Absent Pulmonary Valve Syndrome, Tetralogy of Fallot Type, with Associated Aortopulmonary Collaterals case report and literature review

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Background

Absent pulmonary valve syndrome (APV) is an uncommon cardiac defect seen in conjunction with Tetraology of Fallot (TOF and TOF/APV) occurring in 3-6% of patients with TOF. (11) TOF/APV is associated with genetic abnormalities, most commonly 22q11 deletion (3). Hallmark anatomic/echocardiographic findings of TOF/APV include absent or rudimentary pulmonic valve, enlarged pulmonary artery (PA), free pulmonic regurgitation (PR) at the absent pulmonic valve, and an increased pressure gradient across the absent pulmonic valve. (4) Though multiple other vascular anomalies have been reported in conjunction with TOF/APV including absent branch PA (2, 11), anomalous origin of branch PA from other vascular structures (1, 8, 10, 13), anomalous pulmonary venous connections (3, 7), anomalous origin of coronary arteries (5), and aortic abnormalites (3,13). Presence of major aortopulmonary collateral arteries (MAPCA) has been rarely reported (6, 7, 12).

Here we present a case report and literature review on TOF/APV and MAPCA.

Clinical Report

The patient was born to a G2P1, 23 year old mother with adequate prenatal care. Prenatal echocardiogram initially diagnosed TOF with pulmonary atresia but echocardiogram on DOL 0 revealed TOF/APV. Cardiac CT on DOL1 revealed MAPCAs. Cardiac catheterization on DOL 11 revealed a MAPCA that was the sole blood supply of the left lower lobe in addition to two dual supply MAPCAs. At six months of age the patient underwent complete repair with placement of a pulmonary homograft, branch PA augmentation, extensive RVOT myectomy and VSD closure. Cardiac repair was uncomplicated and inotropic support was discontinued on post op day 7. Hospital course was complicated by respiratory distress necessitating brief re-intubation thought to be due to an aspiration event and not due to MAPCA. The patient was discharged on post op day 15.

Discussion

Clinical manifestations of TOF/APV include cyanosis, pre and perioperative respiratory distress, as well as failure to thrive. Specifically, the respiratory distress seen in TOF/APV is attributed to airway compression from an enlarged PA with or without enlargement of the branch PAs. Enlargement of the PA is attributed to two theories: First that agenesis of the ductus arteriosis is the cause and second is increased volume load to the PA due to free pulmonic regurgitation and increased pressure gradient seen across the stenotic valve annulus with resultant post stenotic dilatation. (4)

We found a few case reports (7, 12) one case series of refractory respiratory distress and pulmonary hemorrhage resulting from MAPCA in a neonate s/p repair of TOF/APV and a small retrospective study showing potential under diagnosis of TOF/APV which found 7/28 patients that underwent cardiac cath or MRI imaging were found to have MAPCA, though this may be underestimated as only 28/50 patients in the study underwent angiogram or MRI imaging. Only 3 of these 7 patients required coil embolization though all 7 had some degree of perioperative respiratory compromise. (6).

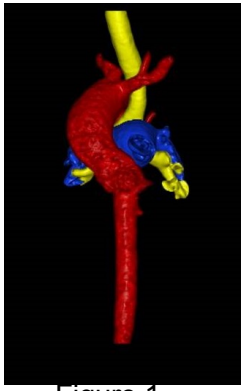

Figure 1

Cardiac CT 3D reconstruction of the aorta (red), pulmonary trunk (blue), and airways (yellow)

Figure 2

Cardiac CT 3D reconstruction of the pulmonary trunk demonstrating stenosis of the PV anulus and dilation of the main PA

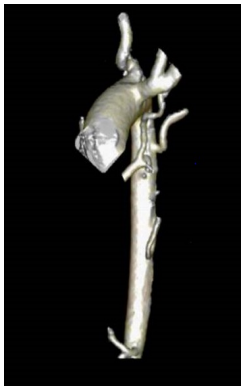

Figure 3

Cardiac CT 3D reconstruction of the aortic arch showing the MAPCAs distal to the arch

Conclusions

- TOF/APV is itself a rare entity that can be associated with abnormal vasculature, though its association with MAPCA may be underestimated.
- Respiratory distress in TOF/APV is common and most frequently attributed to extrinsic airway compression from the enlarged PA.
- MAPCA should be considered in refractory perioperative respiratory distress and intervened on if applicable.

References & Acknowledgements

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