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 present a case of a 31 year old female patient with complex congenital heart disease diagnosed as a common truncus arteriosus with ventricular septal defect after systemic-pulmonary anastomoses with (systemic-to-pulmonary shunts), who was admitted due to the exacerbation of symptoms (NYHA class IV) of chronic heart failure, and suspected endocarditis. Congenital heart disease was recognized at birth and confirmed by cardiac catheterization at the age of 2 years. On admission, the patient body weight was 48 kg, height 160 cm, the pressure of 90/60 mm Hg. Significant scoliosis of the thoracic spine to the left, signs of central cyanosis, clubbed fingers were observed. There was a systolic murmur in the second right intercostal space. Laboratory tests revealed slightly elevated total cholesterol - 253 mg/dL (normal <200), LDL -179 mg/dl (normal <100 m/dl), HDL - 60 mg/dl (normal range <45 mg/dl) elevated level of CRP - 35 mg/l (normal range 0.08-3.1mg/l) mild hyperbilirubinemia - 1.38 mg/dl. Blood cultures were negative. TEE found a common truncus arteriosus (diameter 50 mm), moderate tricuspid regurgitation. The global and segmental left ventricular systolic function was normal (EF - 65%). Sings of left ventricular hypertrophy were present. Perimembranous VSD of a diameter of 20 mm coexisted. There was no vegetation valvular or wall endocardium. In chest X-ray large chest wall deformity prevented an accurate assessment of the size of the heart, vascularity and bronchoalveolar fields of the lungs. Empirical treatment with Amoxycyllin and Gentamicin was used achiving an improvement of the general condition and reduction of the severity of heart failure (NYHA IV to III), including the reduction of CRP from 35.3 to 11.1 mg/l). CMR imagining revealed a wide (recognized as common) truncus arteriosus (diameter of 55 mm) with a large perimembranous VSD, enlargement of the right ventricle (tricuspid valve ring was 52x55 mm - standard 18-33 mm, with a score / + / 4.35). There were two wide vessels originating from the aorta leading towards pulmonary hilus, which were interpreted as collateral arteries. Treatment with beta blocker and spironolactone was initiated. In the course of hospitalization the patient's condition stabilized (NYHA III), NTproBNP - 191 pg/ml (N: 125pg/ml). 6MWT

showed impaired exercise tolerance: 6MWD - 235 m, Sp02 of 80% → 65%. Lung function tests showed a decrease in FVC (1.38 l – 38 of predicted value), FEV1 - 0.94 liters - 30% of pred. value), PEF - 2.22 l - 32% of normal range). On the basis of CTA there was a suspicion of common arterial trunk with pulmonary arteries arising from the descending aorta without stenosis. Furthermore, there was a suspicion of abnormalities of the coronary arteries origin. Venous anomaly was also shown (venous plexus conglomerate passing to the IVC). Angiocardiographic image in cardiac catheterization was interpreted similarly to the result of CTA. ie. common truncus arteriosus with ventricular septal defect and pulmonary arteries arising from the ascending aorta. The patient was discharged recommending Bisoprolol 2.5 mg once a day and Spironol 25 mg once a day. Due to the diagnosis of systemic pulmonary hypertension the use of bosentan was recommended within a national program for the treatment of PH.

DISCUSSION

Performed evaluation, especially CMR, Angio CT and cardiac catheterization indicate the presence of, in this case, so-called type IV, meaning pseudotruncus arteriosus by Collet and Edwards Classification (1). In type I of this classification there is a segment of the pulmonary trunk in the common trunk, from which both pulmonary arteries arise, in type II there is a lack of this segment and two pulmonary arteries arise very close to each other from the ascending part of the common trunk, in type III similarly, but both pulmonary vessels go far from each other. Type IV is actually a variant of pulmonary atresia with ventricular septal defect (PA,VSD) and the pulmonary circulation supplied by truncus arteriosus and/or collateral vessels (collaterals) from a variety of sources (2). In some patients with PA VSD flow into the pulmonary artery with preserved confluence is done by patent truncus arteriosus. In others collateral vessels may arise directly from the aorta, including subdiaphragmatic, from the brachiocephalic trunk arteries, thoracic, bronchial, or even coronary arteries. In these patients, the true pulmonary branches are typically either underdeveloped or even absent. Proper and early diagnosis is very important because in a real common arterial trunk surgical treatment is completely different than in pseudotruncus arteriosus, prognosis is radically different depending on the conditions of the inflow to the pulmonary circulation. In diagnosis at an early stage important issues include identification of sources of inflow to each lung and an explanation of how many broncho-pulmonary segments can communicate with real pulmonary arteries. You should also determine what are the sources of vascularization of each broncho-pulmonary segment. Apart from the different sources of inflow, pulmonary circulation is characterized by variability with age (as a result of angiogenesis, the formation with time of widening or narrowing of existing and emerging after birth collaterals). This affects the condition and the clinical course of patients presenting various symptoms from extreme hypoxia due to reduced pulmonary flow to heart failure due to increased flow and pulmonary hypertension.More detailed discussion of the nature of the collateral circulation and its importance in this defect is beyond the scope of this review. In the present case there has been a development of changes determining the increased flow and hypertension in pulmonary circulation. Therefore, I consider the decision to introduce bosentan as appropriate. There are known, but very rare cases of survival of patients with PA, VSD and developed collateral circulation without treatment up to the age of decades. The presence of

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PA, VSD is rare, estimated 0.07 - 0.1 / 1000 live births and 1.09% of all congenital heart defects (3). In Polish studies of Markowa et al. (4). the occurrence of this defect was estimated to be 0.66%, ie. 3.77 / 100 000 live births. There is a frequent coexistence in the constellation of other abnormalities, including CATH 22 syndrome (23%-40%). There was a concept of the relationship between aorto-pulmonary collaterals, narrow central pulmonary arteries and deletion 22q11 (5). A practical classification of the type of PA, VSD was proposed by the Congenital Heart Surgeons Society. In type A according to this classification there are own branches of the pulmonary artery and the source of vascularity is ductus arteriosus, there is no collateral circulation, in type B there are native pulmonary artery branches with various degree of malformation and the arteries of collateral circulation, while in type C there are no native pulmonary branches and pulmonary vasculature source are only collateral vessels. (6).

Surgical treatment of type B and C is similar, apart that unifocalization of collateral vessels is more extensive. The majority (65%) of children with the defect are treatment during early-infancy. Others are admitted later due to the developed collateral circulation and lack of visible cyanosis. The predominant symptom in about 50% is central cyanosis, in 25% heart failure symptoms, in 25% heart murmur, usually with impaired physical development (7). Surgical treatment of PA, VSD is difficult and multi-stage. In extreme cases of hypoplasia or absence of pulmonary branches surgical correction may not be possible. Then, with an excess of collateral vessels and after a their careful assessment it is possible to close them by intervention.

In neonates with PA, VSD, after determining the type of defect and recognizing that the source of the pulmonary vasculature is patent ductus arteriosus it is necessary to initially apply PGE1 (Prostinu). After evaluating the diameter of the pulmonary arteries modified Blalock-Taussig shunt is performed allowing for a steady supply of blood to the lungs, the growth and development of the pulmonary branch. In the next stage, after a thorough angiographic evaluation of sources of pulmonary vasculature it is necessary to restore the outflow tract of the right ventricle to the pulmonary circulatory system, while leaving the interventricular defect. Larger vessels constituting the only blood supply to the lungs require combining (unifocalization) with the pulmonary artery. In case of double flow from collateral vessels and pulmonary artery, abnormal vessels are closed by intervention (smaller - by means of suitable springs (coils), greater using special plugs Amplatzer - Amplatzer vascular plug) or intra-operatively (8).

In the next step after thorough angiocardiographic evaluation, subsequent unifocalization procedures are performed. With sufficient width of the pulmonary arteries ventricular septal defect can be closed, usually with a perforated patch allowing for temporary decompression of the right ventricular pressure, if the resulting resistance in the pulmonary circulation is excessive. Treatment is therefore phased and requires each time assessment of the pulmonary circulation, the inclusion of a segment for proper perfusion and potential safe elimination of collateral vessels. It is believed that the correction is possible when the Nakata indicator, with a closed collateral vessel circulation is >150mm2/m2.

Postoperative complications of this defect are common. These include stenosis and regurgitation of the RV/PA conduit due to calcification and degenerative changes, RV afterload due to conduit and pulmonary a. branches narrowing, vasoconstriction of unifocalized collaterals, pulmonary branches hypoplasia, tricuspid valve regurgitation due to

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RV dysfunction, LV deficiencies due to volume overload associated with pulmonary blood flow and/or aortic valve regurgitation, endocarditis, supraventricular arrhythmias (atrial due to right heart failure., and also but less frequently ventricular arrhythmias.
In patients undergoing palliative treatment or non-operated cyanosis increases with age as a result of developing collateral circulation. Pulmonary hypertension may be limited to certain segments of the lung. Stokes may occur as a result of paradoxical embolism. Repeated surgery or intervention is required that involve 10-15% of patients within 20 years. The risk of pregnancy is dependent on the type of pre-and post-operative changes. The most common are right heart failure and arrhythmias. RV-PA conduit stenosis, or branch narrowing increase the risk of cardiac insufficiency and arrhythmia.
In this case, probably at the age of two years when performed cardiac catheterization revealed changes, which at that time did not allow to take unifocalization treatment. Over time, there has been a development of increased flow and consolidation of pulmonary hypertension.
In the material of Amarka K et al. (9) out of 206 pts in the observation period from 1975 to 2000 84% were treated surgically. There was no treatment in 32 (16%) patients, of whom 21 died. Various procedures involved 180 patients of whom in 62% of cases the repair operation was possible.

EXPERT'S OPINION

In the presented case, there was the diagnosis of a type of common arterial trunk, which is not correct. In fact, it is a variant of the interventricular septal defect with pulmonary atresia and developed collateral circulation, which became the reason for the development and consolidation of pulmonary hypertension. In this case, the patient should be considered and treated as a patient with Eisenmenger's syndrome, which also has been implemented.

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