

## Left atrial isomerism – sonographic signs at 12 weeks

## Isomerismo esquerdo – sinais ecográficos às 12 semanas

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### Abstract

Left atrial isomerism is a rare syndrome responsible for multiple cardiac, visceral and vascular malformations. Due to its high phenotype variability, prognosis can be poor. This report aims to share some unusual ultrasonographic findings of an affected fetus at 12 weeks of gestation. The discrepancy between atrial and ventricular rates hinted towards the assessment of this disorder, at such an early stage. Ultrasonographic superb microvascular imaging technique corroborated the diagnosis. This case highlights the importance of a thorough morphological evaluation at the first trimester scan.

**Keywords:** Left atrial isomerism; Left isomerism; Fetal ultrasound; Early morphology.

### Resumo

O isomerismo atrial esquerdo é uma síndrome rara responsável por múltiplas malformações cardíacas, viscerais e vasculares. Devido à sua alta variabilidade fenotípica, o prognóstico pode ser desfavorável. Este estudo visa partilhar os achados ecográficos incomuns de um feto afetado, às 12 semanas de gestação. A discrepância entre as frequências auriculares e ventriculares sugeriram a pesquisa desta patologia num estadio tão precoce. O diagnóstico foi documentado com o uso do modo Doppler *Superb Microvascular Imaging*. Este caso destaca a importância de uma avaliação morfológica detalhada na ecografia do primeiro trimestre.

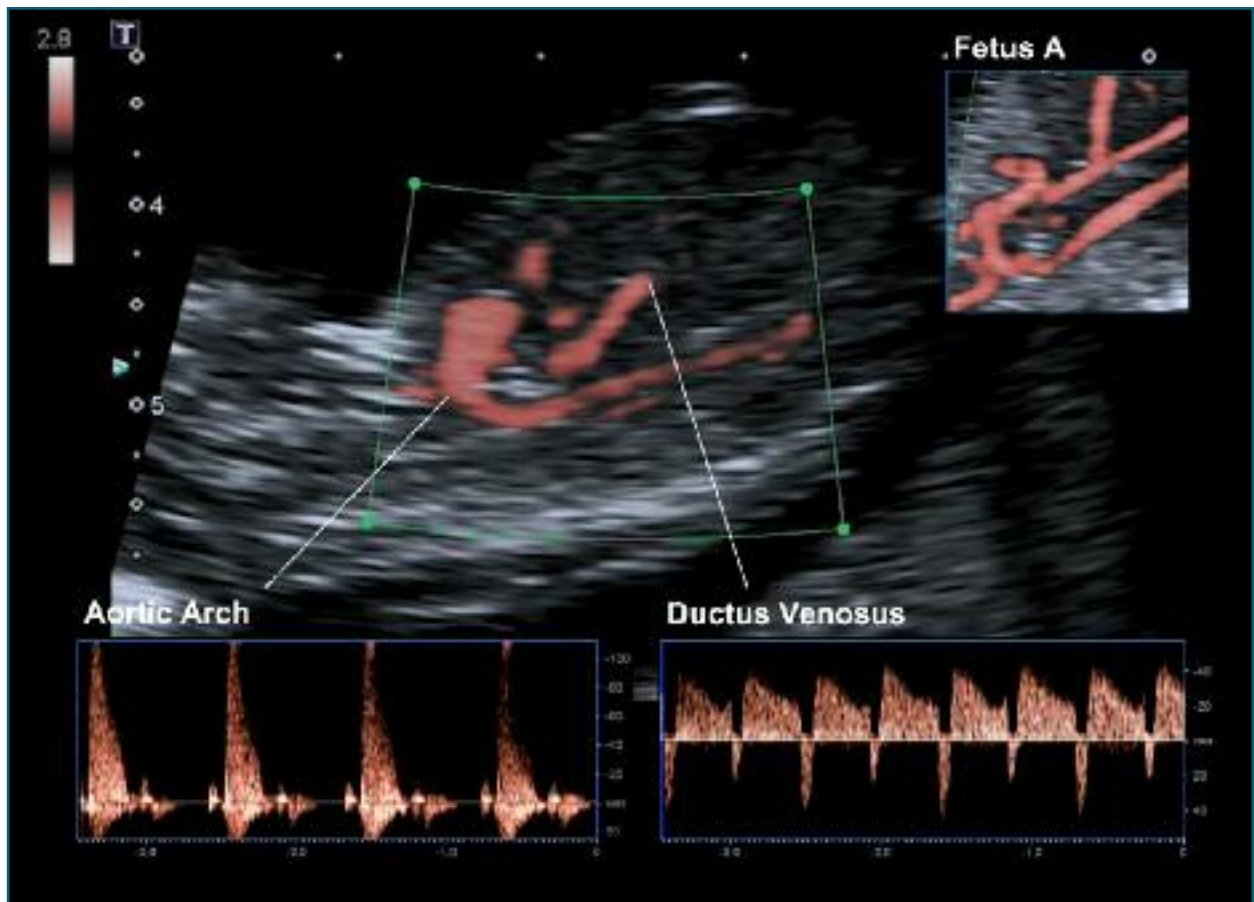
**Palavras-chave:** Isomerismo atrial esquerdo; Isomerismo esquerdo; Ecografia fetal; Morfologia precoce.

Left atrial isomerism (LAI) is a rare syndrome (1 in 10,000-40,000 live births)<sup>1</sup>, that is characterized by the symmetric left-sided aspect of the naturally asymmetric thoracoabdominal organs, as a result of left-right axis disruption. This disorder incorporates a group of cardiac, vascular and visceral anomalies. Multiple combinations are possible and thus prenatal diagnosis can be challenging. The most common findings of this syndrome include several cardiac anomalies (83%) with the reliable finding of two morphological left atrial appendages. Additionally, this syndrome may present conge-

nital heart block (29%), interruption of inferior vena cava with azygos continuation (89%), mispositioned stomach (59%), polysplenia (57%) and midline liver (33%)<sup>2</sup>. Some post-natal diagnostic criteria are not useful *in utero* since visceral-situs is somewhat inconsistent and difficult to screen, especially the number of spleens or hepatic position. Hence cardiac and vascular anomalies are the most common fetal diagnostic pointers<sup>3</sup>. In the first trimester, the presence of increased nuchal translucency and bradycardia should raise suspicion of an underlying cardiac anomaly, including those associated with LAI<sup>4</sup>. In one series of 41 cases, the mean gestational age of diagnosis was 18 weeks (range 12-29)<sup>5</sup>. However, up to the best of our knowledge, no illustra-

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**FIGURE 1.** SMI image showing the aortic arch plane, with azygos parallel to the descending aorta, and interruption of the inferior vena cava (not identified). The two insets at the bottom demonstrate the discordant atrial and ventricular rates, reported by ductus venosus (136bpm) and aortic (68bpm) flow rates, respectively. The right-upper inset demonstrates the normal aspect of the aortic arch from fetus A.

tive reports of such an early diagnosis have been published. The prognosis depends mainly, but not exclusively, on cardinal cardiac defects, with the heart block increasing the risk of fetal or perinatal demise<sup>3</sup>. The global prognosis reported in one large meta-analysis is poor, with 25% of termination of pregnancies, 7% intrauterine and 17% post-natal deaths<sup>2</sup>. The need of surgery was reported in 73% of cases, with a 27% overall death rate during or after the procedure.

Here, we report a case of a thirty-year-old woman with a spontaneous dichorionic/diamniotic pregnancy (3G1P). Uneventful medical or family history, and no consanguinity was reported. This case was sent to our prenatal diagnosis center due to the suspicion of cardiopathy (atrioventricular septal defect and atrioventricular block), associated nuchal translucency above

the 99<sup>th</sup> centile and jugular lymphatic sacs, in one of the fetuses. At 12<sup>3/7</sup> weeks, the ultrasound (Toshiba® Aplio 400) re-evaluation of fetus A was unremarkable, but fetus B revealed a persistent heart rate below 80 bpm. A thorough evaluation exposed several anomalies, including an atrioventricular septal defect with apparent 2 left atrial appendages, “double vessel” sign, right-sided stomach, discrepancy between the heart rate of the ductus venosus and the aortic arch (Figure 1), and interruption of inferior vena cava with azygos continuation, which was recorded with superb microvascular imaging (SMI) technology (Figure 2). After being elucidated of the meaning of these findings and their prognostic impact, the couple requested selective termination of pregnancy, which was accepted by the institutional board. Concerning the fetus A, the pregnancy



**FIGURE 2.** SMI mode showing bi-caval view with interruption of inferior vena cava (IVC), azygos vein (AzV) drainage to the superior vena cava (SVC) and digitiform appearance of the atrial appendage, suggesting a left atrial appendage (LAA) in the right atria (RA). SMI technology also allowed the identification of the left hepatic vein (LHV), the ductus venosus (DV) and the umbilical vein (UV).

occurred unremarkably resulting in a successful full-term birth (3350 g, Apgar score 9/10).

The first trimester fetal bradycardia was crucial to guide the investigation for other LAI-related anomalies. This case highlights the importance of a thorough first trimester morphologic evaluation, especially in twins, where early management of discordant congenital defects benefits perinatal outcomes.

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## INFORMED CONSENT

Patient consent for publication was obtained.

## DECLARATIONS OF INTEREST

The authors have no conflicts of interest to declare.

## AUTHOR CONTRIBUTIONS

All authors made a substantial contribution to the information or material submitted for publication.

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