What’s Your Diagnosis? A Case Study

Case Studies | August 01, 2013
By Joe Antony, MD

When this 32-year-old woman went for an ultrasound at 27-weeks into her pregnancy, a rare anomaly was found. Can you diagnose it?

This is the case of a 27-week-old gestation for which the mother underwent routine ultrasound imaging.

History and symptoms: This 32-year-old mother came for routine sonographic examination at 27-weeks gestational age with a history of normal menstrual cycles prior to the pregnancy. She had no history of any major illness before the sonographic examination.
Family history: This patient, the mother, was one of two children. This was her first pregnancy.
Present pregnancy: The patient had a history of 27-weeks amenorrhea which corresponded with the age of the fetus on ultrasound scan. She had no history of antepartum hemorrhage or discharge during this pregnancy.
Clinical examination: Her blood pressure was normal (BP: 118/78 mm. of Hg); her other vital signs were normal. On examination of the chest there were no abnormalities. On abdominal examination, the fundal height of the patient corresponded to the gestational age of 27-weeks.
Imaging studies: This patient underwent routine transabdominal ultrasound imaging to study the biophysical profile of the fetus and to exclude any congenital anomalies. She had not undergone any previous ultrasound scan. Ultrasound images of the current sonographic examination are shown on the following pages.

Image-01: Transabdominal ultrasound

What do you observe in this ultrasound image of the fetal thorax?

Firstly, let us try to identify the parts of the fetal body being studied in these images. The first image shows the fetal chest. The question that arises is whether there is any abnormality in this image? Yes! Of course! Let us try to describe the findings in this image. There is a mass-like lesion along the anterior aspect of the fetal chest. What can this be? Is it a sternal mass? Is it a loop of the umbilical cord? Is it an umbilical cord mass? Study the image carefully- you will observe that this “mass” is continuous with fetal interior. So the question that arises is—is it a pulmonary mass? Is it a cardiac
mass? Look carefully at image 1. The lesion appears continuous with the fetal heart. So is it a cardiac mass? This axial section image shows that the lesion is part of the heart itself. So do we have a diagnosis based on this image? What can give rise to a lesion that is continuous with the heart and extends outside the fetal chest? The most obvious answer is a possible ectopia of the fetal heart—what do you call this? Yes! This is ectopia cordis; or herniation of the fetal heart outside the thoracic cage. Now, shall we study the second image?

**Image-02: Transabdominal ultrasound of the fetal abdomen**

The second image shows the fetal abdomen. What do you think is abnormal in this image? This is an axial section through the fetal abdomen. Is it normal? What is the most striking abnormality here? Yes—there is an obvious defect in the fetal abdominal wall. What are the other findings in this image? What has happened to the fetal intestines? They appear to freely float inside the amniotic cavity. What does it mean? Is this then an omphalocele? An omphalocele is described as a herniation of abdominal contents including the intestine into the amniotic cavity but having a thin covering membrane. Is that the case here? Observe the image carefully- the answer is no! What do we call this condition? The answer is gastroschisis.

Now the case gets murkier. There are obviously two major fetal anomalies in this case—the first is ectopia cordis and the second is gastroschisis. Are these two isolated findings which have occurred in the same fetus? The question that arises is—is this a part of a major syndrome? If so, what is the name of the syndrome?

What do all these findings point to? What are the diagnostic possibilities? What do all these findings suggest? Let us list the findings:
1. Fetal sternal and anterior chest wall defect
2. Fetal ectopia cordis
3. Fetal abdominal wall defect
4. Fetal gastroschisis

Does it now strike a chord? Shall we see some more images of this case? In these images, the affected parts are labeled.

**Image-03: Transabdominal image of the fetal chest**
What's Your Diagnosis? A Case Study
Published on Physicians Practice (http://www.physicianspractice.com)

Now do we have a diagnosis? What are the diagnostic possibilities now which can explain all these ultrasound features? Let us list some of them:
1. amniotic band syndrome
2. body stalk abnormality
3. pentalogy of Cantrell

Let us now examine each possibility.

Amniotic band syndrome is characterized by constricting amniotic bands around the fingers, toes or limbs causing swelling of the limbs or the affected parts. In addition a band can often be seen traversing across the amniotic cavity. Is that the case here? No! What we have here has nothing to do with the fetal limbs or fingers. So we can exclude this possibility safely.

Let us now examine the possibility of body stalk abnormality. This syndrome is characterized by multiple anomalies, namely- abdominal wall defects primarily and limb and spine anomalies. In our fetus, the limbs appear normal (not shown here), while there is also ectopia cordis. So we can safely exclude this particular syndrome.

What does that leave us with? Do we have a diagnosis now? Yes! The only possibility that we are left
with is Pentalogy of Cantrell!

**Final diagnosis:** Pentalogy of Cantrell

**Discussion:** Pentalogy of Cantrell is characterized mainly by two features, namely ectopia cordis and a large abdominal wall defect with omphalocele or gastroschisis. As the word pentalogy suggests, the true pentalogy of Cantrell is defined as a syndrome of five major anomalies—ectopia cordis, omphalocele or gastroschisis, sternal defect, defect in the diaphragmatic pericardium, and anterior diaphragmatic defect. In a complete form of pentalogy of Cantrell such as this, the prognosis is very bleak as the chance of survival in the postnatal period is almost nil. Medical termination of pregnancy is often offered if a prenatal diagnosis is convincing.

**Sonographic features:**
The chief diagnostic features of pentalogy of Cantrell in a fetus are:

1) ectopia cordis
2) abdominal wall defect
3) gastroschisis or omphalocele
4) the other features include sternal defect and
5) defect in the diaphragmatic pericardium

Very often, the chief features seen are only omphalocele or gastroschisis with an associated ectopia cordis. There are also milder forms of pentalogy of Cantrell, wherein the features may be limited to one or two findings with the other findings absent or partial.

**Source URL:**
http://www.physicianspractice.com/obgyn-ultrasound/what%E2%80%99s-your-diagnosis-case-study

**Links:**