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Prenatal diagnosis of absent aortic valve with continuous mitral regurgitation in Turner's syndrome
Too many things gone awry

BACKGROUND

Congenitally absent aortic valve (AAoV) is a rare anomaly associated with a high incidence of perinatal mortality.

CASE

A female fetus of twin gestation underwent a fetal echocardiogram at 24 weeks gestation, following unexpected demise of the twin.

Findings:

- Rudimentary AoV leaflets with severe aortic insufficiency and aortic arch flow reversal.
- Left ventricular (LV) non-compaction with severe dysfunction.
- Continuous moderate regurgitation across a dysplastic, functionally atretic mitral valve.
- Borderline restrictive left to right flow through the foramen ovale (FO) was noted.

The presence of a compliant LV cavity and continuous MR appeared to be pivotal findings facilitating an inverse circular shunt.

Delivery occurred by emergent cesarian section at 27 weeks followed by death on day 8 of life.

Microarray revealed Turner's syndrome.

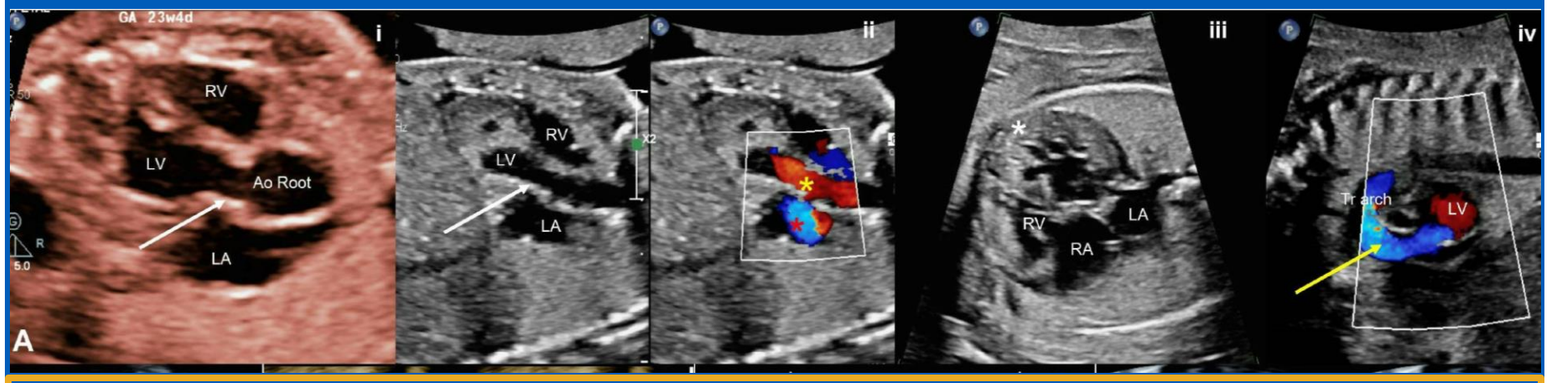

Image A- Fetal echocardiogram: i. Left ventricular outflow view reveals a mildly hypoplastic aortic valve annulus with rudimentary leaflets (white arrow). ii. Color-compare image of the same view showing aortic regurgitation in diastole with concomitant mitral regurgitation (yellow and red asterisks respectively). iii. LV noncompaction with endocardial fibroelastosis (asterisk). iv. Long axis view of the aortic arch with flow reversal across the transverse arch and ascending aorta (yellow arrow).

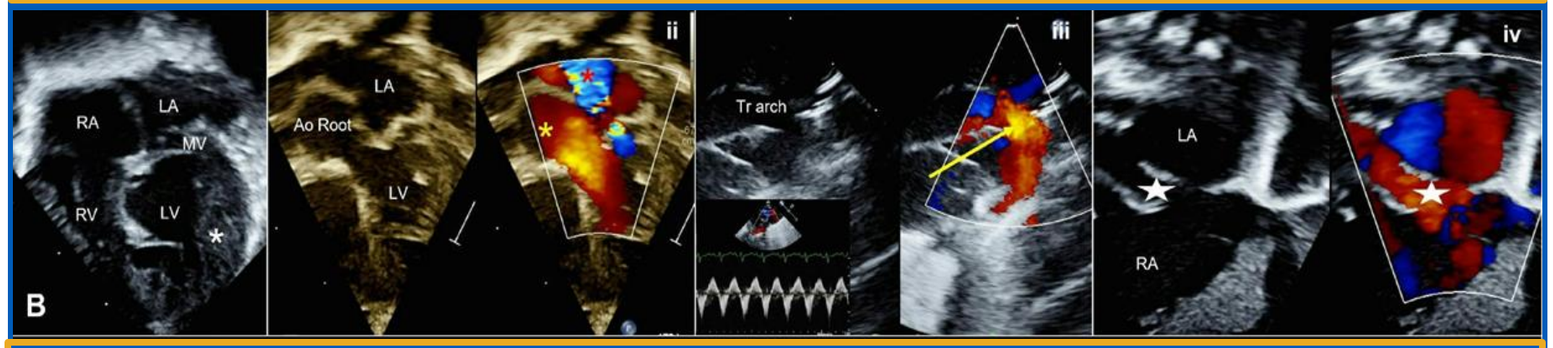

Image B- Postnatal transthoracic echocardiogram: i. Apical 4-chamber view showing dysplasia of MV and LV noncompaction. ii. Apical 5-chamber view showing severe aortic insufficiency with concomitant mitral regurgitation (yellow and red asterisks respectively). iii. Suprasternal view showing flow reversal across the arch and brachiocephalics contributing to the circular shunt (inset shows Doppler pattern across descending aorta with diastolic flow reversal and low-velocity forward flow in systole). iv. Subcostal sagittal view showing borderline restrictive left-to-right flow across foramen ovale (white star).

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DECISION MAKING

Postnatal intervention with Norwood procedure and oversewing of the AoV is typically an option.

In our case, the continuous mitral regurgitation with functional mitral atresia further mitigated the inverse shunt and cardiovascular collapse. Prematurity and preclusion from extracorporeal circulatory support candidacy due to severe aortic insufficiency limited therapy to vasoactive agents which proved ineffective resulting in the newborn's demise.

CONCLUSION

Fetal echocardiographic evaluation of hemodynamics in AAoV is essential.

Prematurity, an inverse circular shunt and chromosomal anomaly are bad prognostic factors that should be considered in parental counselling and postnatal management planning.

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