Patent Ductus Arteriosus Stent to Create Reverse Potts Shunt Physiology in an Infant with Heritable Pulmonary Hypertension

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Background

- Heritable Pulmonary Arterial Hypertension (HPAH) is a rare, progressive condition that is clinically indistinguishable from Idiopathic Pulmonary Hypertension (IPAH) and includes patients that have an identified genetic cause of PAH [1,5].
- HPAH has poor prognosis despite advances in medical therapy [1,5].
- Surgical reverse Potts shunt creation has been described in children with severe HPAH as an alternative to lung transplantation [6].
- Patent ductus arteriosus (PDA) stent placement to create a functional reverse Potts shunt has been reported in children and infants with PAH associated with congenital heart disease as well as IPAH as young as 3.5 months of age [2,3,6].
- PDA stenting for functional reverse Potts shunt creation has not been performed in young infants with HPAH.

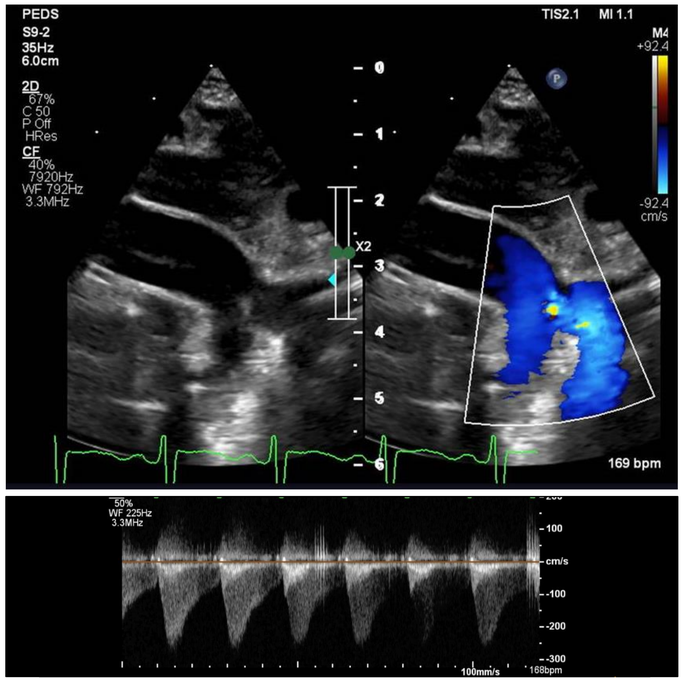

Figure 1: Echocardiogram with doppler at 1 month of life demonstrating all left to right shunting across the PDA

Case Presentation

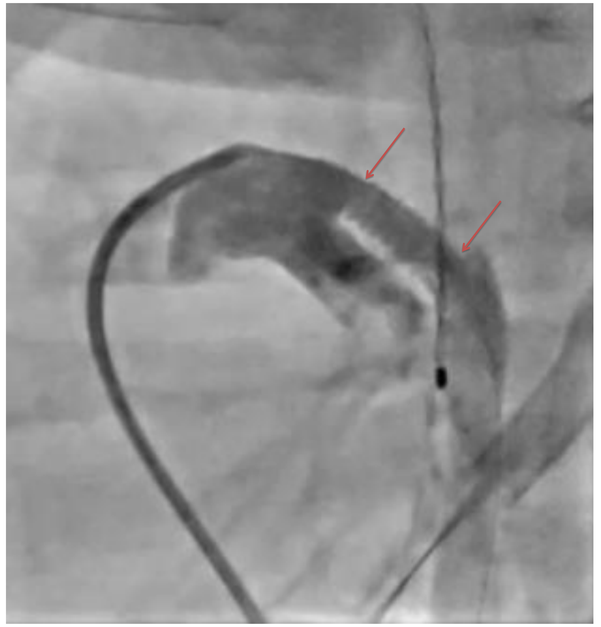

Figure 2: Angiogram of the PDA stent in the lateral view shows contrast filling stent, descending aorta and pulmonary artery with arrows indicating the two ends of the stent.

- Full-term neonate developed cyanosis shortly after birth
- Echo: severe PAH with no structural CHD
- At 1 month of age: Cardiac catheterization with suprasystemic RV pressures. PVRi of 28 U*m^2 consistent with severe PAH. PGE was initiated.
- Patient had persistent suprasystemic RV pressures despite multi-drug therapy for PAH (iNO, Sildenafil, Treprostinil, Bosentan)
- At 2 months of age: transcatheter PDA stent placed to create reverse Potts shunt physiology
- After stent placement patient weaned off iNO and respiratory support
- Patient was discharged from NICU at 6 months of life and has been thriving at home
- Whole genome sequencing (WGS): De novo missense variant in SOX17

REPORT CATEGORY	GENE	VARIANT	CONDITION	ZYGOSITY (INHERITANCE)	VARIANT CLASSIFICATION
VARIANTS RELATED TO PATIENT PHENOTYPE	SOX17	c.208C>G p.Arg70Gly	SOX17- RELATED DISORDERS	Heterozygous (de novo)	Likely pathogenic

Figure 3: Whole Genome Sequencing showing the patient's de novo SOX17 mutation

Discussion

- This case is the first report of a neonate diagnosed with HPAH associated with a pathologic SOX17 variant shortly after birth. It is also the first case report of an infant with HPAH who underwent creation of reverse Potts shunt physiology using a PDA stent.
- PAH is difficult to manage and has a high mortality rate particularly in the pediatric and neonatal population.
- HPAH is likely underdiagnosed. Early genetic testing should be considered in neonates and children with PAH.
- Further identifying and characterizing SOX17 variants associated with PAH through WGS can continue to improve clinical risk stratification and provide guidance for medical and surgical management of these rare neonatal cases.

Conclusion

PDA stent placement should be considered in children and infants with HPAH who are refractory to maximal medical management.

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