Journal of Pediatric Surgery 58 (2023) 1090–1094

Contents lists available at ScienceDirect

Journal of Pediatric Surgery

journal homepage: www.sciencedirect.com/journal/journal-of-pediatric-surgery

Prenatal Sonography in Suspected Proximal Gastrointestinal Obstructions: Diagnostic Accuracy and Neonatal Outcomes

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ARTICLE INFO

Article history:
Received 26 January 2023
Accepted 10 February 2023

Keywords:
Proximal gastrointestinal obstruction
Double bubble
Polyhydramnios

ABSTRACT

Background: The purpose of this study was to assess diagnostic accuracy and neonatal outcomes in fetuses with a suspected proximal gastrointestinal obstruction (GIO).
Methods: After IRB approval, a retrospective chart review was conducted on prenatally suspected and/or postnatally confirmed cases of proximal GIO at a tertiary care facility (2012–2022). Maternal-fetal records were queried for presence of a double bubble ± polyhydramnios, and neonatal outcomes were assessed to calculate the diagnostic accuracy of fetal sonography.
Results: Among 56 confirmed cases, the median birthweight and gestational age at birth were 2550 g [interquartile range (IQR) 2028–3012] and 37 weeks (IQR 34–38), respectively. There was one (2%) false-positive and three (6%) false-negatives by ultrasound. Double bubble had a sensitivity, specificity, positive predictive value, and negative predictive value for proximal GIO of 85%, 98%, 98%, and 83%, respectively. Pathologies included 49 (88%) with duodenal obstruction/annular pancreas, three (5%) with malrotation, and three (5%) with jejunal atresia. The median postoperative length of stay was 27 days (IQR 19–42). Cardiac anomalies were associated with significantly higher complications (45% vs 17%, $p = 0.030$).
Conclusions: In this contemporary series, fetal sonography has high diagnostic accuracy for detecting proximal gastrointestinal obstruction. These data are informative for pediatric surgeons in prenatal counseling and preoperative discussions with families.
Level of Evidence: Diagnostic Study, Level III.

1. Introduction

Proximal congenital gastrointestinal obstructions (GIO), which include duodenal atresia and jejunal atresia, are congenital surgical anomalies that have an incidence of 1 in 2000 live births and require surgery in the early neonatal period [1,2]. Within the proximal gastrointestinal tract, there are a wide range of pathologies that can be intrinsic (e.g., atresias, webs, stenosis) or extrinsic (e.g., volvulus, annular pancreas, vascular defects) in nature. In patients with duodenal atresia, 30–45% are associated with trisomy 21 [3–5] whereas cardiac defects and additional anomalies are seen in 50–65% of cases [6–8].

Most cases of duodenal atresia are now detected antenatally by ultrasound. Well documented hallmark findings on fetal ultrasound are a dilated stomach with adjacent duodenal dilation (double bubble). Polyhydramnios often develops after 24 weeks gestation. However, false-positive cases have been reported, and the overall diagnostic accuracy of the fetal double bubble for proximal GIO has not been well documented in the recent literature [6–13]. Such data would be of obvious value for pediatric surgeons, neonatologists, and maternal-fetal medicine specialists during counseling and delivery planning discussions with families.

The aim of this study was to determine the accuracy of prenatal sonography in suspected proximal GIO and to characterize early postnatal outcomes. We hypothesized that the widespread use and enhanced resolution of fetal ultrasound would enable the diagnosis of proximal GIO with high sensitivity and specificity.

List of abbreviations: GIO, gastrointestinal obstruction; ICD, International Classification of Diseases; IQR, interquartile range; LOS, length of stay; NPV, negative predictive value; PPV, Positive predictive value; TPN, total parenteral nutrition.

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https://doi.org/10.1016/j.jpedsurg.2023.02.029
0022-3468/

2. Materials and methods

2.1. Study cohort

This was a retrospective study of all neonates with a prenatally suspected and/or postnatally confirmed proximal GIO who were referred to Johns Hopkins Medicine in Baltimore, Maryland, between January 1, 2012 and April 30, 2022. Institutional Review Board approval (IRB 00296389) and waiver of consent were obtained.

A prospectively maintained fetal database was used to identify all suspected proximal GIO fetuses referred to Johns Hopkins Maternal-Fetal Medicine. Cases suspected by the presence of a double-bubble sign, with or without polyhydramnios, and those with postnatally affirmed proximal GIO in which prenatal sonograms had been performed were included. Ultrasound reports and images were reviewed by two maternal-fetal medicine colleagues (ACJ, KJB) to evaluate the presence of a dilated stomach with adjacent proximal bowel dilation (double bubble) and/or polyhydramnios (amniotic fluid index > 25 or deepest vertical pocket > 8). Prenatal cases were managed expectantly and counseled by a multidisciplinary team, including a maternal-fetal medicine specialist and a pediatric surgeon. Additionally, a supplemental chart review was conducted using a pediatric surgical database based on International Classification of Disease (ICD) codes for annular pancreas (751.7, Q45.1), duodenal (751.1, Q41.0), jejunal (Q41.1), and unspecified intestinal atresia (Q41.8 and Q41.9).

2.2. Eligibility criteria and outcomes

Fetuses with a suspected esophageal atresia or obstructions felt to more likely involve the mid or distal small bowel were excluded. Additional exclusion criteria were termination, fetal death, and incomplete data. For the diagnostic accuracy calculations of the sonographic findings, we also excluded those with no recorded prenatal care.

The primary study outcome was diagnostic accuracy of fetal ultrasound for proximal GIO. Secondary study outcomes included types of proximal GIO, hospital length of stay, and postoperative complications rates.

2.3. Statistical analysis

The diagnostic accuracy, including sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV), were calculated. Patient demographics and hospitalization characteristics were assessed using frequency and central tendency measures. p values were generated using Chi-squared, Fisher's exact, and Kruskal–Wallis tests (R Core Team, v3.6.1, Vienna Austria). For all analyses, two-sided $p < 0.05$ was considered significant.

3. Results

Demographic and patient characteristics data are shown in Table 1. Among the 56 infants with confirmed proximal GIO who met inclusion criteria, there were 29 (52%) males and 27 (48%) females. The median birthweight was 2550 g [interquartile range (IQR) 2008–3008], and the median gestational age at birth was 35.7 weeks (IQR 33.4–37.9). Sixteen (29%) had trisomy 21, and 20 (36%) had cardiac anomalies. Two patients had cardiac procedures during the initial admission.

Table 1
Characteristics of proximal gastrointestinal obstructions, n = 56.

Characteristic	
Birthweight (grams), median (IQR)	2500 (2008, 3008)
Gestational age, median (IQR)	35.7 (33.4, 37.9)
Sex	
Female, n (%)	27 (48.2)
Male, n (%)	29 (51.8)
Race	
Asian, n (%)	1 (1.8)
Black, n (%)	19 (33.9)
White non-Hispanic, n (%)	19 (33.9)
White Hispanic, n (%)	10 (17.9)
Other, n (%)	7 (12.5)
Year	
2012, n (%)	5 (8.9)
2013, n (%)	3 (5.4)
2014, n (%)	7 (12.5)
2015, n (%)	8 (14.3)
2016, n (%)	2 (3.6)
2017, n (%)	7 (12.5)
2018, n (%)	4 (7.1)
2019, n (%)	5 (8.9)
2020, n (%)	6 (10.7)
2021, n (%)	5 (8.9)
2022, n (%)	4 (7.1)
Additional distal GIO, n (%)	7 (12.5)
Anomalies	
Trisomy 21, n (%)	16 (28.6)
Cardiac, n (%)	20 (35.7)
Heterotaxy, n (%)	3 (5.4)
Renal, n (%)	2 (3.6)
Vertebral, n (%)	2 (3.6)
Outborn	16 (28.6%)

3.1. Diagnostic accuracy

Of the 54 cases with documented prenatal ultrasounds, the median gestational age at diagnosis was 26.4 weeks (IQR 21.2–29.7). Twenty percent had a normal finding at the initial anatomic survey scan at 20 weeks. Sonographic findings consistent with proximal GIO included both double bubble and polyhydramnios (n = 37, 69%) and double bubble without polyhydramnios (n = 9, 17%). Three (6%) with a confirmed proximal GIO had normal fetal ultrasound reports and were therefore false-negative cases (Fig. 1). Once suspected, none of the imaging findings disappeared or regressed later in pregnancy. Only one patient was a false-positive diagnosis, having been referred for a double bubble without polyhydramnios at 37 weeks gestation. This neonate tolerated enteral feeds and was discharged on day of life 2 without surgical intervention. There were three affected neonates who had normal prenatal ultrasounds (i.e., false negatives) and were found to have feeding intolerance at birth and duodenal webs at operation.

Diagnostic accuracy data are shown in Table 2. The presence of a double bubble had a sensitivity, specificity, PPV, and NPV of 85%, 98%, 98%, and 83%, respectively. The presence of both a double bubble and polyhydramnios had a sensitivity, specificity, PPV, and NPV of 69%, 100%, 100%, and 70%, respectively.

3.2. Type of proximal obstruction

Table 3 reveals the different types of proximal GIO identified in the study cohort. Forty-nine (71%) had intraoperative findings of a duodenal obstruction, which included 22 (39%) with type 2 or 3 duodenal atresias and 18 (32%) with type 1 disease secondary to a duodenal web. Nine (16%) were noted to have an annular pancreas. Other pathologies were three (5%) with malrotation and midgut volvulus, three (5%) with jejunal atresia, and one (2%) with pyloric

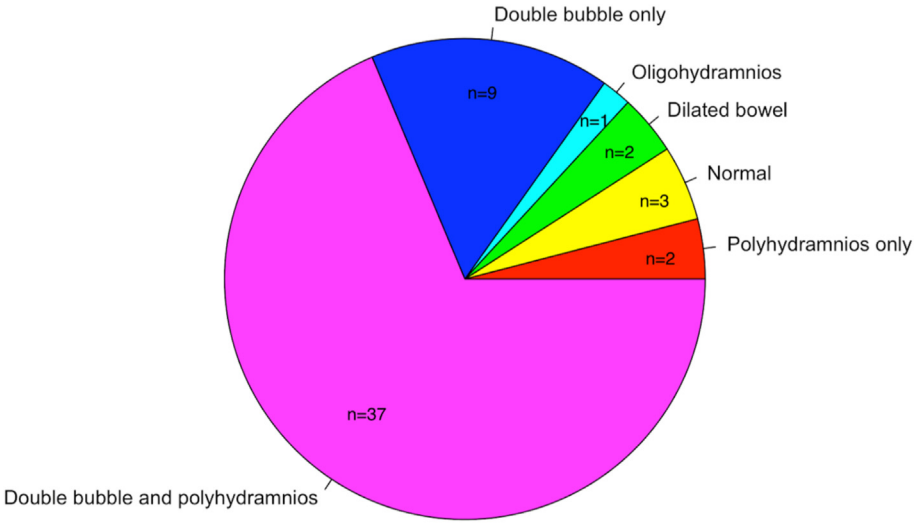

Fig. 1. Prenatal ultrasound findings.

atresia. There were three (5%) patients who had more than one intestinal atresia.

3.3. Hospital course

In all duodenal obstruction cases, a diamond-shaped duodenoduodenostomy was performed. Most infants underwent open surgery (77%), but 13 (23%) underwent laparoscopic repair. The conversion rate to open repair in this latter group was 31%. Four (7%) patients had a gastrostomy tube placed during the initial repair. Postoperative feeds were started at a median of 8 days (IQR 5–12).

The median postoperative length of stay was 27 days (IQR 19–42). Of the three (5%) neonates with midgut volvulus, there was a significantly longer LOS postoperatively (66 days, IQR 44–148 vs 22 days, IQR 0–201; p = 0.021) and longer time to full enteral feeds (40 days, IQR 38–52 vs 18 days, IQR 0–76; p = 0.012). There was a trend of longer time to initial feeds (20 days, IQR 9–31 vs 8, IQR 3–62; p = 0.056). One infant (33%) with midgut volvulus required total parenteral nutrition (TPN) at discharge whereas none of the non-volvulus patients were discharged on TPN (p = 0.037). There was no significant difference in complication rates between the two groups.

3.4. Postoperative complications

There were no in-hospital deaths. Seven (13%) patients underwent a second operation during the same hospitalization at a median age of 43 days (IQR 31.5–142.5): Four (7%) were re-explorations, and two (4%) had gastrostomy tubes placed. Postoperative complications based on the presence or absence of cardiac anomalies are compared in Table 4. The frequency of proximal GIO patients having any complication was 27%, but cardiac anomalies were significantly associated with increased complications (45% vs 17%, p = 0.030). Trisomy 21 was not associated with any significant differences in postoperative complications.

Table 3
Types of proximal gastrointestinal obstructions, n = 56.

	Overall (%)
Duodenal atresia (type 2 or 3)	22 (39)
Duodenal atresia (web)	18 (32)
Annular pancreas	9 (16)
Jejunal atresia	3 (5)
Malrotation with volvulus and jejunal atresia	2 (4)
Malrotation with volvulus	1 (2)
Pyloric atresia	1 (2)

4. Discussion

In this study of infants with congenital proximal GIO managed at single tertiary care referral center, the presence of a double bubble on prenatal ultrasound was associated with a relatively high diagnostic accuracy rate, including a sensitivity of 85% and a specificity of 98%. Three (5%) neonates with proximal GIO escaped antenatal detection. Although the presence of a prenatal double bubble in conjunction with polyhydramnios increased the specificity for proximal GIO to 100%, having both features simultaneously was associated with reduced sensitivity (69%) as some GIO had either double bubble or polyhydramnios. Our fetal detection rates for proximal GIO based on the double bubble sign are notably higher than previously reported rates ranging from 52 to 73% [8,11]. Most of these studies evaluate a smaller series of patients and/or were conducted in an era or setting when ultrasound technology was less widely employed in prenatal care.

Table 2
Diagnostic accuracy of prenatal ultrasound for proximal gastrointestinal obstruction.

Test	Sensitivity	Specificity	Positive predictive value	Negative predictive value
Double bubble, % (95% CI)	85.2 (72.9–93.4)	97.5 (86.8–99.9)	97.9 (86.9–99.7)	83.0 (71.9–90.3)
Double bubble and Polyhydramnios, % (95% CI)	68.5 (54.5–80.5)	100 (90.5–100)	100 (99.9–100)	70.2 (61.4, 77.8)

Abbreviations: CI – confidence interval.

Table 4
Postoperative outcomes of proximal gastrointestinal obstruction neonates, by cardiac anomaly status.

	Cardiac Anomalies n = 20	No Cardiac Anomalies n = 36	Overall	p values
LOS (days), median (IQR)	33.0	21.5	27.5	0.099
Postop LOS (days), median (IQR)	30.0	19.5	24.5	0.099
Postop day for initial feeds, median (IQR)	8.0	7.0	8.0	0.255
Postop day for full feeds, median (IQR)	21.0	18.0	19.0	0.949
TPN at discharge, n (%)	0 (0)	1 (3)	1 (2)	>0.999
Tube feeds at discharge, n (%)	8 (40)	9 (25)	17 (30)	0.363
Any complication, n (%)	9 (45)	6 (17)	15 (27)	**0.030**
Revisions, n (%)	2 (10)	0 (0)	2 (4)	0.123
Pneumonia, n (%)	1 (5)	0 (0)	1 (2)	0.357
Surgical site infection, n (%)	2 (10)	0 (0)	2 (4)	0.123
Blood transfusion, n (%)	7 (35)	5 (14)	12 (21)	0.092
Small bowel obstruction, n (%)	2 (10)	1 (3)	3 (5)	0.288

Bold signifies $p < 0.05$.

Another important finding from our study was that 20% of proximal GIO fetuses had no evidence of a double bubble noted at the 20-week anatomic survey, but eventually developed suspicious findings later in gestation. In our cohort, increasing maternal fundal height was the most common indication for rescanning these fetuses. Our median gestational age at diagnosis was approximately 26 weeks, which is similar to reported of 26–31 weeks as described elsewhere [8,10,14,15]. A recent study showed that women receiving prenatal care beginning in first trimester were likely to be diagnosed with fetal proximal GIO after 24 weeks [14].

Nearly 90% of the proximal GIOs in our series were duodenal atresia/webs with or without annular pancreas that most pediatric surgeons would manage by open or laparoscopic duodenoduodenostomy. However, it is important for clinicians to maintain a wide differential diagnosis that would include a duplication cyst and *in utero* midgut volvulus, which was present in 5% of our study and has a dramatically different prognosis in the majority of cases. Like congenital duodenal obstruction, fetal midgut volvulus can present with double bubble and dilated bowel, but not always with polyhydramnios. Midgut volvulus can result in short gut syndrome and parenteral nutrition dependance along with well-documented morbidities such as central line infection and liver failure [16,17] Our data revealed that these volvulus patients had a longer postoperative LOS, took longer to initial and full feeds, and required tube feeds and/or total parenteral nutrition at discharge. Mortality rates in this cohort range from 14 to 40% have been reported [18,19].

Our study confirmed the high concomitant rate (35%) of cardiac anomalies and duodenal atresia as documented elsewhere [20]. The presence of cardiac anomalies has been associated with higher morbidity and mortality in duodenal atresia [20,21] Consequently, parents and practitioners should be aware the complication rate was significantly higher (28%) in those with cardiac anomalies. Our findings support the use of prenatal and postnatal echocardiography should prenatal proximal GIO be suspected given its implications on early neonatal outcomes.

Despite the aforementioned findings of this study, there are several noteworthy limitations. First, most of the data were retrospectively collected, and thus subject to errors due to misclassifications, clinical documentation, or admissions at other facilities. Second, this was a single-center analysis of a relatively rare disease and are thus subject to the shortcomings of smaller sample sizes. We would encourage multi-institutional collaboration among fetal centers to validate our data. Finally, not all of the ultrasounds were done at the same facility or initially read by the same maternal-fetal medicine specialist and thus are subject to potential issues with inter-rater reliability. Our results may therefore have limited generalizability based on institutional expertise and resources.

In conclusion, this contemporary series shows that fetal sonography had high diagnostic accuracy for detecting proximal gastrointestinal obstruction and that the presence of double bubble and polyhydramnios was highly specific for the disease. Although uncommon, *in utero* volvulus is an important anomaly in the differential diagnosis since it was associated with an increased LOS and longer time to full enteral feeds. These data should guide pediatric surgeons, neonatologists, and other perinatal providers in accurately counseling families and directing care.

Disclosure

The authors report no proprietary or commercial interest in any product mentioned or concept discussed in this article.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Conflicts of interest

The authors have nothing to declare.

Acknowledgments

None.

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