Isolated Double Chamber Right Ventricle in a newborn with Mosaic Genome-Wide Paternal Uniparental Isodisomy.

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Introduction

- A double-chambered right ventricle is an uncommon abnormality that represents 0.5 to 2% of all the congenital heart defects, and it has been infrequently reported as an isolated entity.
- This condition causes sub-infundibular right ventricular outflow tract obstruction, originated by either hypertrophic trabecular tissue, atypical muscle bands or an anomalous moderator band, that divides the right ventricle into a proximal high-pressure and distal low-pressure chamber.
- The clinical presentation can range from asymptomatic patients to development of hydrops fetalis or heart failure.
- There have been few cases describing a relationship with Noonan syndrome but to our knowledge it has not been reported in association with a uniparental isodisomy pattern.

Patient Presentation and Evaluation/Intervention

- We describe the case of a female born at 29 weeks of gestation due to preterm labor, to a mother who had a pregnancy complicated by anti-D isoimmunization that required cordocentesis and fetal transfusion, development of severe polyhydramnios treated with amniocentesis, and had a fetal echocardiogram at 26 weeks of gestation that demonstrated mild bi-ventricular hypertrophy.
- She initially required respiratory support with continuous positive airway pressure and was found to have a double-chambered right ventricle as an isolated defect, with severe hypertrophy and moderate-to-severe sub-pulmonary outflow tract obstruction.
- Beta-blocker therapy was initiated, and serial echocardiograms continued to demonstrate a stable but severe right ventricular outflow tract obstruction.

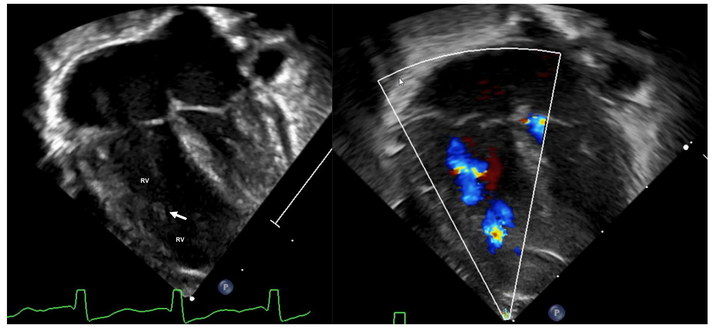

Figure 1. Transthoracic echocardiogram apical four chamber view, 2D (left) and color Doppler interrogation (right), that demonstrates the double-chambered right ventricle separated by anomalous muscular bundles (white arrow).

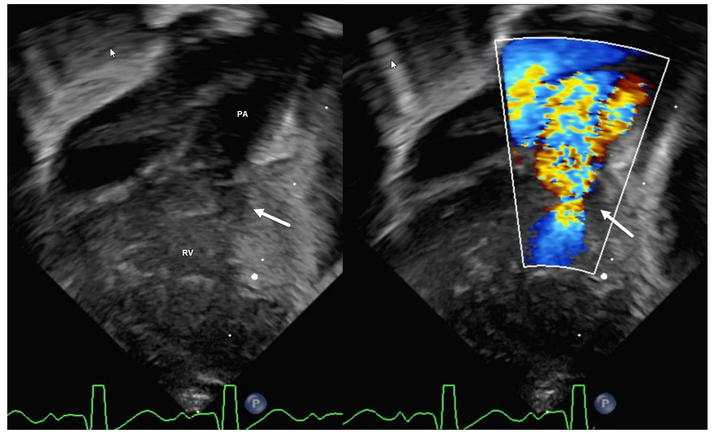

Figure 2. Transthoracic echocardiogram apical view of the right ventricular outflow tract, 2D (left) and color Doppler interrogation (right), that demonstrates the sub-pulmonary muscular right ventricular outflow obstruction (white arrow).

- Additionally, she had feeding difficulties with persistent hypoglycemia, and further investigation revealed a non-classical congenital adrenal hyperplasia.
- A single-nucleotide polymorphism analysis was performed, that showed an unusual B-allele frequency pattern suggestive of a mosaic, genome-wide paternal uniparental isodisomy.
- Ultimately, she underwent right ventricular muscle bundle resection at eleven weeks of age with excellent post-operative results with a widely patent right ventricular outflow tract.

Discussion

- Double-chambered right ventricle is a rare progressive condition that can have detrimental complications if left untreated.
- Most of the cases are associated with other congenital cardiac malformations, such as pulmonary valve stenosis, tetralogy of Fallot, Ebstein's anomaly, transpositions of the great arteries, or anomalous pulmonary drainage, but it is more commonly reported in association with membranous ventricular septal defects.
- As an isolated congenital heart defect, it becomes a diagnostic challenge, scenario in which further investigation could help as guidance to identify associated conditions, such as Noonan syndrome or as with our case uniparental isodisomy.

Conclusion

- Overall, it is important for clinicians to have a high index of suspicion of double-chambered right ventricle, particularly in the case of an isolated defect.
- Early diagnosis, identification of other associated congenital heart diseases and early surgical correction will result in favorable long-term outcomes.