Prenatal diagnosis of an uncommon form of a hypoplastic left heart syndrome variant.

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Title: Prenatal diagnosis of an uncommon form of a hypoplastic left heart syndrome variant.

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Background: Mitral valve dysplasia syndrome (MVDS) is a rare form of congenital heart disease, similar to hypoplastic left heart syndrome (HLHS). Prenatal identification is important for counseling, delivery planning and postnatal management.

Case: A 39 year-old woman underwent fetal echocardiography at ~34 weeks gestation for evaluation of fetal cardiomegaly and hydrops revealing: biatrial enlargement, mild-to-moderate mitral and tricuspid valve insufficiency, echobright mitral valve apparatus, biventricular dilation, severe LV fibroelastosis (EFE) and systolic dysfunction, mild hypoplasia of aortic valve annulus and aortic arch, and a thick restrictive atrial septum (RAS) with left to right flow (Fig 1a-c).

Decision-making: Although critical aortic stenosis was considered initially, MVDS seemed more likely given above characteristic findings. Delivery planning included elective C-section with standby catheterization laboratory and ECMO teams, given RAS and cardiac dysfunction. Patient was listed for transplant as biventricular or single ventricle repair were deemed unfeasible, given valvar insufficiency, LV dysfunction and EFE (Fig 1d-g).

Conclusion: MVDS is uncommon but should be considered with HLHS differential, in presence of a normal-dilated left atrium and ventricle, LV dysfunction and EFE, RAS and aortic valve and arch hypoplasia. Planned delivery and immediate postnatal atrial septostomy is warranted and cardiac transplantation is often the only therapeutic option.