Prenatal diagnosis of left isomerism with normal heart: a case report

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Summary

Left isomerism, also called polysplenia, is a laterality disturbance associated with with paired left-sidedness viscera (left atrial appendages, bilobed lungs and long hyparterial bronchi) and multiple small spleens. Left isomerism, heart congenital abnormalities and gastrointestinal malformation are strongly associated.
In this paper we present a case of prenatal diagnosis of left isomerism in a fetus with a structurally normal heart.

KEY WORDS: left isomerism, normal heart, right sided stomach, gut malrotation

Case report

A 36 year old pregnant came to our attention at 25 weeks of gestation because of diagnosis of "isolated" right sided stomach at anomaly scan. The woman was at her first pregnancy and her personal and family history was unremarkable. Genetic amniocentesis, performed at 16 weeks due to advanced maternal age, revealed a normal fetal kariotype (46, XX).
We performed a detailed ultrasonographyc examination included fetal echocardiography.
Presence of right sided stomach was confirmed. Fetal echocardiography showed a normally developed heart but absence of inferior vena cava with azygos continuation to superior vena cava. Not rhythm abnormalities were present during the scan.
Not other abnormalities were seen and fetal biometry and amniotic fluid index were normal.
After the first scan, we performed ultrasonographic examinations every two weeks until delivery to ascertain a normal heart rhythm and to exclude other anomalies. After the 31 weeks of gestation the fetus showed an increased amniotic fluid index and developed a polyhydramnios at 35 weeks.
The baby was delivered at 37 weeks. Newborn weight was 3070 gr. The apgar score at birth was 9 at 1 minute and 10 at 5 minutes.
ECG and chest RX at birth were normal. Neonatal echocardiography confirmed all prenatal findings.
Abdominal ultrasonography, liver scan and upper gastrointestinal study revealed presence of right sided stomach, polysplenia and gut malrotation. Surgery was performed because of presence of partial gut occlusion.
Following abdominal and cardiac examinations during the first 4 months of life were normal (Figs. 1 and 2).

Discussion

The syndrome of left atrial isomerism, also called polysplenia, is associated with paired left-sidedness viscera (left atrial appendages, bilobed lungs and long hyparterial bronchi) and multiple small spleens.
In echocardiography the situs is traditionally deduced from the arrangement of the great vessels in abdomen. Usually, the aorta is posterior and at the left of the spine, while inferior vena cava is more anterior and at the right of the spine.
An important marker of left isomerism is an interrupted inferior vena cava (IVC) with azygos continuation (1). The azygos vein can be seen as a venous structure posterior to the aorta both in sagittal and transverse section of fetal abdomen and chest.
Diagnosis of fetal left isomerism is made in presence of combination of viscerocardiac heterotaxy, complex cardiac malformations, heart block, and interruption of the inferior vena cava with azygos continuation (2). In our case diagnosis of left isomerism, even in presence of normal heart, was made due to the presence of interrupted inferior vena cava and right sided stomach.
The mortality in fetuses and neonates is high in the pre-

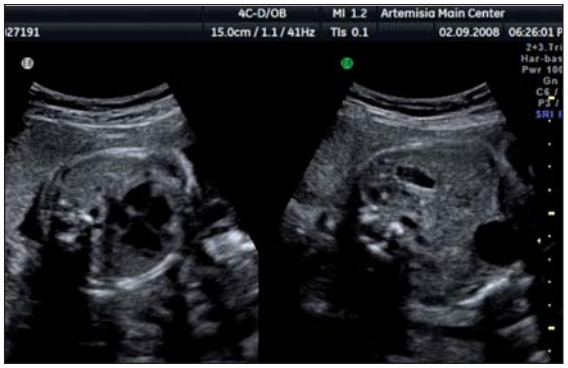

Figure 1 - On the left: trasversal section of the chest showing the heart in normal position. Behind the left atrio is possible to see the aorta and azygos vein. On the right: trasversal chest of the abdomen showing stomach on the right (head above the the level of the abdominal scanning plane).

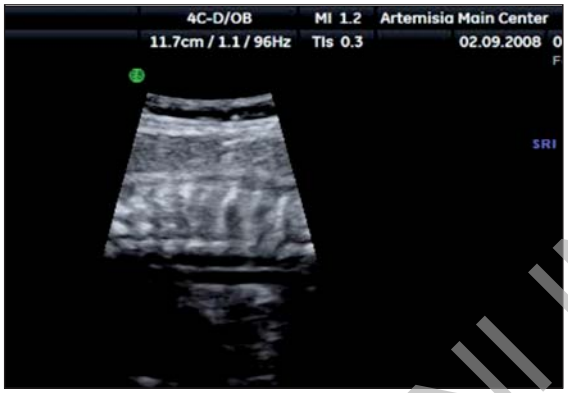

Figure 2 - Sagittal view of the chest showing the azygos vein posterior to the aorta.

sence of heart block and hydrops, whereas the morbidity is mainly determined by the cardiac and extra cardiac defects (2).

Heterotaxy, congenital cardiovascular malformation and gastrointestinal abnormalities are strongly associated. (1-5). Furthermore in left isomerism early fetal heart block and hydrops (6, 7) may be associated as cardiac looping and asymmetrical gut morphogenesis are the first events in the embryo which involve left-right asymmetry.

Gut malrotation and biliar atresia are the most common abnormalities reported in about 40-50% of cases (3-5). Disturbances of cardiac rhythm in patients with left isomerism range from 60% in prenatal series (6) to 15% in postnatal series (13).

Presence of normally developed heart have been reported in fetal and pediatric series with a prevalence variable from 3% to 18% (6, 8-12).

For this reason, prenatal diagnosis of left isomerism in presence of normal heart may be overlooked despite possible presence of gastrointestinal abnormalities and rhythm abnormalities.

In this case report we would like to stress on the importance of follow up through the pregnancy in order to exclude signs of gastrointestinal obstruction every times there is a suspicion of left isomerism. In fact, an important aspect in the management of left isomerism syndrome is the high association with gastrointestinal malformation and rhythm abnormalities that may influence neonatal morbidity and mortality.

In conclusion left isomerism syndrome may coexist with a structurally normal heart. For this reason we recommend that the first step during every fetal echocardiography should be the study of great vessels arrangement in the abdomen in order to ascertain the situs solitus.

If prenatal left isomerism is suspected, even in presence of a normal heart, is mandatory to exclude signs of gastrointestinal abnormalities, as late polyhydramnios, and cardiac rhythm disturbance during the pregnancy and neonatal age.

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