

## LEFT ATRIAL ISOMERISM AT 12 WEEKS OF GESTATION - CASE REPORT

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*Left atrial isomerism, also called polysplenia, is a laterality disturbance associated with double left-sidedness and right-sided structures such as the inferior vena cava and the right atrium with sinus node are absent or may have developed abnormally. We report a case of a 12 weeks of gestation fetus presenting: nuchal edema (5.5mm) and beginning hydrops, bradycardia with the heart rate of 50-60 beats/min, heart axis rotated to the right and atrioventricular septal defect. These findings suggested left atrial isomerism. Interruption of the inferior vena cava with azygos continuation or outflow tract abnormalities could not be demonstrated at this early stage of gestation. The mother opted for expectant management and requested invasive testing which showed normal karyotype. The pregnancy stopped at 17 weeks of gestation. The subject is discussed, noting that left atrial isomerism is a rare and severe cardiac abnormality that can be seen more often in early fetal series than at birth due to the increased incidence of fetal demise, as it happened in our case. The systematic evaluation of the fetal heart in the first trimester can diagnose such conditions, and therefore can provide a better understanding of the reasons of the fetal demise and the prognosis for future pregnancies, related to the risk of recurrence and genetic conditions.*

**Keywords:** left atrial isomerism, heterotaxy, first trimester, fetal bradycardia, AVSD

### INTRODUCTION

The embryologic development of the abdominal and thoracic structures follows a spatially controlled and coordinated manner leading to well defined right and left sided anatomic positions within the body. Any arrangement of the abdominal or thoracic organs other than the normal (situs solitus) or the mirror-image of the normal (situs inversus) is referred as heterotaxy syndrome<sup>1,2</sup>. Heterotaxy syndrome is found in 2.2-4.2% of infants with congenital heart disease<sup>3</sup>. Left atrial isomerism, also called polysplenia, is a laterality disturbance associated with double left-sidedness and underdevelopment of right-sided structures. Left isomerism, heart congenital abnormalities and gastrointestinal malformation are strongly associated<sup>1</sup>. The most common association is the absence of the intrahepatic part of the inferior vena cava. In fetal series, left isomerism is more common than in postnatal series due to the increased incidence of fetal demise. Heterotaxy has a risk of recurrence of 10% and the genetic etiology include autosomal dominant, autosomal recessive, X-linked and single gene disorder<sup>3</sup>.

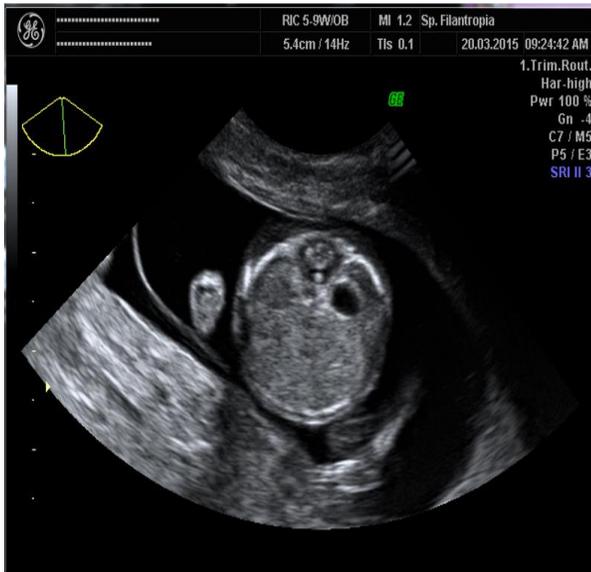
### MATERIAL AND METHODS

We present a case of prenatal diagnosis of left isomerism in a fetus at 12 weeks of gestation. The scan was performed both transabdominal and transvaginal, using Voluson 730 ultrasound system, in mode B, Doppler and pulsed Doppler.

### CASE REPORT

A 28 years old primigravida was referred to our department at 12 weeks of gestation because for bradycardia and nuchal edema. Her personal and family history was unremarkable. We performed a detailed ultrasonographic examination included fetal echocardiography. The ultrasound confirmed these findings (heart rate 50-60b/min and NT- 5.5mm) as well as heart axis rotated to the right and atrioventricular septal defect. Interruption of the inferior vena cava with azygos continuation or outflow tract abnormalities could not be demonstrated at this early stage of gestation. The mother opted for expectant management and requested invasive testing which showed normal karyotype. After the first scan, we performed ultrasonographic examinations every two weeks until 17 weeks of gestation when fetal demise was diagnosed. Figure 1, 2 show transversal

sections of the chest and upper abdomen showing the discordant positions of the fetal heart and stomach. Figure 3 shows a large atrioventricular septal defect.



**Figure 1** Ultrasound image showing a transverse section of the upper abdomen of the fetus with the fetal stomach on the left side. Also, fetal edema can be noted.



**Figure 2** Ultrasound image showing a transverse section of the fetal chest, with the heart pointing to the right side, abnormal and discordant with the stomach. Fetal edema is clearly seen.



**Figure 3** Ultrasound image showing a transverse section of the fetal chest, at the level of the 4-chambers view.

A normal cardiac crux can not be seen. The interatrial septum can not be seen and the interventricular septum has a large defect at the insertion level of the tricuspid and mitral valves, leading to a single atrioventricular valve above a large septal defect, findings consistent with a large atrioventricular septal defect. The cardiac walls appear thick, and there is clear fetal edema, findings consistent with fetal cardiac insufficiency.

## DISCUSSIONS

Left atrial isomerism is most commonly associated (80-90% of cases) with the absence of the intrahepatic part of the inferior vena cava<sup>2-4</sup>. Due to the size of the fetus, and subsequently the size of the thoracic and abdominal structures, this abnormality could not be demonstrated. Another feature of left atrial isomerism is the absence of the morphologic right atrium with its sinus node, leading to bradyarrhythmia up to complete block in 40-70% of cases<sup>3,4</sup> and it was also found in the case we present. The presence of the heart block, associated sometimes with cardiac and extracardiac malformations leads in 30% of the cases to cardiac failure, hydrops and fetal death<sup>3,5</sup>. Other abnormalities that can be associated with left isomerism are: symmetric, left sided liver, right-sided stomach, upper gastrointestinal obstruction, polysplenia, bilobed lungs, abnormal pulmonary venous connection<sup>6,7</sup>. The cardiac axis can be either to the left, middle or right side of the chest<sup>3</sup>. The most common cardiac abnormality was also found in our case is an atrioventricular septal defect, while other cardiac abnormalities

that can be associated with left isomerism include double outlet right ventricle, obstruction of the outflow tracts, left superior vena cava and abnormal venous connection<sup>5</sup>.

## CONCLUSIONS

The subject is discussed, noting that left atrial isomerism is a rare and severe cardiac abnormality that can be seen more often in early fetal series than at birth due to the increased incidence of fetal demise, as it happened in our case. The systematic evaluation of the fetal heart in the first trimester can diagnose such conditions, and therefore can provide a better understanding of the reasons of the fetal demise and the prognosis for future pregnancies, related to the risk of recurrence and genetic conditions. The mortality in fetuses is high in the presence of heart block and hydrops, whereas the morbidity is mainly determined by the cardiac and extra cardiac defects<sup>8</sup>. Findings like hydrops and fetal bradycardia are signs of a poor prognosis that be easily observed<sup>9</sup>. The detailed first trimester assessment can reveal other abnormalities such as discordant position of the fetal heart and stomach, atrioventricular septal defect that can lead to the diagnosis of a rare fetal condition such as left atrial isomerism and can be useful in establishing the recurrence risk in other pregnancies. Therefore, the assessment of the fetal anatomy and a detailed evaluation of the fetal heart must be performed in fetuses with abnormal findings.

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