Tetralogy of Fallot with Pulmonary Atresia and a Major Aortopulmonary Collateral Artery (MAPCA) in a Young Adult: A Case Report

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Abstract

Clinical Presentation: This report describes a case of a 24-year-old, acyanotic, female patient presenting with mild effort dyspnea, a right ventricular heave, a displaced cardiac apex, a loud single S2, a holosystolic murmur on the left parasternal area, and a continuous murmur at the left posterior chest. Transthoracic and transesophageal echocardiography demonstrated dilated left atrium and ventricle, hypertrophied right ventricle and a large ventricular septal defect with overriding of the aorta. However, both studies failed to visualize the pulmonic valve. The patient underwent cardiac catheterization which revealed the presence of pulmonary atresia. A descending aortogram showed the presence of a single large major aortopulmonary collateral artery (MAPCA) arising from the descending thoracic aorta which supplies confluent left and right pulmonary arteries. The pulmonary arterial vasculature is enlarged and pruned in appearance. The main pulmonary artery is non-atretic. Mean proximal pulmonary artery pressure was 68 mmHg, indicating severe pulmonary hypertension. Patient was started on digoxin and sildenafil.

Significance: This case report emphasizes the importance of a well-developed central pulmonary circulation and an adequate but not excessive pulmonary blood supply from an aortopulmonary vascular connection in the survival of patients with pulmonary atresia into adulthood.

Keywords: tetralogy of Fallot; pulmonary atresia; major aortopulmonary collateral artery

Synopsis

This report describes a case of a young adult patient presenting with mild effort dyspnea, a right ventricular heave, a displaced cardiac apex, a loud single S2, a holosystolic murmur on the left parasternal area, and a continuous murmur at the left posterior chest. Transthoracic and transesophageal echocardiography demonstrated a large ventricular septal defect with overriding of the aorta. However, both studies failed to visualize the pulmonic valve. Angiography revealed pulmonary atresia and a large major aortopulmonary collateral artery arising from the descending thoracic aorta.

Case

This is a case of F.C.L., a 24-year-old female who consulted at our institution complaining of mild dyspnea at moderate levels of exertion. She was initially suspected to have a congenital heart disease when she was noted to have a cardiac murmur at age one month during a routine vaccination at a medical clinic. She was then referred to a tertiary hospital institution where diagnostic work ups were done and was eventually diagnosed with truncus arteriosus (PDA). She was prescribed digoxin and was advised cardiac surgery but failed to comply. Due to persistence of her symptom, she eventually consulted at our institution for further evaluation.

During her first two decades of life, she was asymptomatic and was able to participate in competitive sports activities.

Four years prior, she began complaining of mild effort dyspnea. She consulted a private physician and a transthoracic echocardiogram was requested. The echocardiogram result was congenital heart disease, probably pink tetralogy of Fallot (TOF) with a patent ductus arteriosus (PDA). She was prescribed digoxin and was advised cardiac surgery but failed to comply. Due to persistence of her symptom, she eventually consulted at our institution for further evaluation. Physical examination revealed a right ventricular heave, a displaced cardiac apex, a loud single S2, a holosystolic murmur with thrills at the left parasternal border and a continuous murmur at the left posterior chest. There were no clubbing or cyanosis noted. The result of her 6-minute walk test was 504 meters.

A 12-lead electrocardiogram showed left ventricular hypertrophy by voltage criteria and ST-T wave changes consistent with left ventricular strain and/or digitalis effect. A chest radiograph revealed cardiomegaly with hypervascularized lungs.

An arterial blood gas taken at room air showed compensated respiratory alkalosis (pCO2 30.9 mmHg, HCO3 - 20.8 meq/L, pH 7.44) and moderate hypoxemia (PO2 55.6 mmHg, O2 saturation 90.6% at 21% FIO2).

Transthoracic echocardiography revealed a large ventricular septal defect (VSD) with overriding of the aorta. The left ventricle was dilated with adequate overall systolic function (LVEDD: 51 mm; EF: 71%). The left atrium was also dilated (AP diameter 47 mm) and the right ventricle was

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hypertrophied. The pulmonic valve could not be visualized. Bubble contrast showed right-to-left shunting across the VSD. A transesophageal echocardiography done also failed to visualize the pulmonic valve. All four pulmonary veins were shown to drain into the left atrium. The mitral, tricuspid and aortic valves were structurally normal. There was no PDA noted. Presumptive diagnosis was TOF with probable pulmonary atresia and major aortopulmonary collateral arteries (MAPCAs).

The patient underwent cardiac catheterization which confirmed the presence of pulmonary atresia (Figure 1). Her right ventricular pressure was systemic at 115/5 mmHg.

![Fig. 1. Right ventriculography in AP view demonstrates the passage of radiocontrast dye to the left ventricle through a ventricular septal defect. The aorta overrides the ventricular septal defect. The main pulmonary artery was not visualized indicating a pulmonary atresia. BC=Berman catheter; Ao=aorta; RV=right ventricle; LV=left ventricle; VSD=ventricular septal defect.](image1)

Her aortic saturation at room air was 90.7%. A descending aortogram showed the presence of a single large major aortopulmonary collateral artery (MAPCA) arising from the descending thoracic aorta which supplies confluent left and right pulmonary arteries (Figures 2, 3 and 4). The pulmonary arterial vasculature is enlarged and pruned in appearance. Proximal pulmonary artery pressure was sub systolic at 90/45 with a mean of 68 mmHg, indicating severe pulmonary hypertension.

![Fig. 2. Retrograde proximal descending aortogram shows a large single major aortopulmonary collateral artery (MAPCA) originating from the thoracic descending aorta. Ao=aorta; MAPCA= major aorto-pulmonary collateral artery; PC=pigtail catheter.](image2)

![Fig. 3. Angiography was done following the cannulation of the MAPCA and selective cannulation of a lower branch of the LPA using a reverse Berman catheter. Contrast is seen anterogradely filling a branch of the left pulmonary artery, but with poor capillary blush. LPA=left pulmonary artery; BC=Berman catheter, reverse.](image3)

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![Fig. 4. Selective injection into the MAPCA defines the confluent pulmonary artery vessel course and distribution. Contrast is seen filling the native pulmonary arterial tree. Again there is note of pruning of the pulmonary vasculature. RPA=left pulmonary artery; RPA= right pulmonary artery; MPA=main pulmonary artery; BC=Berman catheter, reverse.](image4)

Patient was started on digoxin and sildenafil. No surgical intervention was planned because of the presence of severe
pulmonary hypertension.

Discussion

Among patients with TOF, pulmonary atresia occurs in approximately 12 percent of cases. In these cases, the native pulmonary arteries may be of normal size, but are commonly hypoplastic. In the presence of good-sized central pulmonary arteries, pulmonary blood supply is via a PDA (about one half to two-thirds of cases). In the remaining cases, characterized by hypoplastic or absent pulmonary arteries, pulmonary blood supply is from MAPCAs. Our patient has a non-atretic main pulmonary artery, good-sized and confluent right and left pulmonary arteries which are supplied by a single MAPCA, a quite unusual finding which does not fit into any of the types of aortic-to-pulmonary circulation in TOF with pulmonary atresia patients categorized by Castaneda and colleagues (Table 1).

<table>
<thead>
<tr>
<th>Nonatretic MPA</th>
<th>PDA</th>
<th>RPA-LPA confluence</th>
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<td>Type 1</td>
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Table I. Classification of Pulmonary Blood Flow in TOF with Pulmonary Atresia

Patients with large MAPCAs and unrestricted pulmonary blood flow are at risk of future development of congestive heart failure and pulmonary vascular disease. We believe our patient already has beginning cardiac failure hence we started her on digoxin. Sildenafil was given for the treatment of pulmonary hypertension. Palliative treatment should be emphasized. The oldest survivors are typically those with adequate but not excessive aortic-to-pulmonary collateral circulations but the long-term consequence is chronic volume overload of the left ventricle resulting to cardiac failure. Such patients have limited cardiac reserve. All of these characteristics were exemplified by our patient. The average age of death of such uncorrected adult patients is 33 years and longevity beyond the fourth decade is already exceptional.

Conclusion

We discussed a rare adult case of TOF with pulmonary atresia and a MAPCA arising from the descending thoracic aorta supplying the non-hypoplastic central pulmonary arteries. This case report emphasizes the importance of a well-developed central pulmonary circulation and an adequate but not excessive pulmonary blood supply from an aortopulmonary vascular connection in the survival of patients with pulmonary atresia into adulthood. Future care is directed at preserving ventricular function and temporizing the development of severe and irreversible pulmonary hypertension. Palliative treatment should be emphasized.

References