

Transposition of the great arteries at first-trimestre scan

G. Soro, A. Guarque, B. Muñoz, R. Colell

Introduction

Congenital heart diseases represent the most common severe congenital malformations and only 14.1% are diagnosed before 25 weeks of gestation.

Case report

A 39-year-old primigravida without risk factors attended a first-trimester ultrasound at 13 weeks of gestation. The evaluation showed: horizontalization of the cardiac axis with normal four-chamber view, with equal-sized ventricles, position and size.(Figure A)

Color Doppler imaging revealed a ventricular septal defect (VSD). (Figure B) Adequate visualization of the aortic and pulmonary outflow tracts was not correctly achieved in a transverse view.



Figure A

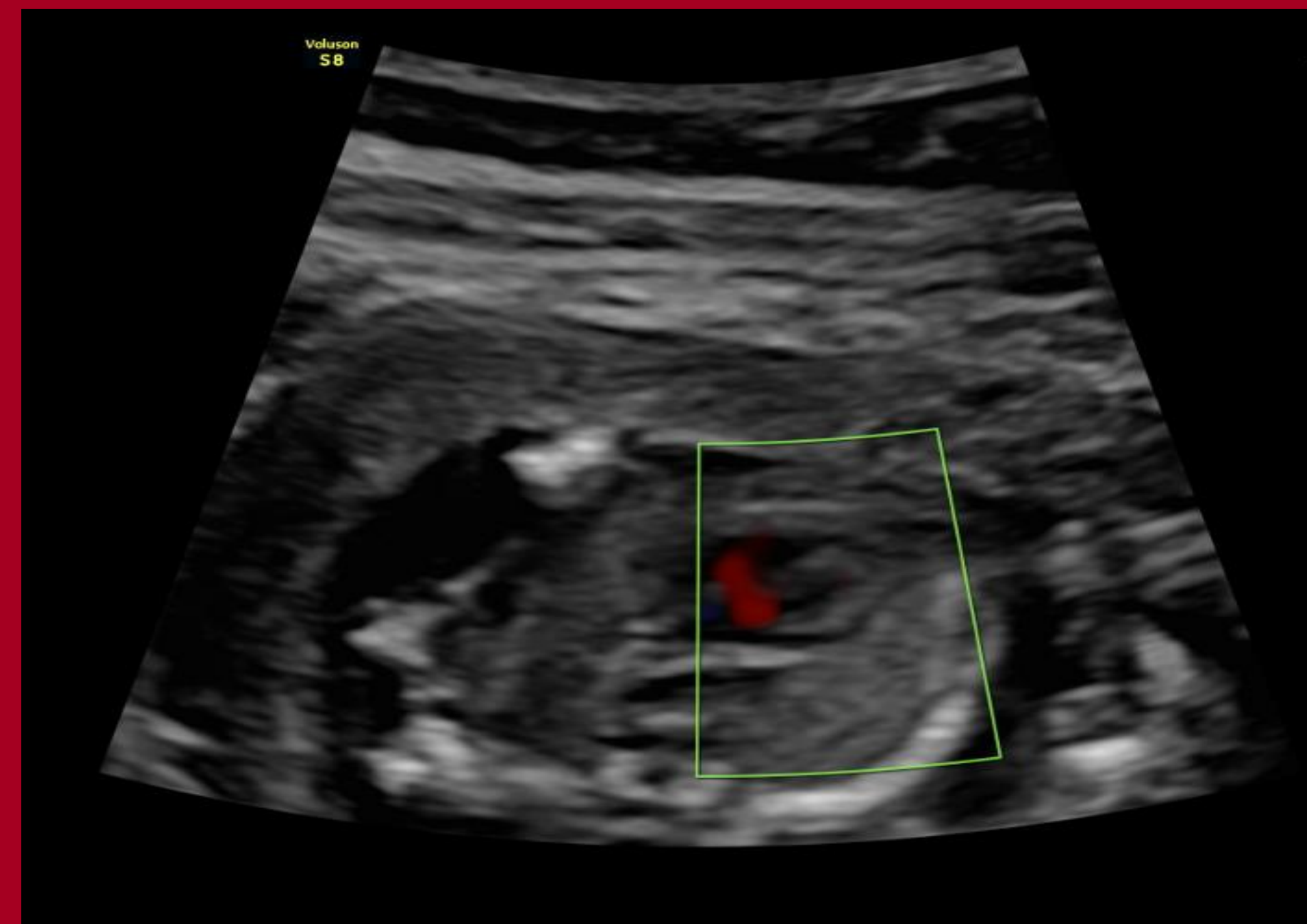


Figure B

Longitudinal views demonstrated parallel alignment of the great vessels. Aorta, with supraaortic branches, arose from the right ventricle, while the pulmonary artery, with its bifurcation, originated from the left ventricle, confirming the diagnosis of transposition of the great vessels associated with VSD. (Figure C and D)

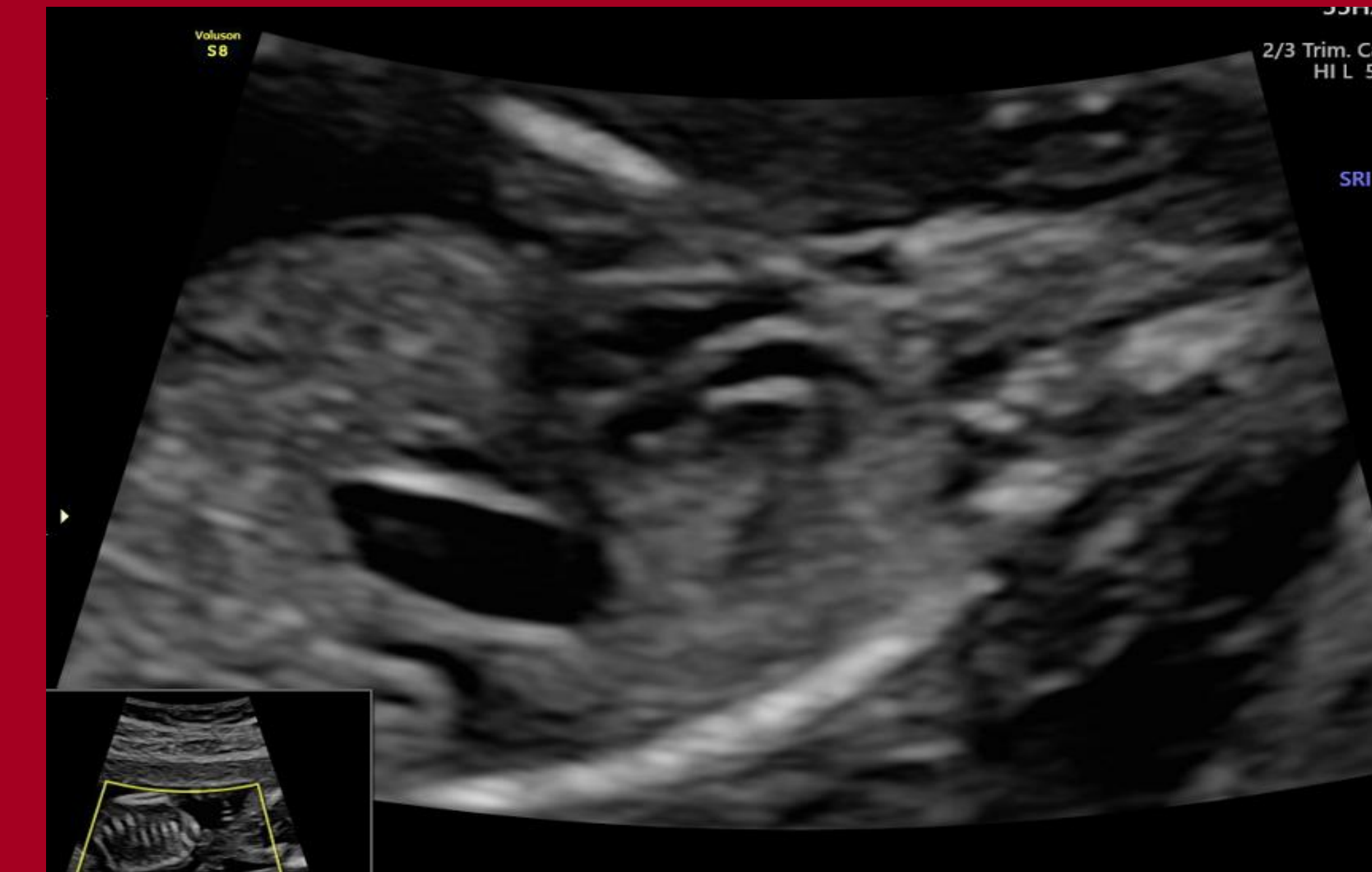


Figure C

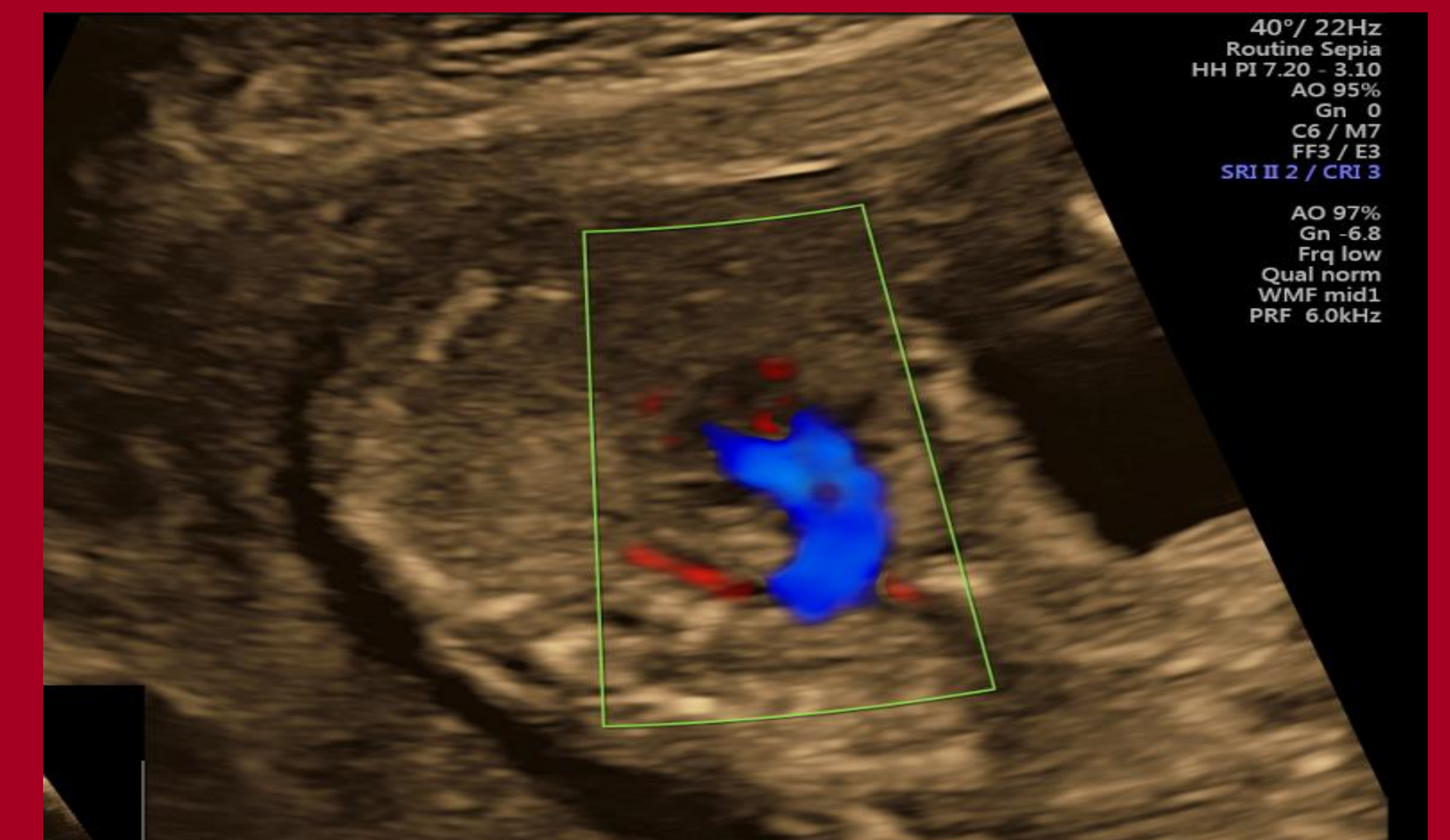


Figure D

Quantitative Fluorescence Polymerase Chain Reaction(QF-PCR), and Comparative Genome Hybridization (CGH) array were normal for the affected fetus. An early fetal echocardiography at 15+5 weeks confirmed the diagnosis. Parents opted for legal termination of pregnancy at 17 weeks, anatomopathological exam confirmed ecocardiography findings.

Conclusion

First-trimester ultrasound should be considered the initial step in the diagnosis of morphological abnormalities, including congenital heart diseases. With appropriate ultrasound equipment and adequate training, it should serve not only as a “genetic” scan but also as a “morphological” assessment without limitations based on fetal size.