of this work is a retrospective analysis of fetal gastroschisis diagnosed in the Maternal-Fetal Medicine Centre in the years 1998–2011.

Material and methods: The analysis included 35 cases of fetal gastroschisis diagnosed in the Maternal-Fetal Medicine Centre in the years 1998–2011.

Results and discussion: The most numerous group of pregnant women, whose fetuses were diagnosed with gastroschisis, were young women below 35 years of age, more frequently residents of rural than urban areas, in most cases primiparas. Herniated contents most often contained small and large intestine. In 60% of fetal gastroschisis no complications were observed during pregnancy.

Conclusions: Early diagnosis seems to be of particular importance, as it allows monitoring for potential complications of gastroschisis, which may affect prompt decisions of elective preterm delivery, delivery at a referral center, and hence the possibility of preparing neonatal and pediatric surgical team.

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1. Introduction

Congenital malformations are morphological abnormalities acquired in intrauterine life and identifiable at birth. According to statistics, 2.0% of infants are currently born with at least one congenital defect,[1–3] in about 0.4% of newborns multiple congenital anomalies are identified, and in about 0.6–1.0% chromosomal aberrations are detected.[2,3] Congenital fetal anomalies are among utero-placental insufficiency and prematurity the most important risk factor for increased perinatal mortality.[1,2]

According to ICD classification, abdominal wall defects belong to the diseases of the musculoskeletal system, but most often are described together with abdominal disorders. The most common abdominal wall defects include gastroschisis and umbilical hernia.[4] These malformations may be isolated defects or part of complex alterations (pentalogy of Cantrell, Beckwith-Weidemann syndrome, cloacal exstrophy sequence, limb-body wall complex).[2,3,5,6] The most common concomitant disorder is intrauterine growth restriction. Parenteral defects occur in less than 5% of cases.[5,6]

Gastroschisis represents a congenital malformation of the anterior abdominal wall. It is a defect characterized by protrusion of the abdominal viscera through the full-thickness abdominal defect (peritoneum, muscle, fascia and skin).[2,3,7–9] Herniated contents usually include: small and large intestine, to a varying degree, less frequently: stomach and liver.[1,2,5,9,10] The estimated incidence is approximately 0.47–4.40 cases in 10 000 live births.[3] Reports in the literature indicate that gastroschisis occurs as a consequence of premature involution of the right omphalomesenteric artery or right umbilical vein at about 6 weeks gestation. This could explain the localized right abdominal wall defect.[2,7,11,12] Risk factors have not yet been precisely determined. Researches indicate that gastroschisis occurs more frequently in infants of young mothers, cigarette smokers, and possibly in case of maternal use of aspirin, pseudoephedrine, and acetaminophen in the early pregnancy.[13] In the majority of cases the etiology is sporadic, although familial incidence is also possible.[2] This defect occurs with the same frequency in fetuses of both sexes.[5,6]

Diagnosis of gastroschisis can be made as early as the first trimester and routinely in the second trimester by fetal ultrasound. Intestine loops can be seen floating freely in the amniotic fluid.[5,6,12] In rare cases stomach, less frequently liver or spleen may also protrude. Typically, the opening in the abdominal wall is about 2–4 cm in diameter.[9,11] It is important not only to diagnose gastroschisis, but also monitor its size, since reduced dimension may cause auto-amputation of the free intestinal loops. An important element of monitoring the condition of gastrointestinal tract is also the assessment of blood supply of the eviscerated intestines (Color Doppler). Presence of peristalsis and width of the extruded intestinal loops are also evaluated in order to monitor patency of the gastrointestinal tract.

Elevated level of maternal serum alpha-fetoprotein (AFP) may also play a role in the diagnosis of gastroschisis,[13] however it has a marginal significance due to unambiguous result of ultrasound examination, which is nowadays mandatory.

Favorable prognosis is associated with small amount of eviscerated intestinal loops, with preserved blood supply, not dilated, with present peristalsis. Unfavorable prognosis is limited to cases where large portions of small and large intestine are eviscerated, dilated, with no peristalsis and blood supply. Ultrasound findings resulting from the exposure of the uncovered intestines to the harmful effects of the amniotic fluid include: bowel wall thickening and hyperechoic fibrin deposits on its surface. Complications of gastroschisis that may occur during pregnancy include: ischemia, obstruction, stenosis, and intestinal perforation. Sonographic signs suggestive of intestinal perforation are: bowel wall thickening, peritoneal calcification, meconium ascites and pseudocysts.[5,6]

1.1. *Management during pregnancy*

Pregnancy with fetal gastroschisis should be continued to term. In case of worsening of the ultrasonographic signs with suspected obstruction (dilated loops, loss of peristalsis) early delivery may be considered. Usually cesarean delivery is performed, although data regarding the most favorable mode of delivery are unclear.[14] Regardless of the route of delivery, it should be conducted in a referral center with strict cooperation with pediatric surgery unit. After delivery, in the event of respiratory disorders in the newborn, endotracheal intubation is recommended. Eviscerated loops should be covered with a sterile bag in order to prevent fluid loss. Early surgical intervention aiming at restoring normal anatomic conditions, returning intestines back into the abdominal cavity and closing the abdominal defect, is the preferred approach.[15,16]

2. Aim

The aim of this work was a retrospective analysis of fetal gastroschisis identified in the Maternal-Fetal Medicine Centre (MFMC) in the years 1998–2011.

3. Material and methods

The analysis included a database of examinations performed in the years 1998–2011. During this period, 26 000 fetal ultrasound examinations were performed and in 35 cases gastroschisis was diagnosed. Analysis included maternal age and gestational age at time of diagnosis, geographical distribution, type of protruded contents, coexistence of other defects and complications.

4. Results and discussion

The most numerous group among pregnant women, in whom fetal defect was diagnosed, were young women – 21 women (60%) aged up to 25 (Fig. 1). Only 3 (8%) pregnant women were above 35 years of age.

Patients of MFMC were more frequently residents of urban areas (20 cases, 57%), than rural areas (15 cases, 43%). Slightly more than one-fourth of pregnant women lived in the district of Olsztyn, where the MFMC is situated (Fig. 2). The majority of

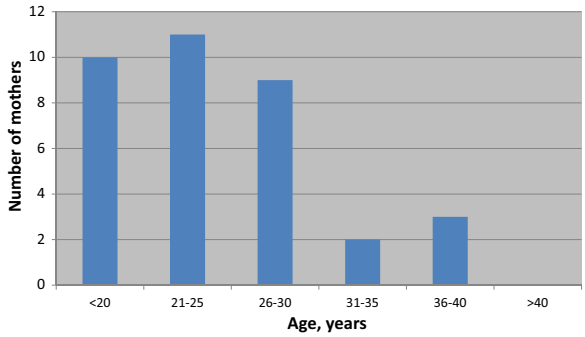

Fig. 1 – Age of mother at diagnosis.

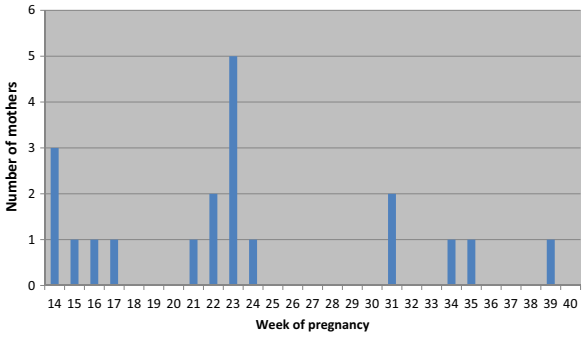

Fig. 4 – Week of pregnancy at diagnosis.

women were primiparas (22 cases, 63%) (Fig. 3). Almost all cases of gastroschisis included in the analysis were single pregnancies (33; 94%), there were two twin pregnancies in the analyzed material (6%). Most defects were diagnosed between 20 and 23 weeks gestation (9; 25%) (Fig. 4). In 22 (60%) cases the contents of evenetration was small and large intestine (Fig. 5). In 5 (14%) cases coexistence of other anomalies was observed, such as: ventricular septal defect, ectopic heart, and dilation of

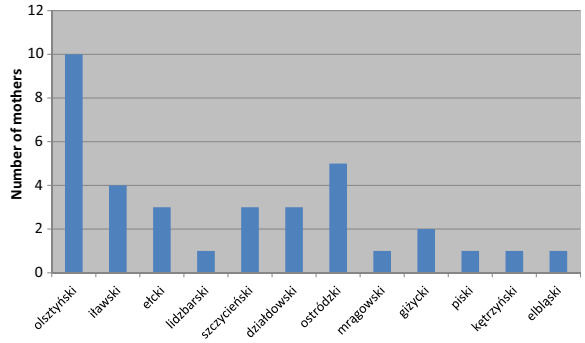

Fig. 2 – County of residence.

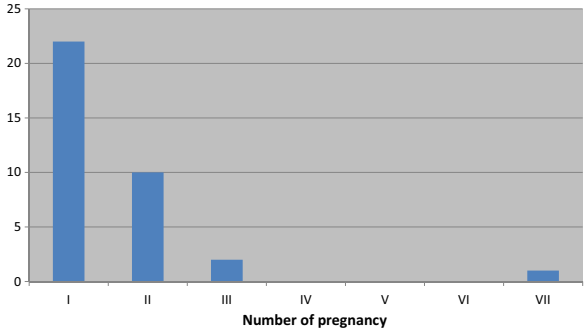

Fig. 3 – Number of pregnancy in which defect is diagnosed.

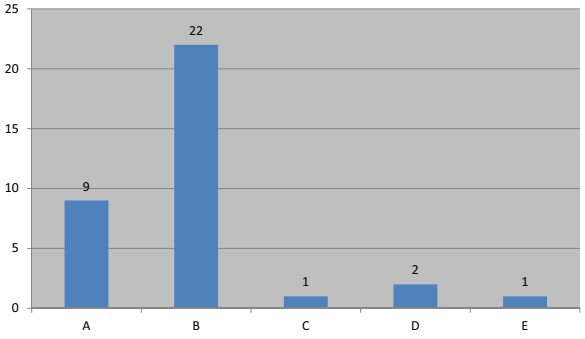

Fig. 5 – Herniated contents in ultrasound image. A – only small bowel; B – only small or large bowel; C – small and large bowel, liver; D – small and large bowel, stomach; E – small and large bowel, liver, stomach.

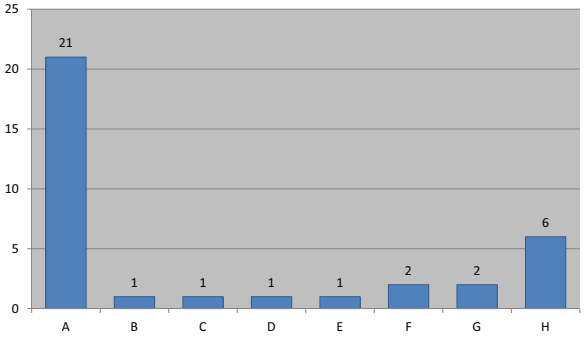

Fig. 6 – Complications during pregnancy. A – no complications; B – polyhydramnios, mesenteric cyst; C – obstruction, amputation of extraabdominal intestinal loops and necrosis; D – obstruction, oligohydramnios, impeded urine flow/substantially filled bladder; E – increased amount of amniotic fluid; F – polyhydramnios; G – oligohydramnios; H – only obstruction.

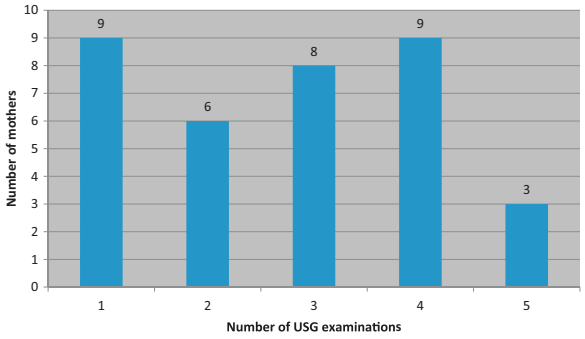

Fig. 7 – Number of ultrasound examinations during pregnancy.

posterior horns of lateral ventricles; in 1 (3%) fetus multiple congenital anomalies were identified: lateral displacement of the heart, abnormal spine curvature, ductus venosus flow was not visualized, and assessment of limbs was indecisive.

In 21 (57%) cases there were no complications of gastroschisis observed during pregnancy. In rest of the cases obstruction, polyhydramnios, decreased amount of amniotic fluid or hypotrophy were noted (Fig. 6). The mode of ultrasonographic monitoring of gastroschisis was dependent on gestational age at the time of diagnosis; in 25 (71%) pregnant women at least one follow-up examination was performed (Fig. 7).

Literature data indicate that in recent years increasing number of gastroschisis has been observed.[17–20] Although risk factors has not yet been precisely identified, similar to other analyses, in the material of MFMC gastroschisis was more frequently observed in infants of young mothers, which may be promoted by the use of stimulants and medication in the early pregnancy. The majority of gastroschisis diagnoses was made between 20 and 23 weeks gestation, which on one hand results from better imaging at this stage of pregnancy, and on the other, from the profile of the Centre, to which patients with suspected fetal malformation are referred.

5. Conclusions

Favorable prognosis is associated with lack of concomitant malformations, which was also confirmed by the very analysis. Early diagnosis seems to be especially important as it allows monitoring for potential complications of gastroschisis, which may affect prompt decisions of elective preterm delivery, delivery at a referral center, and hence the possibility of preparing neonatal and pediatric surgical team.

Conflict of interest

None declared.

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