Progressive pulmonary vein stenosis in Down syndrome infant: a rare cause of pulmonary hypertension

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SUMMARY

Pulmonary veins stenosis in a Down Syndrome infant with normal connection pulmonary vein is rare and precise incidence of this disease is unknown. We report a case of progressive multiple pulmonary vein stenosis in a Down Syndrome infant with congenital heart disease and transient myeloproliferative leukaemia. This case-report aims to improve awareness among physicians and sonographers of this disease and the importance of pulmonary vein assessment in congenital heart disease with unexplained pulmonary hypertension

INTRODUCTION

Down syndrome (DS) patients with associated cardiac defects have a high incidence of pulmonary hypertension (PHTN). Pulmonary vein stenosis (PVS) is rare in these patients.[1,2] A literature review in patients with DS from 1975 to 2011 found only 13 reported cases associated with PVS.[3] We report a DS infant with acyanotic congenital heart disease with PVS as the cause for progression of pulmonary hypertension.

CASE REPORT

A three-month-old boy with DS was admitted for respiratory distress secondary to bronchopneumonia. He had a history of respiratory distress secondary to transient myeloproliferative disease at two weeks of life for which he required non-invasive ventilation, single volume exchange transfusion and three days of intravenous cytarabine. An echocardiogram at the time showed mild dilatation of right atrium (RA) and right ventricle (RV), small secundum atrial septal defect (ASD), small right coronary artery (RCA) fistula to RA and estimated right ventricular systolic pressure (RVSP) of 50mmHg. The high estimated RVSP was attributed to pulmonary hypertension in newborn and elevated pulmonary pressure may also be caused by the myeloproliferative disease. His symptoms subsequently improved and transient myeloproliferative leukaemia resolved on follow up. During admission for bronchopneumonia at three months old, he was in mild respiratory distress with saturations between 80 to 85% in room air.

An echocardiography showed a systemic pulmonary artery pressure with marked right atrial and right ventricle dilatation and new findings of pulmonary veins with smaller calibre measured 1 to 1.5mm in diameter based on colour Doppler (Fig.1 and Fig.2). An attempt to wean him off oxygen caused further suprasystemic increase in his pulmonary arterial pressure.

Cardiac catheterisation revealed elevated RVSP (61mmHg), high mean pulmonary artery (PA) pressure (38mmHg) and PVR 19.3 Wood unit (WU). Angiogram revealed stenosis of right upper pulmonary vein (RUPV), mild stenosis of left upper pulmonary vein (LUPV) and hypoplasia and stenosis of both right and left lower pulmonary vein. His multiple pulmonary veins stenosis was not amenable to catheterisation-based intervention or surgical repair. He was treated conservatively and discharged home with non-invasive ventilation.

DISCUSSION

Patients with DS are known to have high incidence of PHTN due to congenital heart disease with significant left to right shunt and abnormal pulmonary vasculature.[1] It is possible that some DS patients with PVS may go unrecognised and cases were underreported due to poor echocardiographic windows and difficult pulmonary vein imaging. The progressive nature of the disease may cause under suspicion especially if the initial echocardiographic evaluation of pulmonary veins were normal. In our patient with DS and multiple PVS, diagnosis was similarly made at a young age of 3 months old similar to previous literature where patients with three of four PVS tend to present earlier at median age of four months.[5] The suspicion of PVS in our patient from transthoracic echocardiography was made from the small calibre pulmonary veins and flow turbulence from colour Doppler. A detailed echocardiography in particular assessing pulmonary veins in DS patients will make early diagnosis of PVS feasible. All DS patients with PHTN eventually will require a cardiac catheterisation for hemodynamic assessment and evaluation of all four pulmonary veins with direct pulmonary vein angiogram.

The utility of catheter interventions including stenting is limited in children as they provide only short-term relief, technically challenging due to small calibre veins and high restenosis rate which require re-intervention.

The role of pulmonary vasodilators remains controversial with only a few reports showing mixed results. In our patient we opted not use pulmonary vasodilator in view of multiple pulmonary vein involvement, which already carries guarded prognosis and high mortality rates. Pulmonary vasodilator

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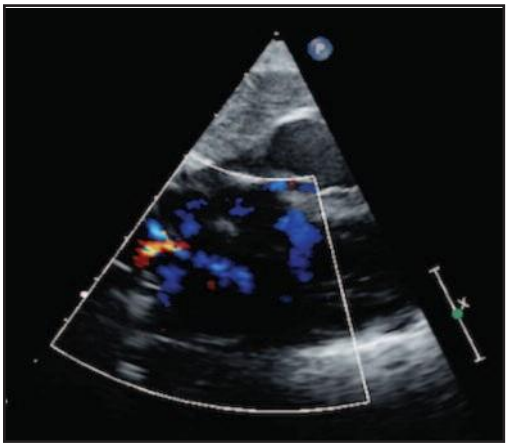

Fig. 1: RUPV and RLPV stenosis.

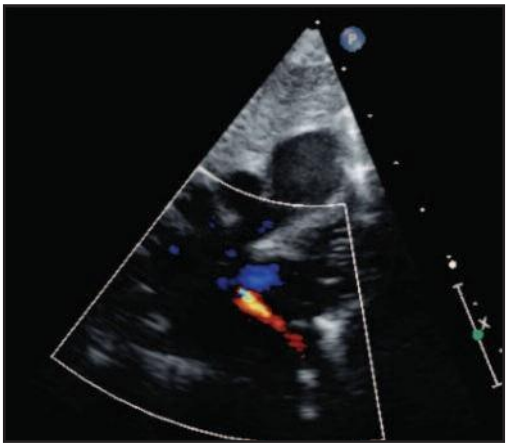

Fig. 2: LLPV stenosis.

may also cause dilatation of other non stenosed segments which further impair the pulmonary venous return to the heart.

In conclusion, pulmonary vein stenosis with normal connection in DS children is rare. High suspicion of PVS in DS children with unexplained PHTN is suggested with initial transthoracic echocardiography and careful evaluation of pulmonary veins.

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