Case Report

A Case of Ultrasound Diagnosis of Fetal Hiatal Hernia in Late Third Trimester of Pregnancy

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Congenital hiatal hernia is a condition characterized by herniation of the abdominal organs, most commonly the stomach, through a physiological but overlax gastroesophageal junction into the thoracic cavity. The prenatal diagnosis of CHH is unusual and in the literature only eight case reports of prenatal diagnosis of this anomaly have been reported [1–5].

The CHH is not commonly considered in the differential prenatal diagnosis of cystic chest anomalies. In this paper we report a case of hiatal hernia, suspected in the late third trimester of gestation, and present a review of the literature on the subject.

1. Introduction

Congenital hiatal hernia (CHH) is characterized by the herniation of the abdominal organs, most commonly the stomach, through a physiological but overlax gastroesophageal junction into the thoracic cavity. The prenatal diagnosis of CHH is unusual and in the literature only eight case reports of prenatal diagnosis of this anomaly have been reported [1–5].

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2. Case Report

A 37-year-old woman (gravid 2, para 1) was referred to our fetal unit at 39 weeks’ gestation for evaluation of what appeared to be a small cystic mass in the fetal thorax. The pregnancy had been otherwise uneventful. Family history was unremarkable. Amniocentesis for karyotyping had been performed at 16 weeks for advanced maternal age and revealed a normal karyotype (46, XY). According to Italian guidelines, the patient had undergone ultrasound examinations at 20 and 32 weeks’ gestation, and the stomach was normally seen located on the left in the abdomen. Routine ultrasound examination performed at 39 weeks showed a fetus with normal growth. The ultrasound examination was performed because, in our hospital, all patients are evaluated with a clinical and ultrasound examination at 39 weeks of gestation. On the transverse section of the thorax the heart appeared normally located, without mediastinal shift, and the lungs appeared to be of normal echogenicity and volume. A round cystic image, diameter 8 mm, was visible behind the heart in the posterior mediastinum (Figure 1); in sagittal section it appeared to be in continuity with a small stomach located below the diaphragm (Figure 2).

During the ultrasound exam period, no modification of the cystic structure was detected. One possible diagnosis was hiatal herniation of the stomach into the thorax and...
counseling about the diagnostic suspicion of hiatal hernia, the
differential diagnosis, and the likely postnatal complications
was performed.
At 40 weeks' gestation, the patient was admitted for
spontaneous labor. A male newborn, weighing 3460 g, was
delivered vaginally with Apgar scores of 9 and 10 at 1st and
5th min, respectively. Postdelivery chest radiography revealed
no pulmonary abnormalities and a normal diaphragmatic
profile (Figure 3).

The upper gastrointestinal tract series using barium
confirmed the prenatal diagnosis of CHH, categorized as
type I. During the examination, gastroesophageal reflux was
observed, together with a sliding up and down of the stomach
into the fetal thorax. The newborn was discharged with
prophylactic therapy with ranitidine and domperidone. At
six-month follow-up the baby is growing well with sporadic
episodes of reflux and he is still on therapy.

3. Discussion

Hiatal hernia (HH) is defined as a herniation of the abdom-
nal organs into the chest through the oesophageal diaphrag-
matric hiatus [3]. This is a well-recognized anomaly in children
but most reported cases refer to adult patients. Its etiology
is not precisely known, but the hypothesis of its congenital
origin is widely accepted. Similarly to other congenital
diaphragmatic defects, HH follows a sporadic pattern of
incidence in most cases. Familial occurrence of sliding HH
has been reported in more than 20 cases, but only one
family with two members affected has been described in the
literature. The presence of HH in the newborn should alert
the clinician to suspect connective tissue disorders such as
Ehlers-Danlos syndrome, cutis laxa, and Marfan syndrome
[6].

Hiatal hernia often involves parts of the stomach and
rarely other abdominal viscera and it is generally classified by
the position of the gastroesophageal junction relative to the
diaphragm. The gastroesophageal junction is a complex valve
composed of a smooth muscle element and a diaphragmatic
element. These normally supplement each other to maintain
competence in a static condition and during dynamic stress
associated with increased intraabdominal pressure [7]. Hiatal
hernia is classified as follows: type I, which is a sliding hiatus
hernia; type II, also called “rolling” or “pure” paragastrohe-
phageal hernia that may result from primary diaphragmatic defects;
type III, which is a complex mix of types I and II, with
migration of the gastroesophageal junction up into the
thorax through the hiatus and an additional paragastrohe-
phageal component; and type IV, essentially a large type III hernia,
which may contain the entire stomach, omentum, or other
abdominal viscera [6]. Prenatal diagnosis of HH is unusual
and to our knowledge only eight cases have been reported in
the literature [1–5].

In all reported cases of CHH, the condition was identified
in the third trimester of gestation. This late onset may be
due to the fact that the fetus may be unable to develop
sufficient pressure to dilate the intrathoracic stomach until
the third trimester. In the third trimester, the development
of adequate pressure during swallowing, probably with the
effect of reflux of gastric secretions into the esophagus, could
result in visualization of the dilated esophagus/stomach at
ultrasound [4].
In the majority of the reports, CHH was an isolated anomaly [1, 3, 4]. However, Yamamoto et al. describe a prenatal diagnosis of type II CHH associated with asplenia syndrome (congenital absence of the spleen, dextrocardia, atrioventricular septal defect, pulmonary stenosis, and intestinal malrotation) [5]. In this fetus, the stomach was observed as intermittently herniating into the thoracic cavity. An intermittent gastric herniation was also described by Ogunyemi in a case of CHH associated with polyhydramnios; in this case the presence of hydramnios was probably due to esophageal atresia and the differential diagnosis with esophageal atresia was made by the authors [1]. In three cases, including ours, the stomach was normally seen in the abdomen, while in three other fetuses the stomach was completely herniated into the chest and no intraabdominal stomach was visible; one of these fetuses was affected by CHARGE syndrome [2]. In none of the reported cases was CHH associated with mediastinal shift or pleural/pericardial effusion.

The presence of a cystic mass in the fetal thorax should alert the sonographer to consider in the differential diagnosis congenital chest anomalies such as diaphragmatic hernia, macrocystic adenomatoid malformation of the lung, or rarer conditions such as esophageal duplication or neurenteric cyst [4].

The sonographic criteria for the prenatal diagnosis of CHH were first proposed by Bahado-Singh et al. [3] and then refined by Ruano et al. [4]. The criteria suggested are helpful for making the correct diagnosis and are as follows: (1) the presence of a hypoanechoic mass in the posterior mediastinum, just behind the heart and anterior to the vertebral body, corresponds to a herniated stomach; (2) there is neither mediastinal shift nor pleural or pericardial effusion; the stomach can be identified in the abdominal cavity but in a median position or is not visible in the abdomen because of its complete herniation into the fetal thorax; (3) the dynamic aspect of the herniated stomach means its up–down movements through the enlarged hiatus into the thoracic cavity.

Although congenital hiatal hernia is never a critical condition, it cannot be considered entirely free from significant morbidity. CHH does not carry the same risks as those seen in adult cases. Nonetheless, in the infants and children the herniated stomach into the thorax with continuing acid regurgitation can be complicated by peptic ulceration, severe esophagitis, pyloric stenosis, vomiting, gastric volvulus, aspiration pneumonia, and pulmonary fibrosis [8]. Complications are more prevalent in paraesophageal hernias than in sliding hiatus hernias and elective repair is advocated on diagnosis even in asymptomatic patient in order to prevent life-threatening complication and to avoid the significant morbidity and mortality of an emergency operation [8]. Hiatal hernia is a mechanical defect that will progress so that medical therapy is ineffective and pediatric literature supports routine elective repair of the hernia in infants with congenital HH. The principles of repair consist of reduction of the hernia, partial or complete excision of the sac to prevent recurrence, and crucial approximation followed by an antireflux fundoplication [6, 8].

Congenital hiatal hernia is uncommon and it is rarely prenatally diagnosed but it should be included in the differential diagnosis of any chest anomalies sonographically detected because an early diagnosis and an appropriate postnatal management avoid unnecessary morbidity and even mortality.

**Conflict of Interests**

The authors (Stefania Di Francesco, Mariano Matteo Lanna, Marcello Napolitano, Luciano Maestri, Stefano Faiola, Mariangela Rustico, and Enrico Ferrazzi) declare that there is no conflict of interests regarding the publication of this paper.

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