# Medical Expertise

"Development of the European Network in Orphan Cardiovascular Diseases"
„Rozszerzenie Europejskiej Sieci Współpracy ds Sierocych Chorób Kardiologicznych"

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### CASE SUMMARY

22-year-old man after pulmonary valve (PV) valvulotomy in childhood was admitted for cardiological evaluation due to progressive loss of exercise capacity. The surgery was performed in 1991. In 1995 he was diagnosed with endocarditis of mitral valve and chronic hepatitis B. In 1999 he underwent a surgical operation of the brain abscess. In 2009 he was hospitalized due to a transient ischemic attack (TIA). At the age of 17 he was diagnosed with epilepsy and currently receives antiepileptic medications.

At present he complains of easy fatigue, dyspnea, and decrease of exercise tolerance. He is consider to be in class II by New York Heart Association (NYHA). On admission he is stable, with no signs of peripheral edema or pulmonary congestion. Physical evaluation reveals systolic murmur in 2$^{nd}$ intercostal area.

ECG showed regular sinus rhythm of 70 bpm with incomplete RBBB, negative T wave in III and aVF. Holter ECG disclosed average heart rate (HR) of 63 bpm, 6 episodes of supraventricular and 4 ventricular arrhythmias, 4 episodes of sinus bradycardia with minimal heart rate of 35 bpm. Chest X-ray did not reveal any significant abnormalities. Laboratory studies showed: RBC – 5600000, Hg- 17,7 g/dl, INR- 1.01, ALAT- 9 U/L, Creatinine – 98umol/L; total cholesterol- 3,6 mmol/l, LDL- 2,13 mmol/l, proteins- 72,3 g/L with albumins- 39,1 g/L. Cardio-pulmonary exercise test showed mildly impaired exercise tolerance with peak oxygen uptake of 23,7 ml/kg/min at maximal workload of 13,1 METs. Bodypletysmography showed normal ventilation and respiratory resistant.

Echocardiography showed enlarged right ventricle, coronary sinus of 23x14 mm, PV gradient of 24/14 mmHg.

Transesophageal echocardiography showed 12 mm ASD with left-to-right shunt, 3 pulmonary veins draining to the left atrium and enlarged coronary sinus. Also persistent left superior vena cava to the coronary sinus was identified.

Right heart catheterization showed left-to-right shunt at the level of interatrial septum with Qp/Qs ratio of 1,22, PV systolic gradient of 13mmHg and normal pulmonary arterial pressure and resistance: right atrium (7/4/4 mmHg), right ventricle (27/0/6 mmHg), pulmonary artery (14/2/8 mmHg), aorta (138/76/94). Oximetry showed: right atrial oxygen saturation (SatO2)



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of 67,6%, right ventricular SatO2 of 76,5%, pulmonary artery SatO2 of 75,6 %, aortic SatO2 of 97.5 %, SatO2 in vena cava inferior - 70 % and in vena cava superior - 67,3 %. Cardiac index was 2.0 l/min/m2 and cardiac output 3.6 l/min.
Angio computed tomography revealed patent left vena cava superior draining to the right atrium, 4 pulmonary veins draining correctly to the left atrium and ASD with maximal diameter of 14 mm. Cardiac magnetic resonance showed moderate regurgitation through pulmonary valve – netto 59 ml/cycle with maximal speed of 1.4 m/s. MR confirmed persistent left superior vena cava. The authors are concerned whether ASD closure with or without PV replacement should be advised to the patient now.

## DISCUSSION

Atrial septal defect (ASD) is the most common congenital cardiac lesion in adults accounting for 10-15% of all congenital heart defects. There is a few types of ASD, that can be distinguished: ostium secundum (70%), ASD type II located in the central part of the interatrial septum, ostium primum (20%) ASD type I located in the lower part of the septum, sinus venosus type ASD (10%) located at the inlet of one of the cava veins or defect involving coronary sinus. There is no racial predilection and it affect female more often than male (ratio F:M 2:1). In childhood most ASD are asymptomatic. Symptoms of an ASD depends on its size and associated left-to-right shunting. These include easy fatigability, dyspnea, arrhythmias, decrease in exercise tolerance. Untreated ASD are associated with significantly shortened life expectancy, right heart failure, pulmonary hypertension, arrhythmias or paradoxical emboli. Definite treatment of ASD type II include percutaneous or surgical closure [3]. Indications for intervention in ASD following the current guidelines [3] are:

1. Patients with significant shunt and PVR < 5 WU should undergo ASD closure regardless of symptoms [class I B].
2. Device closure is the method of choice for secundum ASD closure when applicable [class I C].
3. All ASDs regardless of size in patients with suspicion of paradoxical embolism should be considered for intervention [class II a C].
4. Patients with PVR > 5 WU but <2/3 SVR or PAP <2/3 systemic pressure and evidence of net left- right shunt (Qp:Qs > 1.5) may be consider for intervention [class II b C].
5. ASD closure must be avoided in patients with Eisenmenger physiology [class III C].

Outcome is best with repair at age under 25 years. However, presence of sinus venosus defect or right – sided anomalous pulmonary venosus connections and visualization of normal left – sided pulmonary venosus connections are the most important prerequisites for surgical planning.

Congenital pulmonary valve stenosis is the most common form of pulmonic stenosis. It accounts for 80% of right ventricular outflow tract obstruction [4]. Balloon valvuloplasty tends to be the first choice for treatment. In case of subvalvular or supravalvular right ventricle outflow tract obstruction open-heart surgery is needed. The severity of pulmonary stenosis is determined by echocardiography [4].

## EXPERT'S OPINION


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This is a case of a young adult after surgical correction of congenital PV stenosis in childhood, with left-to-right shunting through an ASD, history of brain ischemia and neurosurgical interventions in the past.

Due to the history of ischemic cerebral stroke likely caused by paradoxical embolus, qualification for the ASD closure is reasonable. However, prior to surgery, more detailed evaluation of the heart and vascular connections is needed. ASD with 14 mm of diameter would probably give bigger shunt expressed by Qp/Qs ratio. The anatomy of the ASD, persistent left vena cava or coronary sinus is vaguely studied.

Gradient over pulmonary valve is mildly elevated. Long-term effect of the PV valvulotmy seems to be good. There are no indications for PV replacement.

Detailed neurological evaluation and qualification for the open-heart procedure is mandatory. It is yet necessary to define whether extracorporeal circulation may cause worsening of patients neurological conditions.

## CONCLUSION

Surgical closure of ASD should be recommended. Reevaluation of the anatomy of the heart and vascular conncetions on angioCT and CMR is needed prior to the operation. Pulmonary valve replacement is not recommended. Neurological qualification for the operation is mandatory.

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