What is your diagnosis?

A 27 year-old pregnant woman was referred to our perinatology clinic for an abnormal fetal ultrasound finding. She was a nulliparous lady with three gestations of one ectopic pregnancy, one miscarriage and this ongoing pregnancy at its 37th week. In her gestational history she was followed carefully by the same obstetrician with low-risk double and triple test results and second-level prenatal ultrasonography.

We detected a hypoechogenic, regular bordered, sausage-like cystic mass behind the heart, between the thoracic aorta and right atrium (Figure 1, 2). The fetus had compatible measurements with its gestational age except that its head circumference was 100 mm (41 weeks). There was no other ultrasonographic abnormality detected. Amniotic fluid index was within the normal range (9 cm). The heart was left sided and had normal 4 chambers with normal three vessels and trachea view. What is your diagnosis?

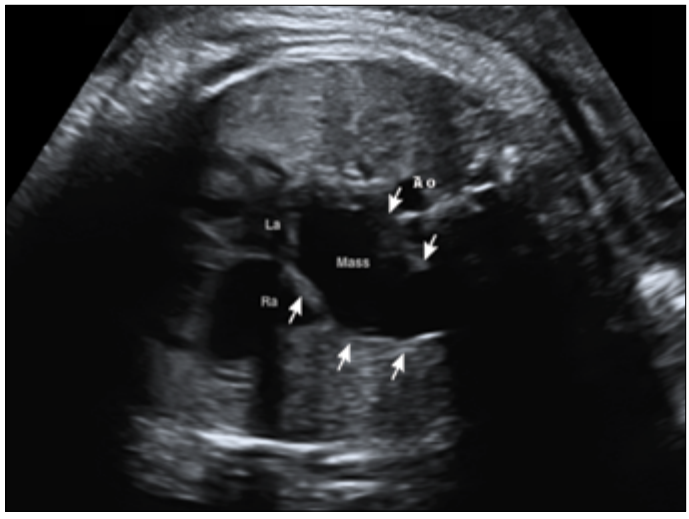

Figure 1. The arrows show the borders of the mass. Ao indicates the thoracic aorta , La indicates the left atrium and Ra the right atrium

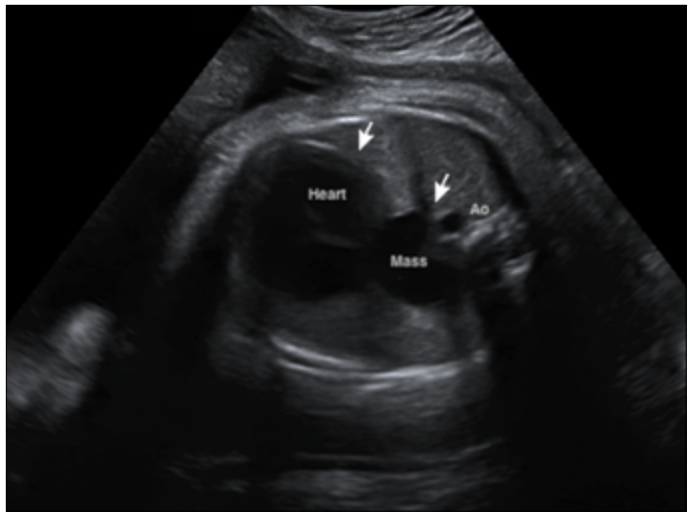

Figure 2. The relation of the heart, mass and aorta

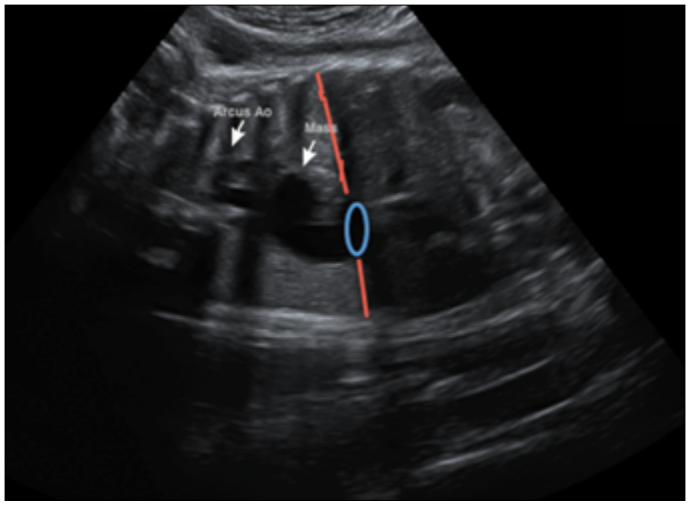

Figure 3. In this longitudinal view the red line shows the diaphragm and the blue circle shows the diaphragmatic hole

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Answer

Detailed examination of the internal organs revealed an absent stomach. With attention directed to this, although the diaphragm seemed intact and the lungs were normal in shape and volume, a stomach sliding to the thorax through diaphragm defect was detected (Figure 3).

Congenital diaphragmatic hernia (CDH) has an incidence of approximately 1/3,000 live births (1, 2). The primary embryological events leading to hernia remain largely unknown (3). Identification of the causes is challenging because of its heterogeneous and multifactorial character (4). It occurs as an isolated malformation in around 60% of cases, usually being sporadic.

Congenital diaphragmatic hernia can generally be classified into two types depending on its position: posterolateral and nonposterolateral. It is often located on the left side (85%) and least frequently located bilaterally (2%). The posterolateral defect- generally named as a Bochdalek hernia- is seen in 70-75% of cases (1). Nonposterolateral defects can be sub-divided into anterior and central types. The anterior type occurs in approximately 23-28% of cases. As in our patient, central hernias account for the remaining 2-7% of cases.

In our case, the fetus had a small, central defect of the diaphragm. Despite the expected, the fetus had no other abnormalities. These small defects may not be visible at the time of second level ultrasonography and may become prominent with advancing gestation.

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