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

The authors presented a case of a 27 years old caucasian male with low exercise tolerance and dyspnea - New York Functional Class II (NYHA). He had a history of Tetralogy of Fallot. In 1987 cardiosurgical operation was performed, probably Blalock-Taussig shunt – no documentation was available. There was no follow-up till 2012. He had a history of alcohol abuse and was a heavy smoker. On admission he presented dyspnea (NYHA class II), reported burning chest pain at rest and on exertion, dizziness, heart palpitations. There was an episode of syncope on exercise with no further diagnosis.

On physical examination blood pressure (BP) was 110/70 mmHg on right arm and 95/70 mmHg on left arm. Heart rate was 75 bpm, regular. Cyanosis of lips and oral mucosa and nail clubbing was present. Murmur over the base of the heart was heard.

ECG showed sinus rhythm 75 bpm with right axis deviation, P pulmonale, signs of RVH. Laboratory tests revealed red blood cells count 6,09x10^6/uL, HCT 60,9%, HGB 21,0 g/dL, elevated level of LDL cholesterol.

Cardiopulmonary exercise test showed poor exercise tolerance. It was terminated after 10:09 minutes, due to general fatigue at 6,3 METs, peak VO2 - 15,4 ml/kg*min, AT - 7,0 ml/kg/min, VE/VCO2 - 39,9. In echocardiography right ventricle (RV) enlargement was observed, RV free wall was 11mm, left ventricle ejection fraction 55%. Moreover VSD 11 mm was present with maximal gradient of 32 mmHg, dilated ascending aorta, hypoplastic pulmonary trunk – 7 mm, pulmonary valve gradient of 5/3 mmHg, mild regurgitation. Visible shunt between left subclavian artery and left pulmonary artery, visible accessory vessel originating from the aorta.

Angio-CT showed stenosis of RVOT, pumlonary valve – 20x14 mm, pulmonary trunk over the valve – 13x11 mm, right pulmonary artery – 15x18 mm, stenosis of middle segment of left pulmonary artery 13x14 mm, than dilated 28x22 mm. Right aortic arch, ascending aorta: 41x40 mm, no abnormalities of coronary arteries were detected.
carvedilol, cilazapril, spironolactone and atorvastatin were prescribed.

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DISCUSSION

Tetralogy of Fallot (ToF) is a rare, cyanotic, complex heart defect which occurs in about 5 out of 10000 newborns. It affects boys and girls equally and is more common in patients with Down Syndrome or DiGeorge Syndrome. ToF comprises of ventricular septal defect (VSD), pulmonary stenosis, right ventricle hypertrophy (RVH) and overriding aorta. This heart defect is most often diagnosed in infants or young children and surgically treated in early childhood. There are two major types of surgery. In classical form of ToF children are operated on between 3-6 months of life. The complete repair consists of VSD closure and RVOT obstruction widening. In case of pulmonary artery hypoplasia, shunt operation is performed. Some patients have shunt operation in childhood and never undergo complete repair. Nevertheless they may still be able to undergo complete repair later in their lives.

In ACC/AHA 2008 Guidelines for the Management of Adults With Congenital Heart Disease complete repair is considered palliative treatment in patients without irreversible PAH or unfavorable pulmonary artery anatomy and as a primary operation, usually performed in the first year of life. An adult who has undergone palliation earlier in life can be considered for surgery for complete repair after thorough evaluation indicates favorable anatomy and hemodynamics. For the unusual cases of patients with tetralogy of Fallot who has undergone palliation with a surgical shunt, catheterization should be performed to assess the potential for repair. The presence or absence of additional muscular VSDs may be determined, as well as the course and anatomy of the epicardial coronary arteries. The pulmonary architecture and vascular pressure and resistance should be delineated, because pulmonary artery distortion and PAH are frequent consequences of palliative surgical shunts. Potential catheter interventions include elimination of collateral vessels or systemic–pulmonary arterial shunts, dilation/stent implantation of obstructed pulmonary arteries, and, more recently, the possibility of percutaneous pulmonary valve implantation

Patients with unrepaired ToF are usually cyanotic, after surgical correction many patients are symptom-free or face late problems. Those are: pulmonary regurgitation, right ventricle outflow tract (RVOT) obstruction, arrhythmias, residual VSD, sudden cardiac death (SCD), infectious endocarditis. Some patients need repeated surgeries during their lives due to those complications. Patients with ToF require regular lifelong follow-up in centers specialized in adults with congenital heart disease

EXPERT'S OPINION

In this patient surgical treatment seems possible. Prior the final qualification right heart catheterization in order to evaluate cardiovascular hemodynamics is necessary. Coronary angiography should also be performed in order to evaluate coronary circulation. He is a heavy smoker and abuses alcohol. The patient should be informed about the extension of the operation, its technique, risks and expected results. Prophylaxis of infectious endocarditis is an important issue.

CONCLUSION

Surgical correction after evaluation of cardiovascular hemodynamics in the cardiac catheterization should be considered.

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