Medical Expertise

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

EXPERT: Jakub Podolec MD, PhD, cardiologist

Affiliation: *Department of Hemodynamics and Angiocardiography, Cardiology Institute, Collegium Medicum, Jagiellonian University at the John Paul II Hospital, Krakow, Poland*

CASE SUMMARY

The case of 22-year-old Caucasian male, with no remarkable family history, who underwent pulmonary valve valvulotomy in his early childchood (at the age of 1) due to congenital stenosis. In the medical review he suffered from endocarditis of the mitral valve (1995) and was diagnosed with chronic hepatitis type B. Four years later in 1999 the patient underwent cranial surgery with removal of an abscess of the brain. In the age of 17 years epilepsy was diagnosed. In the year 2009 he was hospitalized with TIA and stroke. He was referred to our Center due to decrease in exercise tolerance and easy fatigability.
12–lead ECG showed sinus rhythm, 70/min, no deviated axis, non-complete right His bundle branch block and negative T waves in III, aVR. The transoesophageal echocardiography revealed atrial septal defect with left-to right shunting after chloride sodium injection to the left ulnaris vein. The MRI of the heart confirmed sinus venosus atrial septal defect with patent superior vena cava and moderate pulmonary regurgitation. Angio-CT scan showed normal four pulmonary veins draining to the left atrium. In the spiroergometry test moderate exercise disability with peak oxygen consumption of 23,7 ml/kg/min was noticed. 24 - hour Holter ECG did not demonstrate any significant ventricular or supraventricular arrhythmias. Patient remains on oral pharmacohteraphy (acidum salicylicum and walproinian acid.

LITERATURE REVIEW

Atrial septal defect (ASD) is the most common congenital cardiac lesion in adults (10-15%). There can be distinguished 3 types of ASD: ostium primum type (20%) ASD I, ostium secundum (70%) ASD II and sinus venosus (10%) type vena cava superior or inferior (SV-ASD). There is no racial predilection known influencing on the frequency of ASD onsets. It affect female more often than male, and ratio is F:M 2:1. Sinus venosus atrial septal

defect (SVASD), encompasses approximately 4% to 11% of atrial septal defects (ASDs) [1, 2]. The typical malformation is an interatrial communication caused by a deficiency of the common wall between the superior vena cava (SVC) and the right-sided pulmonary veins [2, 3]. SVASD is commonly associated with anomalous pulmonary venous connection (APVC) of some or all of the pulmonary veins, which produces additional left-to-right shunting [3, 4]. In childchood most SV-ASD are asymptomatic. Symptomes may develop as the age and depends on the size of the associated left to right shunt : easy fatigability, dyspnea, arrytmias, decrease in exercise tolerance. Untreated ASD are associated with significantly shortened life expectancy due to: right heart failure, pulmonary hypertension, arrhythmias, paradoxical embolus, reccurent pulmonitis. Few case presentations according treatment of sinus venosus ASD in adult patients were published in the literature. Gatzoulis MA et al. report a 31 y.o. patient who underwent successful surgical intervention on ASD despite high idiopathic pulmonary hypertension diagnosed before. . Surgical repair and continued advanced therapy with bosentan 125 mg and aspirin 75 mg proved to be successful. Patient experienced post-operative improvement in exercise capacity, dyspnea, and a reduction in pulmonary pressures from 47 to 25 mmHg after surgery [5].

EXPERT'S OPINION

Due to the anatomical reasons it is not possible to perform percutaneous intervention. Symptoms may be acceptable for the patient at this moment (NYHA II), but if symptoms increase decision of surgical repair of the defect may become the best option for the patient. Due to the stroke that the patient suffered in the past he might be a candidate for surgery earlier than later. As well documented in patients with congenital heart diseases, MRI should be considered at follow up before any decision making [6, 7]. Right heart cath should be done in the nearest future.

CONCLUSION

Right heart catheterization is indicated. Surgical correction with ASD closure, eventually pulmonary venous drainage correction is considered to be the prefered method.

REFERENCES

John Paul II Hospital in Kraków
Jagiellonian University, Institute of Cardiology
80 Prądnicka Str., 31-202 Kraków;
tel. +48 (12) 614 33 99; 614 34 88; fax. +48 (12) 614 34 88
e-mail: rarediseases@szpitaljp2.krakow.pl
www.crcd.eu

1. Peacock TB. Malformations of the heart. In: Peacock TB, ed. On Malformations, &c., of the Human Heart: With Original Cases. London, UK: John Churchill; 1858: 11–102.
2. Oliver JM, Gallego P, Gonzalez A, Dominguez FJ, Aroca A, Mesa JM. Sinus venosus syndrome: atrial septal defect or anomalous venous connection? A multiplane transoesophageal approach. Heart. 2002; 88: 634–638.
3. Van Praagh S, Carrera ME, Sanders SP, Mayer JE, Van Praagh R. Sinus venosus defects: unroofing of the right pulmonary veins: anatomic and echocardiographic findings and surgical treatment. Am Heart J. 1994; 128: 365–379.
4. Kirklin JW, Barratt-Boyes BG, eds. Cardiac Surgery: Morphology, Diagnostic Criteria, Natural History, Techniques, Results, and Indications. 2nd ed. New York, NY: Churchill Livingstone; 1993; 1: 609–644.
5. Gatzoulis MA, Giannakoulas G. Sinus venosus atrial septal defect in a 31-year-old female patient: a case for surgical repair. Eur Respir Rev. 2010 Dec;19(118):340-4.
6. Prompona M, Muehling O, Naebauer M et al., MRI for detection of anomalous pulmonary venous drainage in patients with sinus venosus atrial septal defects. Int J Cardiovasc Imaging. 2011 Mar;27(3):403-12.
7. Gulati GS, Hoey ET, Gopalan D et al., Sinus venosus atrial septal defect in adults: utility of cardiovascular MRI in resolving this diagnostic dilemma. Heart Lung Circ. 2010 Oct;19(10):615-9.

John Paul II Hospital in Kraków
Jagiellonian University, Institute of Cardiology
80 Prądnicka Str., 31-202 Kraków;
tel. +48 (12) 614 33 99; 614 34 88; fax. +48 (12) 614 34 88
e-mail: rarediseases@szpitaljp2.krakow.pl
www.crcd.eu