CASE REPORT

Prenatal diagnosis of megaduodenum using ultrasound: a case report

Kaihui Zeng\textsuperscript{1}, Dongmei Li\textsuperscript{2}, Yao Zhang\textsuperscript{1}, Chengcheng Cao\textsuperscript{3}, Ruobing Bai\textsuperscript{4}, Zeyu Yang\textsuperscript{1} and Lizhu Chen\textsuperscript{1*}

Abstract

Background: Congenital megaduodenum is a rare disorder; however, its prenatal diagnosis has not been reported previously. We report the case of an abdominal cystic mass in a fetus that was later diagnosed as megaduodenum.

Case presentation: An abdominal cystic mass was found during ultrasonography of a fetus at 11 weeks of gestation. The mass progressively enlarged with gestation. The amniotic fluid volume decreased and then returned to normal. During the last prenatal ultrasound examination, the mass was observed communicating with the stomach; therefore, duodenal dilation was suspected. Finally, the patient was diagnosed with megaduodenum caused by a developmental defect in the nerve plexus.

Conclusions: Congenital megaduodenum is a differential diagnosis of massive fetal abdominal cystic masses. Ultrasound examinations of such masses communicating with the stomach may help determine the diagnosis.

Keywords: Megaduodenum, Fetus, Prenatal diagnosis, Ultrasound

Background

Megaduodenum is a rare clinical syndrome caused by mechanical or functional abnormalities, which is characterized by remarkable expansion of the duodenum. Usually, there is significant expansion of the descending part, horizontal part, and ascending part of the duodenum, and the diagnosis can be made when the diameter of the duodenum is larger than 5 cm \cite{1}. Megaduodenum was first reported by Melchior \cite{2} in 1924; however, only a few such cases in adults and children have been reported \cite{3–7}. The prenatal diagnosis of megaduodenum has not been previously reported. We report the case of an abdominal cystic mass in a fetus detected at 11 4/7 weeks of gestation that was later diagnosed as megaduodenum following postnatal surgery and histopathological examination.

Case presentation

A 30-year-old woman (gravida 4, para 0) who conceived by in vitro fertilization presented with a fetus with a cystic abdominal mass detected on fetal nuchal translucency examination at 11 4/7 weeks of gestation. She was referred to our hospital for a fetal ultrasound consultation at 20 3/7 weeks of gestation. The findings of the ultrasound examination are shown in Table 1 and Fig. 1. The cystic mass enlarged with gestational age. During the last prenatal examination at 33 3/7 weeks of gestation, emptying of the stomach was observed. Fluid passed through the pylorus into the cystic mass; the fluid content in the mass was thick, allowing clear visualization of the fluid flow from the stomach into the cystic mass (Additional file 1).

Based on this finding, duodenal dilation was suspected. Amniocentesis was performed, and the karyotype was found to be normal. Comparative genomic hybridization (CGH) array revealed microdeletions on chromosome 16 [del (16) (p12.2) chr16: g. 22000000_22440000 del] and microduplications on chromosome 9 [dup (9) (p24.1) chr9: g. 6320000_7420000 dup]. Public databases of DGV, DECIPHER, OMIM, ClinGen, UCSC, gnomAD, and PubMed.

\* Correspondence: aliceclz@sina.com

1Department of Ultrasound, Shengjing Hospital of China Medical University, No.36 Sanhao St, Shenyang 110004, Liaoning, China

Full list of author information is available at the end of the article.
were checked. The microdeletions on chromosome 16 were at high risk of pathogenic variations that could manifest as growth restriction, language delay, mild to moderate mental deficiency, mental and behavioral abnormalities, congenital heart disease, and bone malformations; however, their associations with megaduodenum have not been reported.

Microduplications on chromosome 9 were regarded as variants of unknown significance. Because the abnormalities on chromosome 16 were inherited from the father who presented with a normal phenotype, the woman decided to continue the pregnancy.

Spontaneous preterm labor occurred at 33 3/7 weeks of gestation. Because of the breech position of the fetus, cesarean delivery of a female neonate (birth weight, 2240 g) was performed at 34 1/7 weeks of gestation. The child was kept on fasting, and transabdominal ultrasonography was performed 10 h after birth. The ultrasound findings are presented in Fig. 2. Upper gastrointestinal contrast showed that the gastric antrum had moved upward, the descending part of the duodenum was slightly compressed, the duodenum had expanded, and the contrast agent passed through the duodenum smoothly (Fig. 3a).

The child underwent surgery 2 days after birth. Exploratory laparotomy revealed a giant duodenal loop with a diameter of approximately 6 cm and intestinal malrotation (Fig. 3b). The proximal jejunum had a

| Gestational age | 11 4/7 weeks | 15 4/7 weeks | 20 3/7 weeks | 25 3/7 weeks | 29 3/7 weeks | 33 3/7 weeks |
|----------------|-------------|-------------|-------------|-------------|-------------|-------------|
| Gestational age estimated by ultrasound | 11 5/7 weeks | 15 1/7 weeks | 19 3/7 weeks | 23 3/7 weeks | 28 2/7 weeks | 33 4/7 weeks |
| Cyst size, cm | 0.7 × 0.6 × 0.5 | 1.5 × 1.0 × 0.7 | 3.3 × 1.7 × 1.6 | 4.6 × 4.8 × 2.5 | 7.1 × 6.7 × 4.0 | 9.0 × 7.9 × 5.8 |
| Internal echo findings of the cyst | Oval shape, anechoic | Irregular shape, anechoic | Irregular shape, homogenous low-level echogenicity of the cyst fluid, echogenic wall | Irregular shape, homogenous low-level echogenicity of the cyst fluid, echogenic wall | Irregular shape, homogenous low-level echogenicity of the cyst fluid, echogenic wall | Irregular shape, homogenous low-level echogenicity of the cyst fluid, echogenic wall |
| Amniotic fluid volume, DVP | 3.6 cm | 3.4 cm | 3.2 cm | 2.5 cm | 2.7 cm (AFI, 7) | 4.7 cm (AFI, 13) |
| Additional findings | – | – | – | – | – | The cystic mass communicated with the stomach and the cervix was open |

**Table 1** Prenatal ultrasound findings of the fetus

_AFI_ amniotic fluid index, _DVP_ depth of the vertical pocket

Fig. 1 Ultrasound findings of six prenatal examinations: **a** 11 4/7 weeks of gestation; **b** 15 4/7 weeks of gestation; **c** 20 3/7 weeks of gestation; **d** 25 3/7 weeks of gestation; **e** 29 3/7 weeks of gestation; **f** 33 3/7 weeks of gestation. The arrow indicates the cystic mass
diameter of approximately 0.5 cm, and no obvious obstruction was observed. The large duodenal loop was surgically removed, and duodenoejejunostomy was performed. Most of the intermuscular plexus was not visible and ganglion cells were not observed on histopathological examination (Fig. 3c).

The child was in a good condition and was discharged from the hospital on day 13 after surgery. During the follow-up examination at 3 months after surgery, the child was found to be in good health.

Discussion and conclusions
Megaduodenum is a clinical syndrome with multiple causes; the condition is divided into two categories, namely, obstructive and non-obstructive, based on whether there is genuine obstruction at the distal end of the duodenum. Obstructive factors may be extra-duodenal (superior mesenteric artery syndrome, annular pancreas, tumors, congenital bands), related to the duodenal wall (duodenal diverticulum, duodenum inflammation), or related to the duodenal lumen (atresia, gallstones, bezoars, duodenal septum stenosis, parasite infections) [8]. Non-obstructive factors include congenital diseases (familial inherited megaduodenum, congenital ganglion cell deficiency) and acquired diseases (systemic lupus erythematosus, amyloidosis, diabetes) that affect the smooth muscles of the intestine or the plexus of the intestinal wall [9, 10]. This case was attributed to ganglion dysplasia, which is a non-obstructive factor.

To the best of our knowledge, this is the first report of a case of fetal megaduodenum. The dilated intestine lost its normal shape and underwent massive expansion during the second and third trimesters. The expanded intestine was connected to the stomach, which was not dilated and could be emptied. In addition, the colon and rectum were found to be normal, and polyhydramnios did not occur. Because this case was caused by abnormal development of the nerve plexus, its indications appeared early on ultrasound.
Furthermore, the lower duodenal obstruction was not obvious; therefore, the manifestations found during ultrasound were different from those of obstructive diseases. Fetal megaduodenum must be distinguished from meconium pseudocyst and duodenal stenosis or atresia; the main identification points are summarized in Table 2.

Patients with megaduodenum often present with recurrent abdominal distention, nausea, vomiting, diarrhea, malnutrition, and dystrophia [15]. Because this case was diagnosed before birth, the child was kept fasting immediately after birth and she had no other clinical manifestations except abdominal distension. The earliest reported case of megaduodenum was discovered in a 3-month-old infant; the clinical symptoms included failure to gain weight, an abdominal mass, nausea, and vomiting [5]. That child was followed up for 3 years after surgery, gained adequate weight, and was able to lead a healthy life [5].

Megaduodenum is mainly treated by surgery. The tension-free duodenal wall should be completely removed to prevent postoperative functional anastomotic obstruction. For megaduodenum secondary to other diseases, the primary disease must be addressed first [6, 8, 16]. In our case, extreme dilation of the duodenum was caused by a defect in the development of the nerve plexus, and radical duodenectomy was performed. Three months after surgery, the child was in good health. Further follow-up is required to determine the long-term prognosis.

In conclusion, although congenital megaduodenum is rare, it should be considered as part of the differential diagnoses of fetal abdominal cystic masses. In particular, if a giant cystic mass communicating with the stomach is detected on ultrasonography, the prenatal diagnosis of congenital megaduodenum should be considered.

**Table 2** Differential diagnosis of megaduodenum

| Differential diagnosis       | Time of discovery | Stomach size | Communication between the mass and stomach | Amniotic fluid | Mass characteristics | Other manifestations on ultrasound |
|-----------------------------|-------------------|--------------|-------------------------------------------|----------------|----------------------|-----------------------------------|
| Megaduodenum                | Can be found during early pregnancy | Normal       | Yes                                       | Normal         | Enlargement with gestational week, irregular shape, echogenic wall | –                                 |
| Duodenal stenosis or atresia [11, 12] | Usually found during the second trimester | Dilated       | Yes                                       | Often accompanied by polyhydramnios | Spherically dilated duodenum | “Double-bubble” sign, a shriveled bowel can sometimes be observed at the distal end |
| Meconium pseudocyst [13, 14] | Mid-to-late pregnancy | Normal       | No                                        | Often accompanied by polyhydramnios | Large cystic mass with echogenic walls and some septa | Ascites, scattered intra-abdominal calcifications |

**Supplementary Information**
The online version contains supplementary material available at https://doi.org/10.1186/s12884-021-03843-0.

**Additional file 1.** Gastric contents emptying into the cystic mass. We clearly observed the emptying of the stomach and fluid passing through the pylorus into the cystic mass.

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**Authors’ contributions**
KHZ was responsible for the original draft preparation and editing. DML, YZ, CCC, and RBB participated in collecting the data and were major contributors during the writing of the manuscript. LZC and ZYY were responsible for conceptualization, manuscript editing, and reviewing the manuscript. All authors read and approved the final manuscript.

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**Availability of data and materials**
All data analyzed during this study are included in this report.

**Declarations**

**Ethics approval and consent to participate**
The patient described in this case report provided informed consent. Identifying data (birthdate and name) has been removed. The study was approved by the Ethics Committee of Shengjing Hospital, China Medical University.

**Consent for publication**
Written informed consent was obtained from the patient for publication of this Case report and any accompanying images.

**Competing interests**
The authors declare that they have no competing interests.

**Author details**
1Department of Ultrasound, Shengjing Hospital of China Medical University, No.36 Sanhao St, Shenyang 110004, Liaoning, China. 2Department of Ultrasound, Shenyang Women’s and Children’s Hospital, Shenyang, Liaoning, China. 3Department of Pathology, Shengjing Hospital of China Medical University, Shenyang, Liaoning, China. 4Department of Radiology, Shengjing Hospital of China Medical University, Shenyang, Liaoning, China.

**Abbreviations**
AFI: Amniotic fluid index; CM: Cystic mass; DVP: Depth of the vertical pocket; GB: Gallbladder; ST: Stomach
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