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Clinical presentation heterogeneity in four cases with left atrial isomerism

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Introduction

Heterotaxy syndrome is a rare, complex disorder involving structural cardiac defects in combination with symmetrical development of abdominal viscera that are normally asymmetrical organs. The vast assortment of cardiac manifestations with accompanying abdominal visceral variations determines the prognosis of heterotaxy syndrome. In this case study, we aimed to present the prenatal evaluation and clinical variations of four cases diagnosed with left atrial isomerism who were referred to the Perinatology Clinic of Karadeniz Technical University, Farabi Hospital, between 2024 and 2026.

Case 1. A 29-year-old G1P0 pregnant woman, currently at 22 weeks of gestation, was referred to our center with suspected fetal cardiac anomaly. The patient's medical history was unremarkable. Detailed prenatal ultrasonography revealed a complex constellation of findings consistent with situs ambiguus, characterized by the stomach being located on the right side and dextrocardia. The liver was positioned in the left hemiabdomen, while the gallbladder was located in its normal anatomical position. The spleen could not be visualized within the splenic fossa. The four-chamber (4CH) view showed distortion, with the diameter of the left ventricle significantly smaller (hypoplastic) than that of the right ventricle. A minor muscular defect was also detected. The atrial appendages exhibited features consistent with left isomerism. Both the aortic arch and the pulmonary artery originated from the right ventricle, indicating a double-outlet right ventricle with a relatively hypoplastic left ventricle. The blood vessels arose in parallel, and a minor muscular ventricular septal defect (VSD) was noted. The pulmonary artery was also hypoplastic, and an atrial septal defect (ASD) was identified in the 4CH view. A characteristic double aortic arch was observed, with the aortic branches arising from the right arch. There was a persistent left superior vena cava and a confluent vein behind the heart, indicating abnormal pulmonary venous return.

Case 2. A 37-year-old G2P1pregnant woman, currently at 17 weeks of gestation, was referred for suspected fetal cardiac anomaly. The patient's medical history was unremarkable. Ultrasonographic evaluation revealed the stomach in its normal anatomical position on the left, and the gallbladder was on the right. The intestines appeared echogenic. Cardiac assessment demonstrated a 4CH view with an echogenic intracardiac focus (EIF). The morphological right ventricle was left-sided and hypoplastic, while the morphological left ventricle was positioned anteriorly (to the right) and was also hypoplastic. The aorta was located on the left, originating from the morphological right ventricle, and the pulmonary artery was on the right, originating from the morphological left ventricle. Furthermore, both atrial appendages exhibited features consistent with left atrial isomerism. The primary diagnosis was considered left atrial isomerism and congenitally corrected transposition of the great arteries (ccTGA). The three-vessel trachea (3VT) view revealed a right aortic arch with a left ductus arteriosus. The fetal heart rhythm was bradycardic (75 bpm) with confirmed atrioventricular (A-V) block. Additionally, a reversed a-wave was observed in the ductus venosus.

Case 3. A 40-year-old G4P3 pregnant woman was referred to our perinatology department at 13 weeks of gestation with a preliminary diagnosis of cystic hygroma. The patient's obstetric history was significant for active smoking and parental consanguinity. Initial first-trimester screening revealed a significantly increased nuchal translucency (NT) of 11 mm, a single umbilical artery (SUA), and an atrioventricular septal defect (AVSD). Additionally, Doppler assessment showed a reversed a-wave in the ductus venosus. During the follow-up of the patient, ultrasonographic evaluations revealed a small stomach displaced to the left posterior position, and the heart was in a mesocardial position. The gallbladder was not visualized. In addition to an unbalanced AVSD, both atrial appendages exhibited morphological features of a left atrial appendage, consistent with left isomerism. A single common arterial trunk was observed originating predominantly from the right ventricle. Furthermore, a dilated coronary sinus associated with a vertical vein was identified, raising high suspicion for Total Anomalous Pulmonary Venous Connection(TAPVC). No additional extracardiac anomalies were detected during the detailed anatomical survey.

Case 4. A 34-year-old G4P1 pregnant woman, currently at 14 weeks of gestation, was referred to our perinatology department for further evaluation due to increased NT, a reversed a-wave in the DV, and a right-sided stomach. The patient's medical history was notable for maternal obesity, Type 2 diabetes mellitus, and recurrent pregnancy loss. Initial ultrasonographic assessment revealed an NT measurement of 3.2 mm, a reversed a-wave in the DV, mesocardia, and a right-sided stomach. Furthermore, both atria exhibited morphological characteristics of the left atrial appendage, suggesting left atrial isomerism. Follow-up ultrasonographic evaluations demonstrated that the 4CH view showed distortion, with the diameter of the left ventricle significantly smaller (hypoplastic) than that of the right ventricle. In the upper abdominal cross-section, the stomach was again visualized on the right side, and a dilated azygos vein (double vessel sign) was observed to the right of the aorta. The gallbladder and spleen could not be visualized due to the early gestational age and maternal obesity.

Conclusion

As demonstrated in the four cases presented above, left atrial isomerism exhibits considerable variation in ultrasonographic findings. A consistent ultrasonographic feature across all cases is the presence of bilateral atrial appendages that morphologically resemble the left atrial appendage (LAA), characterized by its narrow junction with the left atrium and a distinct finger-like, hooked appearance. Notably, conotruncal anomalies and septal defects were present in three out of the four cases; additionally, two of the four cases presented with anomalous pulmonary venous return. Furthermore, complete atrioventricular (A-V) block-a common finding in left atrial isomerism-was prominently observed in one case. While prenatal evaluation of the spleen is technically challenging, it was successfully visualized in the first three cases; however, it could not be identified in the final case due to the early gestational age and maternal obesity. Furthermore, genetic investigations for all cases yielded a normal karyotype, and chromosomal microarray analysis (CMA) results for cases 1 and 2 were also reported as normal. Despite the generally more favorable prognosis of left atrial isomerism compared to right atrial isomerism, the families in Cases 1 and 2 opted for the termination of pregnancy due to the severity of the associated major cardiac anomalies. The pregnancies for cases 3 and 4 are currently ongoing. In conclusion, the assessment of fetal situs and the normal anatomical positioning of organs can provide critical clues regarding the presence of severe fetal anomalies.

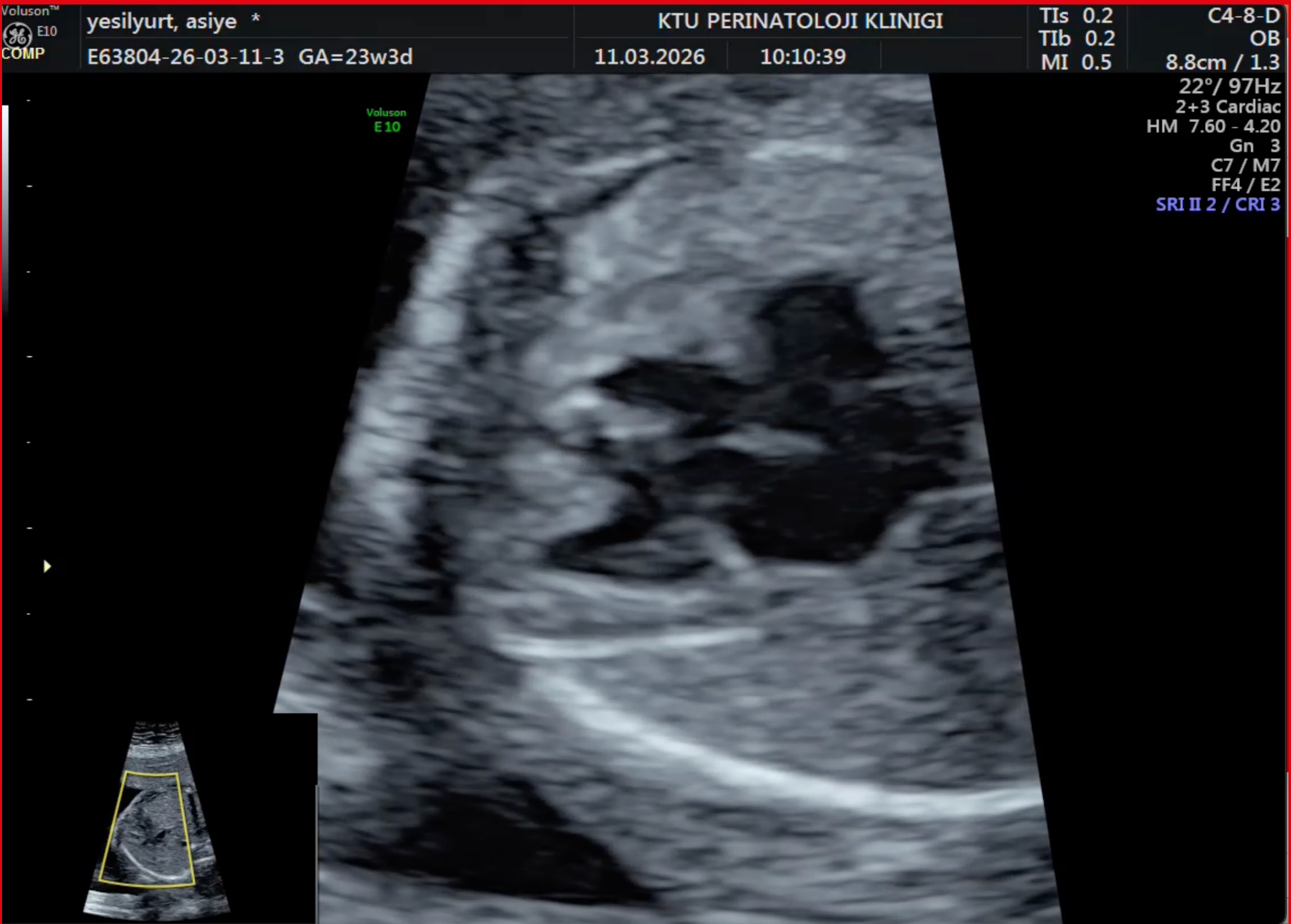


Figure 1. Case 3 AVSD and TAPVC



Figure 2. Case 1 Double aortic arch