Univentricular atrio-ventricular connections: Understanding the prenatal presentations

The Fetal clinic, Pondicherry, India

Objective
Univentricular AV connection defect represents a heterogenous group characterized by the common morphological feature of atrial chamber(s) connecting to a single ventricular chamber. The significance lies in the fact that functional biventricular repair is unlikely in the vast majority. Understanding the complete prenatal spectrum will help in the correct identification of this disorder. We aimed to elucidate the prenatal ultrasound characteristics of this group of cardiac defects.

Methods
This was a retrospective cross-sectional study conducted in a single tertiary level fetal medicine centre over a period of 5 years from January 2017 to January 2022, with gestational age ranging from 12-36 weeks at The Fetal Clinic, Pondicherry. Defects that demonstrated the following patterns were classified as univentricular AV connection defects and were included in the study: a. Atriaresia of right or left AV valve; b. both atria draining into a dominant ventricle; c. hearts with a single ventricle. Data regarding maternal demographic aspects, cardiac segmental analysis, extra cardiac defects were retrieved from the fetal database and review of the images; perinatal outcome data was obtained through telephone follow up from the patients. Cardiac segmental analysis data was recorded as follows. a. Atrial anatomy: Normal atria were documented when both right and left atrium were seen and when interatrial septum was visible. Common atrium was documented when interatrial septum was not visible. b. Atrial arrangement: Based on the atrial and visceral arrangements, the fetuses were categorized into Situs solitus, Situs Ambiguous (isomerism) and situs inversus groups. c. AV Junction arrangement: AV junction was classified as either normal (when both AV valves were patent) or Left / Right AV valve atresia or as common AV valve. d. Common AV valve orifice was classified as one or two depending on the colour flow pattern. e. Ventricle morphology was classified as single ventricle when only one ventricle chamber was visible; or as dominant right ventricle or dominant left ventricle if an additional outlet (rudimentary) chamber was present that was connected to the dominant ventricle through a bulboventricular foramen. f. Outflow tracts were classified as bichambered if each of the outflow tracts arise from different chambers or unichambered if both outflow tracts arise from single chamber.

Results
Results: Over a period of 5 years 57 fetuses were diagnosed with univentricular AV connection defects. The mean gestational age at diagnosis by ultrasound was 21 weeks (12-36 weeks). Of these 39 (68%) had situs solitus, 18(32%) had situs ambiguous. There were no cases of situs inversus. Among the situs solitus group (n = 39), normal atria were noted in 35 fetuses (90%), Unilateral AV Valve atresia in 22 fetuses (56%), Common AV valve in 10 cases and a single AV orifice in 31 fetuses (80%). Single RV was noted in 18 fetuses, Dominant LV in 12 fetuses, rudimentary outlet chamber was present in 18 fetuses and was absent in 21 fetuses. Unichambered outflow tract origin was seen in 20 fetuses, bichambered in 19. Aortic valve stenosis noted in 15 fetuses, pulmonary valve stenosis noted in 11. Truncus arteriosus seen in 2 fetuses. Among the situs ambiguous group (n = 18), common atrium was noted in 10 fetuses. Common AV valve in 15 cases (84%), Unilateral AV valve atresia in 3 fetuses (16%), a solitary ventricle was noted in 14 (77%) fetuses. Rudimentary outlet chamber was present in only 3 fetuses. Unichambered outflow tract origin was noted in 15 fetuses while 3 had bichambered origin; pulmonary valve lesion noted in 15 cases (84%), aortic valve lesion noted in one case, truncus arteriosus was noted in one case. Extra cardiac anomalies were noted in 16/1657, 28% cases. Most common extra cardiac abnormality was agenesis of corpus callosum followed by spine abnormality. Right sided aortic arch was noted in 7 cases. Out of 57 cases 6 were diagnosed in first trimester. Among 57 cases 46 (46/57, 81%) resulted in termination of pregnancy. 4 were livebirths. Among the 4 livebirths, 3 resulted in neonatal death, one awaiting surgery. Invasive testing was done in 5 cases, out of which 4 returned as abnormal KT – 2 were Turner syndrome, one Trisomy 18, one Trisomy 13 and one had normal karyotype. 10 fetuses were given for autopsy after termination and the prenatal findings were confirmed in all the cases.

Conclusion
Conclusion: Univentricular AV connection in the setting of situs inversus is extremely rare. In the setting of situs solitus the ventricular pattern is almost equally distributed between solitary ventricle and the functionally univentricular anatomy with additional outlet chamber. In the setting of atrial isomerism, it is predominantly common AV junction and solitary ventricle anatomy. Prenatal diagnosis is feasible across all gestations.