Medical Expertise

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

EXPERT: Dr Anna Prokop-Staszecka, pulmonologist

Affiliation: *Department of Pulmonary Diseases, John Paul II Hospital, Krakow, Poland*

CASE SUMMARY

The authors presented a case of 30 year old male with non-corrected congenital ventricular septal defect, bicuspid aortic valve, ascending aorta aneurysm and suspection of pulmonary hypertension. At the time patient with central cyanosis, with heart failure symptoms in functional class NYHA II/III. The main issue is further farmacotherapy and prospective surgical treatment.

DISCUSSION

Ventricular septal defect (VSD) is a very common congenital heart defect in children, but due to spontaneous and surgical closure, it is less commonly encountered in adults. Studies have reported the long-term outcome of small VSDs that were not closed during childhood. About one-third of patients with VSD initially managed medically require surgical intervention later in life.

The most common surgical indications are AR, LV dilation, and pulmonary hypertension. Patch closure and a transatrial approach were most frequently used. Suture closure was most commonly used in patients with a residual VSD, which was present in 18% of patients post-operatively and in 24% at late follow-up. (1)

Indications for surgery in ascending aorta aneurysm are based mainly on aortic diameter and derived from findings on natural history regarding the risk of complications weighed against the risk of elective surgery. Surgery should be performed in patients with a BAV, who have a maximal aortic diameter ≥55 mm; these face a lower risk of complications than inMarfan. A lower threshold of 50 mm can be considered in patients with additional risk factors, such as family history, systemic hypertension, coarctation of the aorta, or increase in aortic diameter >3 mm/year, and also according to age, body size, comorbidities, and type of surgery. Regardless of aetiology, surgery should be performed in patients who have a maximal aorticdiameter ≥55 mm.(2)

Pulmonary arterial hypertension of variable degree is commonly associated with adult congenital heart disease. Depending on size and location of the underlying cardiac defect as well as on repair status, pulmonary arterial hypertension may present with or without reversed

shunting and associated cyanosis. Cyanosis is common in patients with congenital heart disease and PAH. Right-to-left shunting reduces arterial oxygen content and is associated with secondary hypoxic damage to other organ systems, thus leading to multiorgan disease.Disease-Targeting Therapies Different drugs, including prostacyclin in various forms, nitric oxide, phosphodiesterase inhibitors such as sildenafil and endothelin-receptor antagonists such as bosentan, have been demonstrated to be beneficial in patients with idiopathic PAH. Until recently, there was little evidence to show efficacy of these drugs in patients with congenital heart disease or Eisenmenger syndrome. Furthermore, worsening right-to-left shunting due to concurrent reduction in systemic vascular resistance was assumed. Recently, prostacyclin analogues have been shown to improve functional capacity, oxygen saturations, and pulmonary hemodynamics in patients with congenital heart disease and PAH. (3)

EXPERT'S OPINION

There is little evidence to show efficacy of PAH-specific drugs in patients with congenital heart disease or Eisenmenger syndrome. A typical pharmacotherapy for heart failure should be continued. Basing on actual patient`s condition surgical VSD closure and aortic aneurysm repair should be considered.

REFERENCES

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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