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A 50-year-old unrepaired patient with pulmonary atresia and ventricular septal defect (RCD code: IV-2A.3)

Agnieszka Sarnecka*, Anna Tyrka, Grzegorz Kopeć, Piotr Podolec

Department of Cardiac and Vascular Diseases, Center for Rare Cardiovascular Diseases John Paul II Hospital, Institute of Cardiology, Jagiellonian University Medical College, Krakow, Poland

Abstract

Pulmonary atresia with ventricular septal defect (PA + VSD) is a cyanotic congenital heart disease, also classified as Tetralogy of Fallot with pulmonary atresia. PA + VSD accounts for about 1–2% of congenital heart defects. The intracardiac anatomy is similar to tetralogy of Fallot but there is no direct communication between the right ventricle and pulmonary arteries. Major problems with surgical treatment are related to complexity of the pulmonary vascular bed. We report a case of a 50-year-old woman with congenital heart disease who was admitted to our Centre in July 2012. Congenital heart disease was first diagnosed at the age of 28 and at that time it was classified as a pulmonary valve atresia with ventricular septal defect with right-to-left shunt and common arterial trunk. Diagnostics performed in our Centre confirmed complicated anatomy of vessels in the chest, especially narrow and hypoplastic major aortopulmonary collateral arteries arising from descending aorta and left subclavian artery. The congenital heart disease was reclassified as a pulmonary atresia with ventricular septal defect. JRCD 2016; 2 (8): 270–274

Key words: congenital heart defect, cyanotic congenital heart disease, Tetralogy of Fallot, major aortopulmonary collateral arteries, MAPCAs

Background

Pulmonary atresia with ventricular septal defect (PA + VSD) is a cyanotic congenital heart disease, also called Tetralogy of Fallot with pulmonary atresia. The intracardiac anatomy is similar to tetralogy but there is no direct communication between the right ventricle and pulmonary arteries. PA + VSD accounts for about 1–2% of congenital heart defects and is commonly associated with 22q11.2 microdeletion [1,2].

The pattern of pulmonary arteries anatomy is versatile, from good size pulmonary arteries supplied from patent ductus arteriosus to multifocal non-confluent or confluent hypoplastic pulmonary arteries supplied by major aortopulmonary collaterals (MAPCAs). MAPCAs are arteries present in embryonic life, that regresses as the normal pulmonary arteries develop. In PA + VSD the native pulmonary circulation is underdeveloped resulting in further growth of MAPCAs, that become the main blood supplier to the lungs. MAPCAs typically arise from the descending aorta but can also arise from aortic arch or other systemic arteries like carotid, subclavian or even coronary arteries. Some complementary to echocardiography non-invasive studies such as computed tomography or magnetic resonance that are available nowadays can be very useful in determining the exact anatomy of the heart defect [1,3–5].

Usually surgical treatment is necessary in early childhood but patients with non-confluent pulmonary arteries with adequate pulmonary blood flow could survive until adulthood without surgery. However, reported overall life expectancy without surgery in patients with PA + VSD is as low as 50% at 1 year of age and 8% at 10 years. Unrepaired patients present with progressive cyanosis, chronic heart failure, dilation of the ascending aorta and aortic regurgitation [1,6–8].

Case presentation

A 50-year-old Caucasian woman with congenital heart disease was referred to Department of Cardiac and Vascular Disease in July 2012 for cardiologic evaluation. Congenital heart disease, referred as pulmonary valve atresia with ventricular septal de-

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* Corresponding author: Department of Cardiac and Vascular Diseases, Center for Rare Cardiovascular Diseases John Paul II Hospital, Institute of Cardiology, Jagiellonian University Medical College, Pradnicka 80 str., 31-202 Krakow, Poland; tel. 0048 12 614 22 87, fax 0048 12 423 43 76; e-mail: a.sarnecka@szpitaljp2.krakow.pl

Table 1. The results of right heart catheterisation (performed at the age of 28)

Parameter	Pressure [mm Hg]	Oxygen saturation [%]
aorta	160/84/112	84
left ventricle	160/8/16	87
right ventricle	165/8/12	64
right atrium	8/6/8	56

fect with right-to-left shunt and common arterial trunk, was first diagnosed at the age of 28. She had a history of paroxysmal atrial fibrillation (one documented episode in 2010), hypothyroidism, well-controlled asthma and a history of neurosurgical procedure for cerebral haematoma of the right hemisphere and clipping of an aneurysm of medial cerebral artery in 2007. She had also a family history of heart defects in her mother's family and polycythaemia vera in her father and sister. More detailed data on her family history including types of cardiac defects were unknown. She had never undergone any genetic evaluation, and had no apparent symptoms characteristic for 22q11.2 microdeletion like DiGeorge syndrome or immune deficiencies.

Diagnostic evaluation including right heart catheterization was first performed at the age of 28. Transthoracic echocardiography study revealed VSD, hypertrophy of the right ventricular wall, overriding aorta with biventricular connection and right-sided aortic arch. Pulmonary trunk was not detected. Transoesophageal echocardiographic study confirmed morphology typical for PA with absent pulmonary trunk and the presence of two aortopulmonary collaterals arising from aortic arch. Further information was provided through aortography which revealed a vessel described as the pulmonary trunk arising from descending aorta and two additional bronchopulmonary collaterals draining to lower lobes of both lung. Hemodynamic evaluation revealed left ventricular desaturation to 87% and pressure equalization in both ventricles (Table 1). The diagnosis of PA + VSD with right-to-left shunt and common arterial trunk was established and she was qualified for optimal medical therapy including vitamin K antagonist, digoxin, spironolactone, torasemide, allopurinol, levothyroxine, budesonide, formoterol, tiotropium and continuous oxygen therapy. She had recurrent therapeutic phlebotomies, however with no signs of hyperviscosity syndrome.

On admission to our Centre in 2012 the patient complained of easy fatigue and poor exercise tolerance in functional class III by World Health Organisation (WHO). Physical examination revealed central cyanosis, clubbed fingers, systolic murmur 3/6 in Erb's point and basal crepitation over the left lung. Laboratory workup disclosed elevated levels of haemoglobin, high-sensitive troponin T and N-terminal prohormone of brain natriuretic peptide (NT-proBNP) (table 2).

Transthoracic echocardiography showed PA, VSD, overriding aorta with biventricular connection and dilated ascending aorta to 52 mm (Figure 1). The size of both ventricles were within normal

Table 2. Abnormal laboratory tests revealing polycythemia, hypercholesterolemia and elevated levels of high-sensitive troponin T and NT-proBNP

Parameter	Value	Unit	Reference values
INR	1.38		[0.8–1.2]
TCHOL	**5.79**	mmol/L	[3.10–5.00]
LDL	**3.95**	mmol/L	[<3.0]
HDL	1.17	mmol/L	[>1.2]
TG	1.83	mmol/L	[<1.7]
hsCRP	3.68	mg/L	[<3.0]
eGFR	>90	mL/min/1,73 m^2	[>60]
K+	4.9	mmol/L	[3.5–5.1]
Na+	142	mmol/L	[136 – 145]
TSH	1.550	µmol/L	[0.27–4.20]
Glucose	5.3	mmol/L	[3.4–5.6]
ALT	22	U/l	[<33]
AST	22	U/l	[<23]
NT-proBNP	**300.5**	pg/mL	[<125.0]
Troponin hsT	**0.033**	ng/mL	[<0.014]
D-dimer	<300	mg/L	[<486]
fibrinogen	3.26	g/L	[1.80 – 3.50]

WBC – white blood cells, RBC – red blood cells, HGB – haemoglobin, HCT – hematocrit, MCV – mean cell volume, PLT – platelets count, INR – international normalized ratio, TCHOL – total cholesterol, LDL – low density lipoproteins, HDL – high density lipoproteins, TG – triglycerides, hsCRP – high-sensitive C-reactive protein, eGFR – estimated glomerular filtration rate, TSH – thyroid stimulating hormone, ALT – alanine aminotransferase, AST – aspartate transaminase, NT-proBNP – N-terminal prohormone of brain natriuretic peptide

limits with hypertrophied right ventricular wall to 17 mm in diastole. The contractility of both ventricles was normal without segmental wall motion abnormalities. The ejection fraction of left ventricle was 60%, tricuspid annular plane systolic excursion (TAPSE) was 24mm and tricuspid annular peak systolic velocity (S') was 15cm/s. Mild regurgitation of aortic and tricuspid valve were revealed.

The distance in 6-minutes walking test was 522m with reduction of blood oxygen saturation from 85% to 77% and dyspnea estimated at 7 in Borg scale. The lung perfusion scintigraphy did not reveal any deficits. The results of body plethysmography were normal except for increase airway resistance. The diffusing capacity or transfer factor of the lung for carbon monoxide (DLCO) was decreased to 60%. Further evaluation was performed using computed tomography (CT) angiography of the heart and thoracic aorta which revealed complicated anatomy of vessels in the chest (Figure 2,3,4). The study showed tricuspid aortic valve situated symmetrically over intraventricular septum defect sized 20x19 mm. The aorta was right-sided and was dilated to 43 × 39 mm at the valve level and to 54x54 mm at the ascending part. The first vessel arising

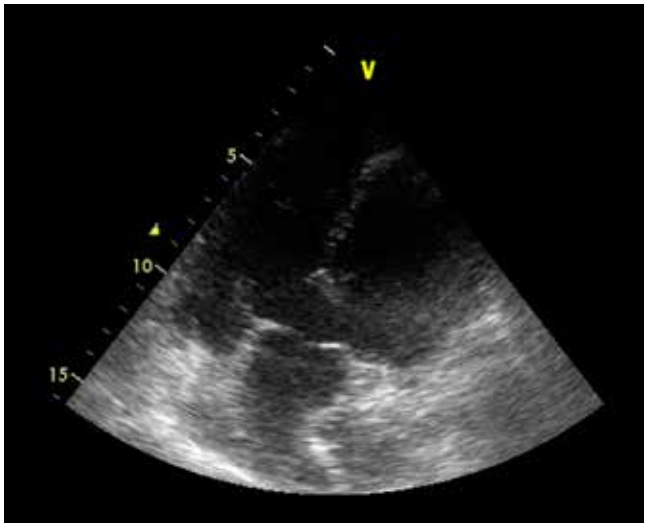

Figure 1. Transthoracic echocardiography showed ventricular septal defect, overriding aorta with biventricular connection and dilated ascending aorta

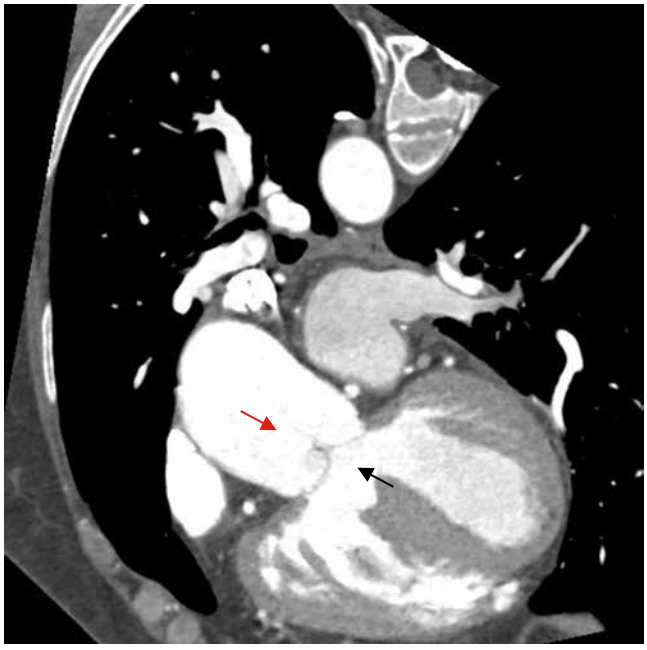

Figure 2. Computed tomography angiography of the heart and thoracic aorta. Ventricular septal defect (black arrow), overriding aorta with biventricular connection (red arrow)

from the aorta was left brachiocephalic artery which divided into left common carotid artery and left subclavian artery. Left main coronary artery was long and arising in an acute angle. The course of the other coronary arteries was typical. The pulmonary venous return into the left atrium was normal. The study showed detailed anatomy of narrow and hypoplastic MAPCAs. The two of them with ostia diameter of 12 mm and 7 mm arose from descending aorta at the level of tracheal bifurcation, then merged and went to the right lung. The third MAPCA with ostium diameter of 14 mm arose from the descending aorta 20 mm below tracheal bifurcation and supplied the lower lobe of the left lung. The upper lobe of

Figure 3. Computed tomography angiography of the heart and thoracic aorta. Dilated ascending aorta (arrow)

the left lung was supplied by the fourth MAPCA that arose from the left subclavian artery. The ostium diameter of this MAPCA was 13 mm but it was narrowed in some segments to 4–5 mm.

Review of literature

Guidelines of the European Society of Cardiology for the management of grown-up congenital heart disease recommend to evaluate cardiological status of patients with PA + VSD in a specialized grown-up congenital heart disease centre at least once a year. Patients surviving unrepaired until adulthood should be reconsidered for surgical or interventional procedures even if they were recognized as inoperable years ago. Indications for surgery may involve: progressive chronic heart failure, dilation of the ascending aorta and aortic regurgitation. Surgical treatment can be suitable for patients without right or left ventricle dysfunction who have good-size confluent pulmonary arteries or large MAPCAs anatomically suitable for unifocalization and who have not developed severe pulmonary hypertension. In some cases, interventional procedures like balloon dilation/stenting of MAPCAs should be considered to improve pulmonary flow. Our patient had non-confluent, hypoplastic and multiple narrowed pulmonary arteries that were not suitable for surgery nor for interventional approach. On the other hand, benefits from pulmonary artery hypertension (PAH) specific treatment in patients with segmental PAH are not definitively confirmed. [1,6–11]

Despite the fact that about 8% of unrepaired patient with PA+VSD survived to 10 years, adult survivors are rare. Life expectancy in unrepaired patients with PA+VSD usually does not exceed three decades. There are only a few documented cases of patients

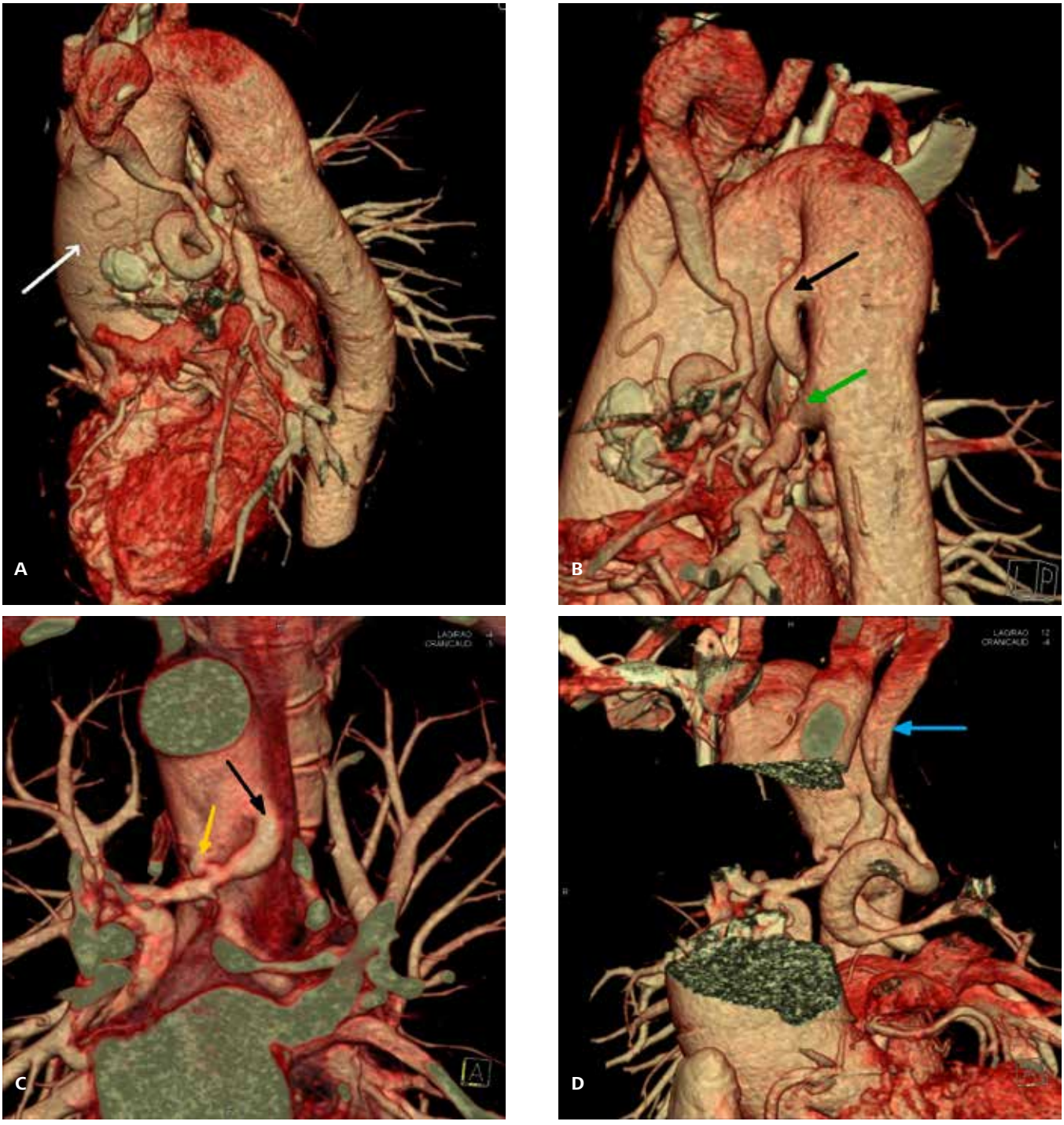

Figure 4. Computed tomography angiography of thoracic aorta. A. Right-sided aorta, ascending aorta dilated to 54 mm (white arrow). B, C, D. The anatomy of major aortopulmonary collaterals (MAPCAs). (MAPCA I – black arrow, MAPCA II – yellow arrow, MAPCA III – green arrow, MAPCA IV – blue arrow)

who survived to 50 years of age – the oldest reported patient was 59. The most important determinant of survival is a presence of sufficient but not too excessive pulmonary circulation. [9,12]

Patient management and follow-up

The previous qualification of the patient as inoperable was sustained. Since no PAH was diagnosed no PAH-specific treatment was initiated. The patient was qualified for further conservative treatment and close observation. Over a few months of follow-up, the patient's cardiological condition was stable. After that the patient was diagnosed with glioblastoma and eventually died at the age of 50 from the tumor.

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