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Review article

Vanishing gastroschisis visualized by antenatal ultrasound: a case report and review of literature

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ABSTRACT

We report a case of vanishing gastroschisis visualized by antenatal ultrasound with a 7-year long term follow-up. Currently, the child is still dependent on daily parenteral nutrition with no signs of hepatotoxicity. To our knowledge, it's the fourth case with a long-term follow-up. Vanishing gastroschisis is a rare complication of gastroschisis. However, physicians should be aware of it because its prognosis is worse than classical gastroschisis. When a vanishing gastroschisis is visualized or suspected by antenatal ultrasound, prenatal counseling is required with explanations about the risk of short bowel syndrome, the need of parenteral nutrition and related complications (inflammatory colitis, sepsis, liver failure and organ transplant). Mortality rate was initially around 93% and dropped to 27% after the years 2000 (versus 10% for classical gastroschisis). After birth, all children will require surgery, and sometimes autologous gastro-intestinal reconstruction. Most survivors (68%) could be taken off the TPN. Unfortunately, long-term outcomes for children with vanishing gastroschisis are still missing in current literature.

Contents

Introduction

Classical gastroschisis (CGS) is an abdominal wall defect (AWD) located on the right side of the umbilicus insertion and associated with small intestine prolapse into the amniotic fluid without any protective covering membrane [1]. Its incidence has increased from 0.1/10 000 birth in 1970 to 5/10 000 in the years 2000. Survival rate after surgery reaches more than 90% [2]. Rare complication of CGS is vanishing gastroschisis (VGS), where spontaneous closure of AWD occurs in utero [3]. VGS's incidence is estimated between 4.5 to 6% of all gastroschisis in series from Vogler et al. (219 cases) [4] and Houben et al. (146 cases) [5], respectively. Closure can be complete with an intact abdominal wall or incomplete with the presence of a useless extra-abdominal bowel remnant, leading to intestinal atresia. Several hypotheses have been postulated regarding the mechanism of vanishing gastroschisis; the common point is that intestinal atresia may result from a vascular accident in utero [6]. Outcomes from vanishing gastroschisis are worse than CGS due to short bowel syndrome (SBS) and to complications related to total parenteral nutrition (TPN) like liver failure or sepsis [7].

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We report a case of VGS visualized by antenatal ultrasound (US) that we have encountered in the obstetrics department of Saint-Luc University Hospital in Brussels (Belgium) and for which we have a 7-year follow-up. To our knowledge, it's only the fourth case with a long-term follow-up. We also propose a review of the literature about this topic. The purpose of this article is to present the antenatal features and postnatal outcomes of VGS based on literature. Because VGS prognosis is different from CGS, it seems important for practitioners to be aware of this complication to adapt prenatal counseling and prepare the parents for multidisciplinary postnatal care.

Case report

A 25-year old primigravida patient was seen at 13 weeks (Figs. 1 and 2) for nuchal translucency evaluation (Down syndrome screening) and a right-sided gastroschisis was detected on that occasion. Amniocentesis was performed for maternal anxiety at 16 weeks and revealed an alpha-fetoprotein level of 3.2MoM (multiples of the median) and a normal male conventional karyotype. At 20 weeks, ultrasound did not show any signs of gastroschisis anymore. The abdominal wall was closed and looked intact but intra-abdominal intestinal dilatation was seen and measured at 7.4 mm. Slightly hyperechogenic bowel was also noted. Evaluation for cystic fibrosis on the amniotic fluid previously collected was performed and screening for the 29 most common mutations was negative.

Repeated intra-abdominal bowel ultrasound inspections carried out at 23, 25, 27 and 30 weeks showed an increasing dilatation, from 8 millimeters to 18.7 millimeters (Fig. 3), with maximal intra-abdominal bowel dilatation (IABD) measured at 23 millimeters at 31 weeks. Maximal bowel wall thickness was measured at 1.2 mm. A fetal MRI was performed at 31 weeks and showed a proximal jejunum dilatation with absence of hyper signal meconium in the colon (Fig. 4). Due to increasing IABD, the patient was admitted at 31weeks for fetal lung maturation and closer surveillance. Spontaneous labor occurred at 32 + 5/7 weeks, despite treatment with intravenous tocolytic drugs (beta-agonists and atosiban). Caesarian section delivery was decided due to breech presentation, primigravida status and prematurity.

Birth weight was 2150 grams (P70) and Apgar score 9/10/10. At birth, the abdominal wall was closed and the abdomen slightly distended. Abdominal X-ray and ultrasound carried out at day zero showed a significant distension of the proximal small intestine and neither colon nor rectal ventilation could be observed. Parenteral nutrition was then initiated. On day one, exploratory laparotomy revealed a dilated jejunum with direct continuity to the colon and absence of ileum, ileocaecal valve and appendix. Two centimeters

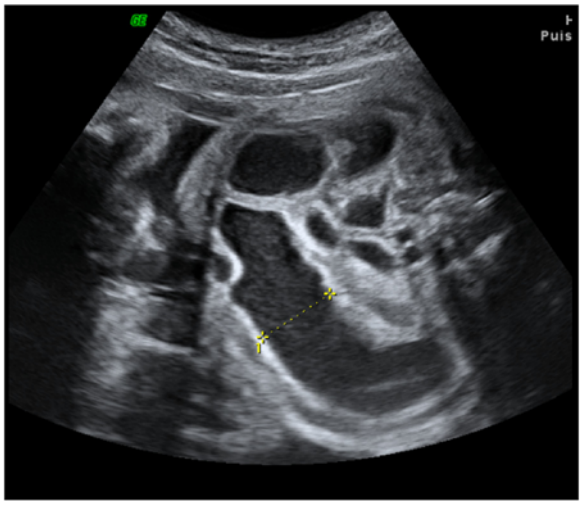
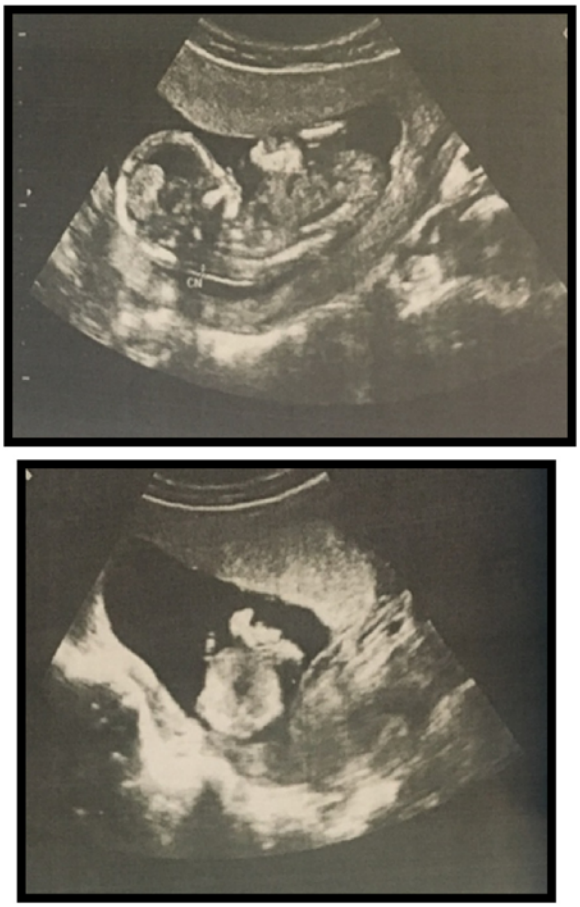


Fig. 3. Antenatal US at 30 weeks showing an intra-abdominal bowel dilatation of 18.7 mm.



Figs. 1 and 2. Ultrasound at 13weeks, diagnosis of gastroschisis.

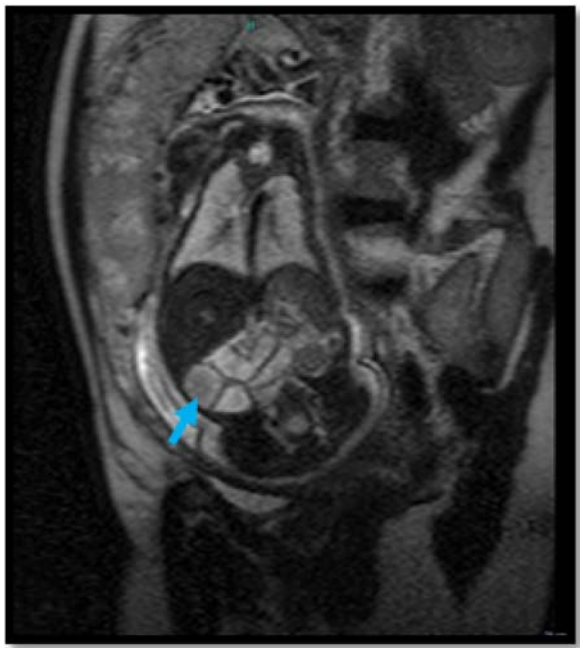


Fig. 4. MRI showing → dilatation of jejunum.

of an atretic colon were resected and a jejunocolic anastomosis created. Small intestine measured 27 centimeters after anastomosis and the colon was almost normal in terms of length and anatomy. These findings led to the diagnosis of short bowel syndrome (SBS). During hospital stay, numerous attempts of enteral feeding failed because they led to significant intestinal dilatation requiring enemas for stool removal. Finally, the child left the hospital at 4 months of age with daily parenteral nutrition and partially oral feeding and nightly enteral feeding.

At 7 months of age, a longitudinal intestinal lengthening and tapering (LILT) surgery (as described by Bianchi) and a gastrostomy were performed. Small intestine final length was 76 centimeters. His subsequent evolution has been marked by several consultations for viral gastroenteritis and respiratory tract infections leading to dehydration, which could be treated at home. Gastrostomy was removed at 3 years of age because it was not more used. He underwent two additional laparotomies. The first one took place at the age of 4 and a half and consisted of the resection of a fistula located between two intestinal loops, creation of an enteroenteric anastomosis and placement of a new gastrostomy. Second surgery took place two months later and was indicated for jejunostomy placement and creation of a colostomy on the left colon due to an inflammation and painful colitis. After three years, a surgery was planned to restore intestinal continuity.

Currently, the child is 7 year-old and is still dependent on daily TPN with no signs of hepatotoxicity. Unfortunately, attempts to wean off the TPN have been unsuccessful so far. Nevertheless, weight gain is correct and body mass index is at the percentile 25. He has good school performances even though he is suffering from moderately hyperactivity.

Discussion

Vanishing gastroschisis is a rare complication of CGS. As we have presented, parents should not be reassured if spontaneous closure of AWD is visualized on antenatal ultrasound because VGS carries out a different prognosis.

According to the literature, the first VGS case was reported by Kiesewetter in 1957 [8]. Keywords used to research in PubMed database were “closing gastroschisis”, “vanishing gastroschisis”, “closed gastroschisis” and “vanishing midgut/bowel”.

We found 34 articles in English [3–36] about 62 cases related to this subject. In 48 cases, closure of the AWD was identified at birth but in only 14 cases this closure was visualized by antenatal US. These 14 cases are summarized in Table 1. The median age at gastroschisis diagnosis was 17 weeks (12-22). Usually, gastroschisis is detected on prenatal ultrasound during the first trimester, sometimes as early as 11 weeks [2]. Closure of the AWD was visualized at the average age of 29 weeks (16-35) (Table 1). For the majority of vanishing gastroschisis, closure of the AWD was a late phenomenon, visualized during the third trimester. In the case reported by Buluggiu et al. [9] as in our case the closure happened during the second trimester. For eight cases among all the published cases, there was even no gastroschisis diagnosed by antenatal US [4,15,16,28,33,34,36]. VGS was only diagnosed at birth.

In gastroschisis, IABD develops more frequently during the second trimester [37]. It has been recognized as a promising sign to identify intestinal atresia and diagnose VGS [5]. Dilatation of intra-abdominal bowel loops was found in about half of the cases reported (30/62) despite lack of information regarding antenatal features. Normal fetal bowel dilatation is rarely found to be greater than 6 mm, independently of gestational age [5]. According to Houben et al.[5] an IABD, in cases of CGS, exceeding 10 mm needs closer prenatal surveillance to give the opportunity for earlier intervention and salvaging of as much bowel length as we can, which would otherwise be lost if the pregnancy were continued without intervention. But preterm delivery is still at debate because there’s no correlation between antenatal scan and postnatal viable bowel length [16]. Additionally, induced prematurity could lead to significant consequences.

Intra-uterine death is another complication of gastroschisis occurring more commonly during third trimester with an overall rate of 10% [2,38]. In VGS, we found three cases of stillbirth [10,12,35] corresponding to an incidence of 5%(3/62).

At birth, 25 described cases had complete closure of the AWD, as in our case. Thirteen had a small right para-umbilical remnant, and in 2 cases the remnant was on the left side (reported by Abdel-Latif et al. [34] and Sing et al. [36]). All the others cases presented an incomplete closure with a small AWD, sometimes as small as 4 mm [15]. All cases suffered from intestinal atresia leading to SBS, no matter if the abdominal wall closure was complete or not.

After birth, mortality rate in VGS based on all the published cases since 1957 is estimated at 42%(25/59) versus 10%[38] in CGS. At the time of the respective publications, 34 children were alive. But when we compared mortality rates before and after the years 2000, there was a significant difference probably due to the progress of antenatal diagnosis, neonatal care and surgery. Before 2000, 13 children died and only 1 survived, corresponding to a mortality rate of 93% After the years 2000, mortality rate dropped

Table 1
14 cases with antenatal diagnosis of vanishing gastroschisis.

First author	Year of publication	GS seen for the first time (weeks)	GS no more visualised (weeks)
Pinette [10]	1994	20	33
Bromley [11]	1995	16	30
Barsoom [7]	2000	19	34
Tawil [12]	2001	15	30
Basaran [6]	2002	20	35
Winter [13]	2005	21	30
Buluggiu [9]	2009	12	16
Khalil [3]	2010	18	33
Dahl [14]	2011	19	>33
Kumar [15]	2012	uns	uns
Wood [16]	2013	18	28
		uns	29
Dennison [17]	2016	13	26
Sergi [18]	2018	22	34
Marin		13	20

GS=gastroschisis
UNS=unspecified

to 27%, with 12 deaths among 45 cases. This amelioration concerning the mortality rate may change the prognosis of the future newborn.

Outcomes from vanishing gastroschisis are worse than CGS due to SBS complications such as inflammatory colitis, motility disorders and surgeries. In some cases, problems related to TPN like liver failure or sepsis, have been described [7]. The 34 VGS survivors are summarized in Table 2.

Neonates with VGS present a significant bowel loss from the duodenum or jejunum extending to the distal colon [16], sometimes requiring surgery in the form of autologous gastrointestinal reconstruction (AGIR) or therapeutic agents such as glucagon-like peptide-2 (GLP2) who promote mucosal growth. Two types of surgery are frequently used: longitudinal intestinal lengthening and tapering (introduced in 1980 by Bianchi) and serial transverse enteroplasty procedure (STEP) (introduced in 2003) [39]. In 18 cases among the 34 that were alive, 3 had an unspecified surgery, 9 had a LILT and the 6 remaining ones had a STEP. Among those 6 cases, one is still fully dependent on TPN, two are weaning off TPN, one received a bowel transplant and one was transferred to a transplant center. So, only one child was able to be independent of parenteral nutrition. TPN withdrawal rate after STEP was about 16%(1/6). Outcomes of nine children after Bianchi surgery were different and seemed to be better. Three children are on enteral feeding including two who received an organ transplant (one liver and one bowel transplant). Four others are on oral feeding. Another one was admitted to the hospital at 8 months of age due to TPN hepatotoxicity. And the last one couldn't be weaned off the TPN. TPN withdrawal rate without organ transplant can be evaluated at 71%(5/7). In our case, LILT surgery was performed at 7 months, but the child remained on daily TPN at 7 years of age, maybe related to intestinal motility disorders. In a review written by King et al. [40] regarding 403 procedures of intestinal bowel lengthening in children with short bowel syndrome, TPN withdrawal rate was nearly similar, about 55% after LILT versus 48% after STEP. They also mentioned a higher rate of organ transplant after Bianchi: 10% versus 6% after STEP. However after reviewing VGS literature, we found that TPN withdrawal rate is better after LILT surgery (71%) than after STEP surgery (16%). Regarding organ transplant, the rate seems to be similar after STEP or after Bianchi's procedure in VGS population.

Organ transplant is another complication directly related to SBS and liver failure (secondary to TPN). In total, 6 children had an organ transplant. Two received a bowel transplant; one had a liver transplant, and the remaining one a combined liver and bowel transplant. Two others were transferred to a transplant center, but there was no more information regarding the type of surgery or the outcome. The prognosis of intestinal transplantation is still hazardous with a survival rate at 5 years of 58% for patients, and 60% for the graft survival in the United States [41].

Table 2
Outcomes of 34 vanishing gastroschisis survivors

First author	Case	Year of publication	AGIR	Organ transplant surgery	Age at the time of publication	Feeding
Kimble [27]	1	1999	LILT at 14w		12y	oral
Barsoom [7]	2	2000	LILT at 5m		8m, alive but hospitalized with TPN hepatotoxicity	TPN
Ogunyemi [30]	3	2001		liver and bowel	53m	oral
Winter [13]	4	2005	LILT	bowel	32m	enteral
Sandy [31]	5	2006	STEP at 30m		40m	weaning TPN
Vogler [4]	6	2008			uns	oral
	7				uns	oral
	8				uns	oral
	9		STEP		uns	enteral
	10				uns	oral
	11				uns	oral
	12		STEP at 6w		uns	weaning TPN
Buluggiu [9]	13	2009	LILT at 5m		25m	oral
Houben [5]	14	2009	LILT		4y	oral
	15		LILT at 12w	liver	11y	enteral
	16				1y	enteral
	17				median follow up 6 (0,5-11) years	enteral
	18		LILT at 6 m			TPN
	19					enteral
	20					enteral
	21					enteral
Khalil [3]	22	2010	LILT at 6 m		2y	enteral
Lawther [32]	23	2010	STEP at 9m		9m	TPN
Dahl [14]	24	2011			21m	oral
Kumar [15]	25	2012	STEP	bowel	2y	uns
	26		STEP	transfer to a transplant center	uns	uns
	27				15d	uns
Wood [16]	28	2013	AGIR		3y	oral
	29		AGIR		uns	weaning TPN
	30		AGIR		uns	uns
	31		AGIR		8m	oral
Abdel-Latif [34]	32	2017	LILT		1m	oral
Sergi [18]	33	2018			10y	oral
Singh [36]	34	2018			48m	oral
Marin			LILT at 7m		7y	TPN

AGIR=autologous gastrointestinal reconstruction
STEP=serial transverse enteroplasty procedure
LILT=longitudinal intestinal lengthening and tapering
d=days,
m=months,
y=years
UNS=unspecified

In VGS, TPN is necessary to feed patients and weaning represents an important goal in SBS treatment. Since 1957, 23 children among the 34 survivors (68%) were weaned off the TPN and were doing well; the older was 12 year-old. Three children are weaning off TPN, and two other are still fully dependent. For six children the diet was unspecified.

Long-term outcomes about gastroschisis survivors are rare [38], as well as for vanishing gastroschisis cases. In VGS, there are only three long term survivors reported by Kimble, Houben, and Sergi [5,18,27]. They are respectively 12, 11 and 10 years old. One is on oral, one on enteral feeding, and for the last one the diet is unknown. Our case is the fourth one reported with a long-term follow-up, but has not yet been weaned off TPN.

In conclusion, the postnatal prognosis of VGS is really different from CGS. The diagnosis of important intra-abdominal bowel loops dilatation in CGS has to bring to our mind the eventuality of VGS and deserved closer surveillance. When VGS is diagnosed or suspected prenatally, parents need to be counseled with respect to the probability of prolonged initial hospitalization, multiple surgeries, ongoing feeding problems due to the high incidence of SBS, and the potential of long-term morbidity.

Declaration of Conflicting Interests

The author(s) declared no conflict of interest with respect to the research, authorship, and/or publication of this article.

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