

Heterotaxy syndrome: right atrial isomerism

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Introduction

Heterotaxy is defined as an abnormality where the internal thoraco-abdominal organs demonstrate abnormal arrangement across the left-right axis of the body. Right atrial isomerism is typically associated with complex cardiovascular malformations.

Case report

A 21-year-old primigravida with consanguineous marriage showed at the first trimester scan an abnormal four-chamber view and discrepancy in the position of the stomach and heart. (Figure A, B) Invasive test was offered but refused by parents.



Figure A



Figure B

At 16 weeks echocardiography was performed: heart in the left chest, four chamber view showed significant disproportion, a wide interauricular and interventricular communication, large right-side morphologic right ventricle with pulmonary artery but neither mitral nor aortic flow was observed.(Figure C, D, E)

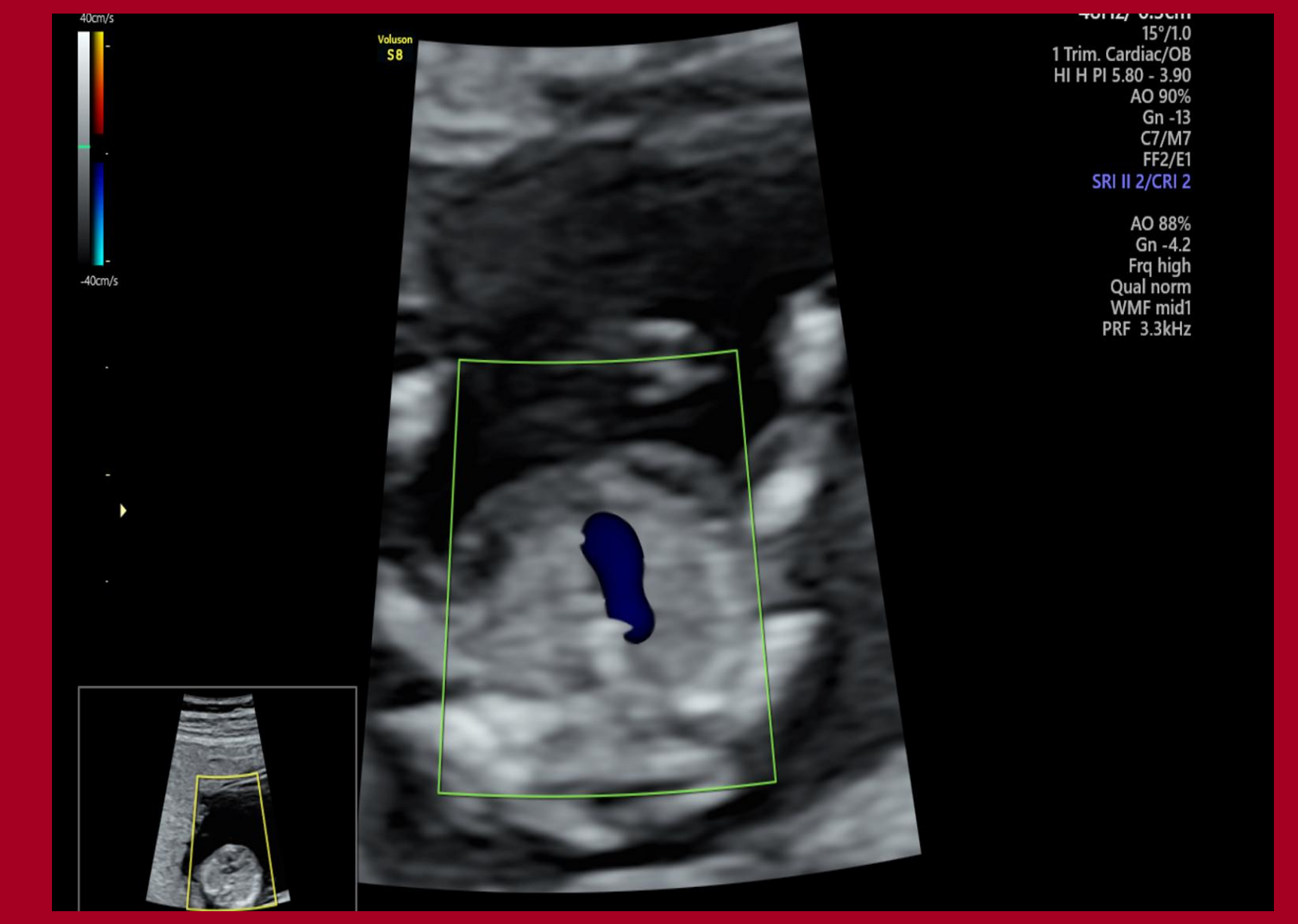
Figure C



Figure D



Figure E



After discussing the prognosis, opted for termination of pregnancy. Necropsy showed: heart in *situs solitus*, pulmonary enlarged artery, absence of aortic arch and ascending aorta. Marked hypoplasia of the left-sided chambers. Coronary arteries in normal position. The right atrium demonstrates normal anatomical features (pectinate muscles, foramen ovale, and septum secundum) with the presence of a large atrial septal defect. Tricuspid valve was identified. Examination of the interventricular septum showed the presence of a septal defect. The left atrium reveals marked hypoplasia, with absence of connection to the mitral valve, consistent with mitral atresia. The left ventricular outflow tract connecting to the aorta was hypoplastic. Liver was located on the left side, and the stomach showed malrotation toward the right side. Placental karyotype was accepted and the result was 46XX but no Comparative Genome Hybridization(CGH) array was performed.