Prenatal diagnosis of left isomerism with normal heart

Sara Costa,[1] Ana Carriço,[2] Francisco Valente[3]

[1]Department of Gynecology–Obstetrics, Centro Hospitalar Universitário do Algarve—Unidade de Portimão, Portugal
[2]Department of Pediatric Cardiology, Centro Hospitalar de Vila Nova de Gaia/Espinho, EPE, Portugal
[3]Department of Gynecology–Obstetrics, Centro Hospitalar de Vila Nova de Gaia/Espinho, EPE, Portugal

Correspondence to
Dr Sara Costa,
saraibcosta@gmail.com

Accepted 27 October 2017

DESCRIPTION

Left isomerism (LI) is characterised by loss of normal asymmetry of the thoracoabdominal organs. Consequently, viscera that normally develop on the left side are found on both sides of the body, whereas right-sided viscera may be absent. This abnormal symmetrical development of organs occurs because in the early embryonic life, there was a disruption of left–right axis orientation.

The most common reported anomalies are bilateral morphological left atria and lungs, cardiac defects, arrhythmias (because the sinus node, located in the right atrium, is absent or incompletely developed), interruption of the inferior vena cava (IVC), multiple spleens and gastrointestinal malformations such as right-sided stomach, midline liver, biliary tract abnormalities and intestinal malrotation.

This rare condition occurs in approximately 1 per 10 000–40 000 live births.[1] The outcome of the fetuses with LI varies depending on the association with cardiac defects, dysrhythmias and other significant malformations.

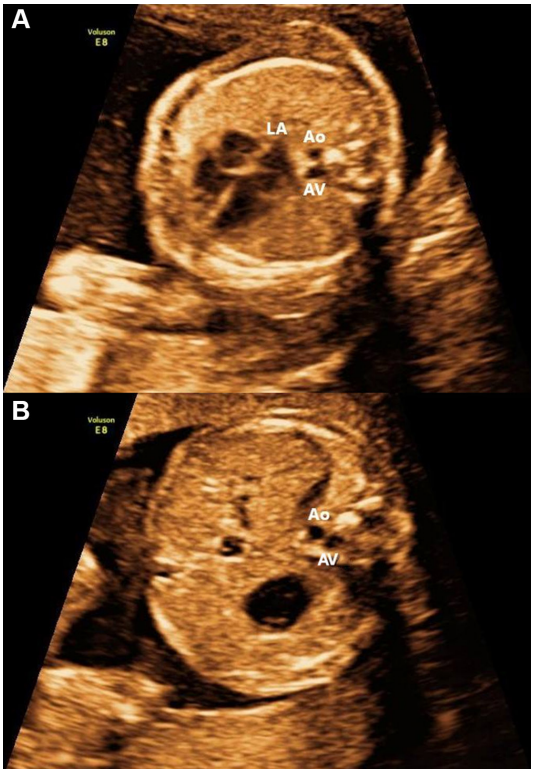

Figure 1 (A) Transverse section of the thorax showing a normal heart and behind the LA, it is possible to see the Ao and AV ('double vessel' sign). (B) Transverse section of the abdomen showing stomach on the left side, absence of the upper part of the inferior vena cava and AV located posterior and lateral to the Ao. Ao, aorta; AV, azygos vein; LA, left atrium.

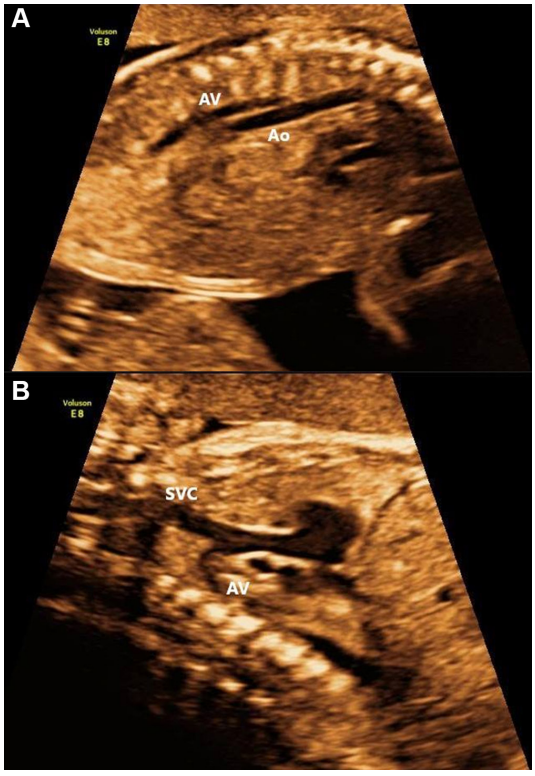

Figure 2 (A) Sagittal view of the fetus showing AV running posterior to the Ao. (B) Sagittal view of the fetus showing interruption of inferior vena cava with AV continuation to the SVC. Ao, aorta; AV, azygos vein; SVC, superior vena cava.

The prenatal diagnosis is usually made by fetal echocardiography. However, about 3%–18% of LI cases demonstrated normal cardiac anatomy.[2] In these situations, the prenatal diagnosis is even more challenging and may be missed.

Traditionally in fetal echocardiography, the situs is confirmed from the anatomical correlations of the great vessels in abdomen. Undergoing normal development, the aorta is located posterior and at the left of the spine, whereas IVC is more anterior and at the right of the spine. In about 80% of patients with LI, the IVC is interrupted in its intrahepatic part, and blood flows via azygos vein into the superior vena cava (SVC) and then into the atrium.[3] Therefore, discontinuity of the IVC with azygos continuation represents an excellent marker of LI. In this condition, during the fetal echocardiography, it is possible to identify the absence of the

To cite: Costa S, Carriço A, Valente F. *BMJ Case Rep* Published Online First: [*please include* Day Month Year]. doi:10.1136/bcr-2017-223097

Learning points

- In left isomerism (LI), instead of distinct left and right sides, there are two left sides resulting in bilateral morphological left atria; in these cases, cardiac defects, dysrhythmias, systemic venous abnormalities, such as interruption of the inferior vena cava, polysplenia and gastrointestinal anomalies are common.
- During fetal echocardiography, it is mandatory to confirm the correct arrangement of great vessels in the abdomen to exclude situs abnormalities such as LI.
- When there is a prenatal suspicion of LI, it is obligatory to perform repeated ultrasound examinations in order to detect dysrhythmias and other anomalies, including if the cardiac anatomy is apparently normal.

IVC, the aorta located medial to the spine and the azygos vein posterior and lateral to the aorta.

We report a case of a 32-year-old healthy pregnant woman referred to our Prenatal Diagnosis Center at 22 weeks of gestation for a fetal echocardiography due to nuchal translucency measurement above the 99th percentile in first trimester. For the same reason, chorionic villus sampling was performed with a normal result for fetal karyotype and microarray analysis. An ultrasound at 20 weeks of gestation found no fetal anomalies.

The fetal echocardiography showed a normal heart but an interruption of the IVC with azygos vein continuation to the SVC (figures 1 and 2). No other abnormalities were found, including heart rhythm disorders.

After this finding, we performed repeated ultrasonographic examinations at two weekly intervals until 30 weeks of gestational age and then once a week until delivery to monitor the heart rhythm and exclude other anomalies. No arrhythmia was detected, but we identified duplicated pyelocaliceal system on the right side.

The patient was delivered at 34 weeks by spontaneous vaginal delivery. After birth, the child underwent an extensive clinical evaluation. All prenatal ultrasound findings were confirmed, and additionally, a multilobulated spleen was detected. We excluded other non-cardiac anomalies such as intestinal malrotation and immune dysfunction. Currently, the child is developing well but maintains surveillance in paediatrics department.

Contributors SC: planning, conducting and reporting of the work. AC: diagnosis and review of the work. FV: diagnosis and review of the work.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

1 Evans WN, Acherman RJ, Restrepo H. Heterotaxy in southern Nevada: prenatal detection and epidemiology. *Pediatr Cardiol* 2015;36:930–4.
2 Berg C, Geipel A, Kohl T, *et al*. Fetal echocardiographic evaluation of atrial morphology and the prediction of laterality in cases of heterotaxy syndromes. *Ultrasound Obstet Gynecol* 2005;26:538–45.
3 Sharma S, Devine W, Anderson RH, *et al*. Identification and analysis of left atrial isomerism. *Am J Cardiol* 1987;60:1157–60.