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Ultrasonographic detection of gastroschisis in the first trimester: Actualities in pregnancy management and outcome

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ABSTRACT

Gastroschisis is a congenital anterior abdominal wall defect that occurs in 4 to 5 in 10,000 births, consisting in the evisceration of bowel in the amniotic cavity without a membranous covering. Gastroschisis cases can be detected during the first trimester using ultrasonography or at the beginning of the second trimester with a detection rate of 100%. Although an isolated condition in most of the cases, further complications such as malrotation, atresia, stenosis, perforation or volvulus can be associated. Periodic monitoring is required to assess the progress of the pregnancy with measurements of the level of amniotic fluid, fetal biometry and bowel diameters at every ultrasound examination. Gastroschisis is most frequently associated with oligohydramnios, intrauterine fetal growth restriction, increased bowel wall thickness and bowel dilatation. The outcome of the disease is favourable with a survival rate of around 90% for born infants. The long time on ventilator and multiple intestinal surgeries that delay the moment of oral feeding with a long-term use of total parental nutrition and short bowel syndrome are the most important comorbidities. Delivery is recommended at 38 weeks of gestation, preserving caesarean delivery for complicated gastroschisis. Although a complex pathology, gastroschisis is associated with normal intellectual development and few to none long-term complications.

Keywords: gastroschisis, first trimester ultrasonography, pregnancy management

INTRODUCTION

Gastroschisis represents a congenital abdominal wall defect associated with the evisceration of the bowel in the amniotic cavity without a membranous covering. The reported incidence worldwide is 4 to 5 in 10,000 live births and in most cases, it occurs on the right of the umbilical ring (1), being similar in both male and female fetuses, with fetuses from young mothers (under 20 years old), with low body mass index, who are smokers, high alcohol consuming or recreational drug users during pregnancy, having a higher risk of developing gastroschisis (2-5).

The vast majority of gastroschisis cases can be detected during the first trimester using ultrasonography examination or at the beginning of the second trimester. As a result of first trimester ultrasound screenings the detection rates of gatroschisis are over 90% with most of the cases being detected between 11 to 14 weeks (6). A complementary method of investigating gastroschisis in cases with risk factors for this pathology is represented by maternal serum alpha-fetoprotein levels, which are considerably higher (7).

The most important ultrasonographic differential diagnosis must be performed with another con-

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genital wall defect, omphalocele, in which the herniated organs are surrounded by a membranous sac (8-10). Other pathologies that must be considered in the differential diagnosis of gastroschisis are represented by cloacal exstrophy, urachal cysts, limb-body wall complex and ectopia cordis (11).

ULTRASONOGRAPHIC IDENTIFICATION OF GASTROSCHISIS

Gastroschisis is usually identifiable from 11 weeks of gestation using transabdominal ultrasonography. A thorough examination is required if the maternal alpha-fetoprotein serum levels are increased as a high level of the protein is associated with gastroschisis (7,12).

During ultrasonographic examination gastroschisis appears as an anterior abdominal wall defect located near the umbilical ring with, in almost all cases near the right side of it, from which bowel protrudes in the amniotic cavity without having a membranous covering. The umbilical ring is not part of the defect and is normal in most cases (13,14). Most frequently only the bowel can be seen protruding and floating in the amniotic liquid, although in rarer cases organs such as the liver or the stomach can also herniate. When they do not herniate, they suffer a process of malposition as a result of the exercised traction by the intestine (14) (Figure 1).

The floating intestine can be visualized like a cauliflower at ultrasonography as a result of the dilatation, inflammation and bowel wall oedema caused by the prolonged exposure to the amniotic fluid. Further complications such as malrotation, atresia, stenosis, perforation or volvulus can occur from the chronic exposure to the amniotic fluid and

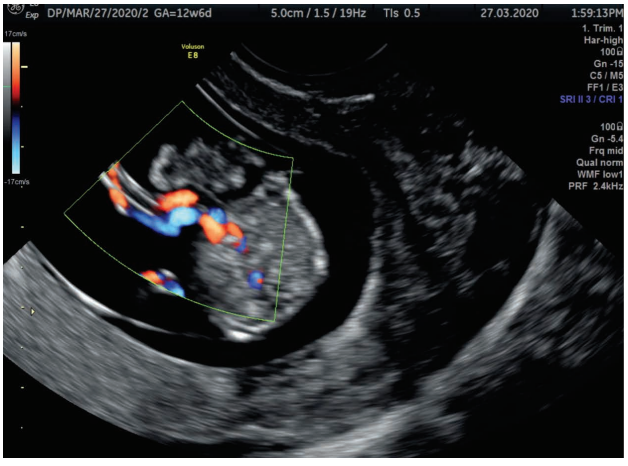

FIGURE 1. 2D color Doppler Transversal abdominal section: on the right side of the umbilical cord insertion and cranial of it the gut is visible floating in amniotic fluid, uncovered by the peritoneum and non-included in the umbilical cord insertion as a differential diagnosis with omphalocele

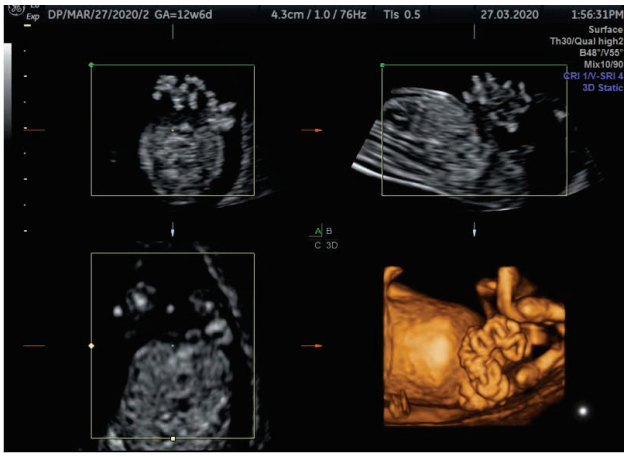

FIGURE 2. 3D ultrasonographic reconstruction image of the eviscerated gut through the anterior abdominal wall

must be identified in order to determine the management of the pregnancy. Gastroschisis is commonly an isolated defect, it can rarely be associated with other congenital abnormalities of other organs or as part of a genetic syndrome (15) (Figure 2).

MANAGEMENT AND OUTCOME OF PREGNANCIES WITH GASTROSCHISIS

Due to the early diagnosis of gastroschisis, as early as the 11th week of pregnancy and around 90% of the cases are diagnosed in the first trimester. Once the abdominal wall defect is discovered, further examination is at utmost importance to discover associated abnormalities. Stephenson et al. reported that from a registry of 3,300 cases of gastroschisis, 85% are isolated (gastroschisis alone or associated with one defect such as any intestinal defect, clubfoot or hip dysplasia, patent ductus arteriosus, patent foramen ovale) while 15% are non-isolated (associated with chromosomal syndromes such as trisomy 18, 13 or 21, or other aneuploidy) (16).

Fetal genetic studies are not required if the gastroschisis is isolated as the prevalence of chromosomal anomalies in these cases is similar to the general population (17). Genetic testing, including amniocentesis, is recommended if other extraintestinal abnormalities are identified using ultrasonography (18) (Figure 3).

Periodic monitoring is required to assess the course of the pregnancy. Overton et al. suggest ultrasound follow-up evaluation every month between 20 and 28 weeks of gestation, every two weeks between 28 and 34 weeks of gestation and weekly after 34th week of gestation until delivery. During all examinations, the practitioner has to evaluate the amniotic fluid using the amniotic fluid index, the normal fetal growth using fetal biometry and also bowel diameters (19).

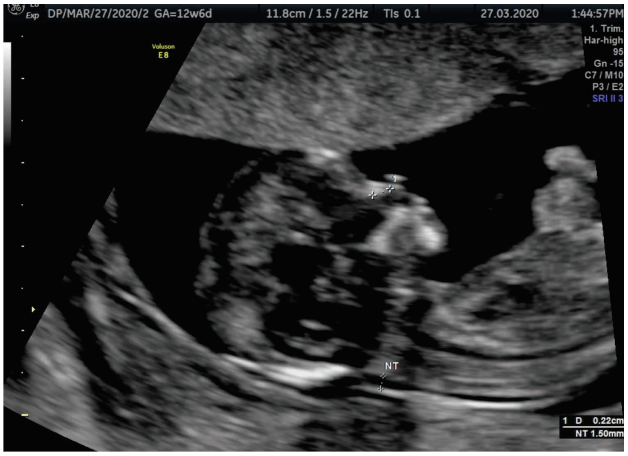

FIGURE 3. Standard sagittal section for NT (nuchal translucency) and NB (nasal bone) measurement (personal collection of Paul Costin Pariza)

Fetal biometry should to be performed during every ultrasound examination in order to detect the onset of fetal growth restriction, which can be significant in abdominal wall defect abnormalities. Growth restriction in fetuses with gastroschisis is predictive for an increased risk of neonatal complications (20).

Amniotic fluid volume level is a vital parameter during the periodic examinations, as it is one of the parameters that modifies. Oligohydramnios is related to fetal growth restriction while polyhydramnios, although rarer, is caused by bowel atresia and it is a predictive for severe complications after birth (21,22).

Bowel wall thickness and bowel dilatation are a result of prolonged exposure to amniotic fluid. If a thickening of the bowel wall of more than 25 mm before 34 weeks is observed during examination, Stephenson et al. (16) recommended the administration of antenatal corticosteroids to accelerate the fetal pulmonary maturation. Page et al. (1) reported an almost doubled spent time in the neonatal intensive care unit for fetuses with bowel dilatation, while a dilatation of over 6 mm is a predictive factor for complex gastroschisis, with an increased risk of bowel obstruction in postnatal life of 4 times higher than baseline population.

The prognosis of this congenital abdominal wall defect is favourable with a survival rate of around 90% for born infants; Fullerton et al. (23) reported a survival rate of 97.8% in 4,420 neonates born with gastroschisis in North America. Although with important healthcare-related comorbidities such as the time on ventilator, multiple intestinal surgeries, delay in time of oral feeding, these infants are at risk of a long-term use of total parental nutrition and short bowel syndrome.

Regarding delivery, it is recommended to be performed in a facility with appropriate structures that can offer assistance in case of emergency. Stephenson et al. (16) recommend delivery at 38+0 weeks of gestation, as well as reserving caesarean delivery for gastroschisis with associated liver herniation because of the increased risk of complications.

Long-term outcome was reported to be favourable, with children between 5 to 17 years old reporting intellectual abilities in the normal range. Long-term issues reported by surviving patients are intestinal adhesions, short bowel syndrome, as well as, psychological issues from the lack of umbilical ring (24,25).

CONCLUSIONS

Gastroschisis, although a congenital malformation with a favourable prognosis regarding long-term intellectual abilities and manageable intestinal comorbidities, represents a challenge for the sonographist and obstetrician to decide the moment of birth in the context of the balance between prematurity complications and intestinal damage. Frequently iatrogenic prematurity is a better option for saving the integrity of the digestive system which is responsible for the entire outcome of the gastroschisis malformation.

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REFERENCES

1. Page R, Ferraro ZM, Moretti F, Fung KF. Gastroschisis: antenatal sonographic predictors of adverse neonatal outcome. *J Pregnancy.* 2014;2014:239406.
2. Mac Bird T, Robbins JM, Druschel C, Cleves MA, Yang S, Hobbs CA; National Birth Defects Prevention Study. Demographic and environmental risk factors for gastroschisis and omphalocele in the National Birth Defects Prevention Study. *J Pediatr Surg*. 2009 Aug;44(8):1546-51.
3. Reid KP, Dickinson JE, Doherty DA. The epidemiologic incidence of congenital gastroschisis in Western Australia. *Am J Obstet Gynecol.* 2003 Sep;189(3):764-8.
4. Waller DK, Shaw GM, Rasmussen SA, Hobbs CA, Canfield MA, et al; National Birth Defects Prevention Study. Prepregnancy obesity as a risk factor for structural birth defects. *Arch Pediatr Adolesc Med.* 2007 Aug;161(8):745-50.
5. Draper ES, Rankin J, Tonks AM, Abrams KR, Field DJ, Clarke M, Kurinczuk JJ. Recreational drug use: a major risk factor for gastroschisis? *Am J Epidemiol.* 2008 Feb 15;167(4):485-91.
6. Syngelaki A, Hammami A, Bower S, Zidere V, Akolekar R, Nicolaides KH. Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11-13 weeks' gestation. *Ultrasound Obstet Gynecol.* 2019 Oct;54(4):468-476.
7. Chen Y, Zhang W, Lu S, Mei J, Wang H, Wang S, Gu L, Zhang L, Chu X. Maternal serum alpha fetoprotein and free β-hCG of second trimester for screening of fetal gastroschisis and omphalocele. *Zhejiang Da Xue Xue Bao Yi Xue Ban.* 2017 May 25;46(3):268-273.

8. Curtis JA, Watson L. Sonographic diagnosis of omphalocele in the first trimester of fetal gestation. J Ultrasound Med. 1988 Feb;7(2):97-100.
9. Brown DL, Emerson DS, Shulman LP, Carson SA. Sonographic diagnosis of omphalocele during 10th week of gestation. *AJR Am J Roentgenol.* 1989 Oct;153(4):825-6.
10. Gray DL, Martin CM, Crane JP. Differential diagnosis of first trimester ventral wall defect. *J Ultrasound Med.* 1989 May;8(5):255-8.
11. Benjamin B, Wilson GN. Anomalies associated with gastroschisis and omphalocele: analysis of 2825 cases from the Texas Birth Defects Registry. *J Pediatr Surg.* 2014 Apr;49(4):514-9.
12. Hu JL, Zhang YJ, Zhang JM, Zhu S, Li DM, Yin YF, Su J, Chan Y, He J, Cao YJ, Zhu BS. Pregnancy outcomes of women with elevated second-trimester maternal serum alpha-fetoprotein. *Taiwan J Obstet Gynecol.* 2020 Jan;59(1):73-78.
13. Prefumo F, Izzi C. Fetal abdominal wall defects. *Best Pract Res Clin Obstet Gynaecol.* 2014 Apr;28(3):391-402.
14. Brugger PC, Prayer D. Development of gastroschisis as seen by magnetic resonance imaging. *Ultrasound Obstet Gynecol*. 2011 Apr;37(4):463-70.
15. Hijkoop A, Lap CCMM, Aliasi M, Mulder EJH, Kramer WLM, et al. Using three-dimensional ultrasound in predicting complex gastroschisis: A longitudinal, prospective, multicenter cohort study. *Prenat Diagn.* 2019 Dec;39(13):1204-1212.
16. Stephenson CD, Lockwood CJ, MacKenzie AP. Gastroschisis. Available from: https://www.uptodate.com/contents/gastroschisis?search=gastroschisis&source=search_result&selectedTitle=1~41&usage_type=default&display_rank=1.
17. Fleurke-Rozema H, van de Kamp K, Bakker M, Pajkrt E, Bilardo C, Snijders R. Prevalence, timing of diagnosis and pregnancy outcome of abdominal wall defects after the introduction of a national prenatal screening program. *Prenat Diagn.* 2017 Apr;37(4):383-388.
18. Committee on Genetics and the Society for Maternal-Fetal Medicine. Committee Opinion No.682: Microarrays and Next-Generation Sequencing Technology: The Use of Advanced Genetic Diagnostic Tools in Obstetrics and Gynecology. *Obstet Gynecol.* 2016 Dec; 128(6):e262-e268.
19. Overton TG, Pierce MR, Gao H, Kurinczuk JJ, Spark P, Draper ES, Marven S, Brocklehurst P, Knight M. Antenatal management and outcomes of gastroschisis in the U.K. *Prenat Diagn.* 2012 Dec;32(13):1256-62.
20. Amin R, Domack A, Bartoletti J, Peterson E, Rink B, et al. National Practice Patterns for Prenatal Monitoring in Gastroschisis: Gastroschisis Outcomes of Delivery (GOOD) Provider Survey. *Fetal Diagn Ther.* 2019;45(2):125-130.
21. Japaraj RP, Hockey R, Chan FY. Gastroschisis: can prenatal sonography predict neonatal outcome? *Ultrasound Obstet Gynecol.* 2003 Apr;21(4):329-33.
22. Mastroiacovo P, Lisi A, Castilla EE, Martínez-Frías ML, Bermejo E, et al. Gastroschisis and associated defects: an international study. *Am J Med Genet A.* 2007 Apr 1;143A(7):660-71.
23. Fullerton BS, Velazco CS, Sparks EA, Morrow KA, et al. Contemporary Outcomes of Infants with Gastroschisis in North America: A Multicenter Cohort Study. *J Pediatr.* 2017 Sep;188:192-197.e6.
24. Koivusalo A, Lindahl H, Rintala RJ. Morbidity and quality of life in adult patients with a congenital abdominal wall defect: a questionnaire survey. *J Pediatr Surg*. 2002 Nov;37(11):1594-601.
25. van Eijck FC, Wijnen RM, van Goor H. The incidence and morbidity of adhesions after treatment of neonates with gastroschisis and omphalocele: a 30-year review. *J Pediatr Surg*. 2008 Mar;43(3):479-83.