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An Unusual Cardiomegaly Culprit: Case Report Of A Hemodynamically Significant Portosystemic Shunt

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An Unusual Cardiomegaly Culprit: Case Report of a Hemodynamically Significant Intrahepatic Portosystemic Shunt

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Clinical Course

Fetal: Diffuse cardiomegaly with preserved biventricular systolic function; no hydrops

→ **Postnatal:** Persistent mild cardiomegaly with structurally normal heart; normal systolic function

→ **14 mo:** Previous findings with new mild septal hypertrophy; asymptomatic; diagnosed with presumed cardiomyopathy (FH negative)

7 yr: Persistent diffuse moderate biatrial and biventricular dilation; normal systolic/diastolic function; asymptomatic

→ **9 yr:** New finding of hepatic vein dilation; normal RV pressures; normal function and SaO2; abdominal CTA and cardiac MRI obtained

→ **Post surgical IHPSS ligation:** Improved ventricular and atrial sizes

Genetic testing at 9 years: NOTCH1 mutation; VUS in TTN gene; microarray negative

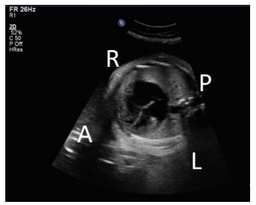

Fig. 2. Fetal echocardiogram (A=anterior, P=posterior, L=left, R=right).

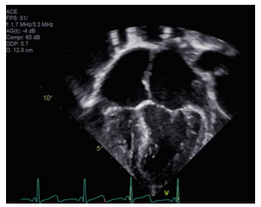

Fig. 3. Echocardiogram at 5 years with biatrial and biventricular dilation.

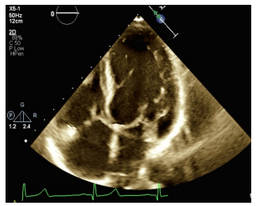

Fig. 4. Post shunt ligation echocardiogram with improved left ventricular and atrial dilation.

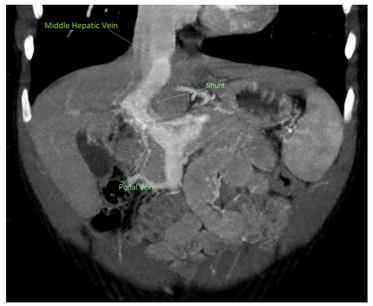

Fig. 1. Abdominal CT angiogram demonstrating intrahepatic portosystemic shunt (IHPSS) between the portal vein and middle hepatic vein.

Cardiac MRI findings:
Mild dilation of the left atrium and right ventricle. Left ventricle dilated with mild concentric hypertrophy.
LV EDV: 130.5 mL/m2 (87-128); LV mass: 101.1 g/m2 (57-88); RV EDV: 115.8 mL/m2 (93-146).
No late gadolinium enhancement seen.

References

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2. Alsamri, M.T., Hamdan, M.A., Sulaiman, M. *et al.* Hypoxia due to intrapulmonary vascular dilatation in a toddler with a congenital portacaval shunt: case report. *BMC Pulm Med* **19**, 49 (2019). https://doi.org/10.1186/s12890-019-0788-8

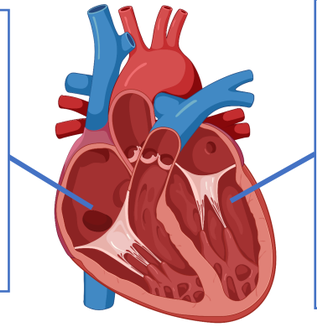
Thank you to Dr. Sherwin Chan (Children's Mercy Hospital) for assistance with radiographic imaging interpretation.

Take-home Points

- Multiple etiologies of unexplained cardiomegaly in a structurally normal heart: Cardiomyopathies, extracardiac shunt lesions, vascular malformations
- Pattern of dilation and systolic/diastolic function can help in narrowing a differential diagnosis.
- Consider evaluation for alternative diagnoses if clinical course and evaluation not consistent with cardiomyopathy.
- Hepatic vein dilation on fetal or postnatal echocardiogram should prompt investigation for portal shunts.
- Prenatal detection may improve early management and prognosis – can be associated with fetal hydrops or pleural effusion
- Outcomes dependent on presence of structural, genetic, other anomalies
- Ligation is often curative.

Keys to Clinical Application

Possible clinical signs/symptoms: Cyanosis, activity intolerance, poor weight gain, fatigue, chronic hypoxemia.



Possible echocardiographic findings: Diffuse cardiomegaly, +/-valvular dysfunction, hepatic vein dilation, elevated RV pressure, positive bubble study if pulmonary AVM present.

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