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 patient with complex heart defect and functionally single ventricle after classical Fontan procedure, after reoperation using modified Fontan method, bodyweight: 54 kg, height 164 cm, BMI: 20.07 kg / m2. Patient non-smoker, not abusing alcohol, no risk factors for coronary artery disease. Type of defect defined at birth as: D-TGA, TA, VSD, with probably (no mention of this) associated pulmonary stenosis and right ventricular hypoplasia. Additionally cleft lip and palate were found as well as inguinal hernia which was later treated by elective surgery. In the first stage, Blallock-Taussig shunt was performed. In the next stage, after 3 years classical Fontan procedure was performed. Due to the diagnosis of the ductus arteriosus, it was closed, but not until 8 years after that surgery. Another modified Fontan procedure was performed three years later with the simultaneous removal of thrombus from the left part of the (common?) atrium. After another 3 years hepatic veins were transplanted into the right atrium. The patient was repeatedly hospitalized due to hypoxia and cyanosis, supra- and ventricular arrhythmia episodes and symptoms of heart failure (NYHA III). The patient also had hepatitis B. Arrhythmia was treated using pharmacotherapy and ablation which reduced ventricular arrhythmias. After treatment with amiodarone iatrogenic hyperthyroidism occurred. Anamnesis revealed melena and an episode of bleeding from the upper and lower gastrointestinal tract. On admission, the patient's condition was defined as fair. Chicken chest, scoliosis, cachectic physique were found. Heart rate was accelerated, irregular 90 bpm, with pulse deficit. 115/80 mmHg Sa02 - 86-90%. In the bottom of the left sternal border a systolic murmur was found - 3/6 by Levine scale. Varicose veins of the lower limbs, bilateral lower leg trophic lesions, clubbed fingers

were present. The results of laboratory tests were normal except for moderately elevated pBNP - 1352 pg / mL (n <125.0). ECG revealed atrial fibrillation with an average heart rate of 85 bmp, isolated ventricular extrasystoles and left ventricular hypertrophy. In Holter monitoring signs of atrial fibrillation were observed with an average ventricular rate of 87 bpm, and isolated premature ventricular extrasystoles (1161), including pairs. In ECHO extended IVC (39 mm) unreceptive to breathing, enlarged common atrium (87cm2) with suspected thrombus, enlargement (67/77 mm) of functionally single left ventricular, EF - 45%, non-restrictive VSD, residual right ventricle, from which aorta originated were found. Mitral valve leaflets were fibrotic, ring diameter 61 mm. There was a significant mitral valve regurgitation (gradient of 5.8 / 3.9 mmHg. Aortic valve was insufficient – moderate regurgitation (gradient of 2.3 / 1.7 mmHg). Ascending aorta was dilated - 42 mm, bulb - 47 mm, ring - 35 mm. Cardiopulmonary exercise test revealed significantly reduced exercise capacity. The test was discontinued after 13 minutes. The patient has reached 7.4 METs and 69% of the maximum heart rate (189 bpm). During the test Sa02 slightly decreased from 70% to 68%. In plethysmography mild impairment of ventilation of restrictive type and increased airway resistance were found. Chest radiography revealed mild curvature of the spine to the right, signs of increased arterial vascularization. The right dome of the diaphragm was higher than the left. Abdomen ultrasound revealed dilation of IVC and hepatic veins, enlarged liver with sings of periportal fibrosis. The patient was treated with Acenocoumarol, diuretics (Spironol, Furosemide), Metizol, Digoxin and Sotahexal due to arrhythmia.

DISCUSSION

Congenital heart defects with functionally single ventricle include the spectrum of rare, usually very complex congenital heart disease, for which the only treatment option besides heart transplant is ultimately Fontan procedure (1). The authors present a case of tricuspid atresia with transposition of great arteries type D, with VSD and possibly stenosis of the pulmonary valve (typ. II B tricuspid atresia – there are no details in the text about the coexistence of stenosis/atresia of pulmonary valve). In infancy in this patient Blallock-Taussig shunt was performed. In the next stage, a few years later the classic operation of Fontan was performed without prior Glenn or hemi-Fontan procedure. In the eighties of the last century it was the method of choice. However, the hemodynamic performance of the classic Fontan (anastomosis of the right atrium or its appendage with the pulmonary artery) was unsatisfactory. This technique favored venous congestion, the occurrence of thromboembolic incidence and arrhythmias resulting in sudden death (2). Therefore, it was modified over time. Gentles TL. et al., in an analysis of 500 consecutive patients with a variety of CHD and CPK operated in a single-center shown that mortality during the initial period of 27.1% fell to 7.5%. The most important risk factors were: the mean pulmonary artery pressure ≥19 mm Hg, heterotaxy syndrome, CPK of the morphology of the right ventricle, pulmonary artery distortion, primary surgical technique (atrio-pulmonary anastomosis), lack of fenestration, CPB time and cross CLMP. The conclusion was that the decrease in mortality over time was partially due to modification of the method of treatment (stages) and type of final procedure (3,4). Long-term pulmonary Blalock-Taussig shunt performed in this patient during the early-infancy and patent ductus arteriosus (closed according to the documents much later) could be the cause of excessive flow and increased

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pressure in the pulmonary circulation. However, there is no hemodynamic data on this subject in the history of the presented case. We should also add that the classic Fontan procedure (and currently used technique of inside or outside atrial tunnel differ significantly reducing the risk of venous thromboembolic events, atrial arrhythmias, as well as the degree of venous congestion (2). Advances in surgical technique and intensive therapy resulted in a significant reduction in mortality between stages (after the 1st 5-25% after the 2nd 5-10% and 5% after the final procedure). Interestingly, it was shown that the mortality rate in the short term after treatment using the old and the present technique differ significantly. In contrast, long-term survival is not significantly different (5). The main causes of late death are still: blood clots, heart failure and sudden cardiac death due to various reasons, largely because of arrhythmias (5). The patient experienced major complications associated with Fontan type circulation, with the exception of exudative enteropathy and *bronchitis plastica*. Prior to determining the possibility of further treatment and its types, including a possible heart transplant, it is necessary to carry out hemodynamic and angiocardiographic evaluation. The liver state is not clear with a history of hepatitis B. It is known that the Fontan type circulation promotes hepatic dysfunction, cirrhosis, and increase the risk of cancer, which in this case also requires appropriate evaluation, including biopsy (6).

EXPERT'S OPINION

This case concerns a patient with type II B atresia of the tricuspid valve (D-TGA, VSD, the RV hypoplasia and pulmonary stenosis presumably), treated at first using pulmo-systemic anastomosis of Blallock-Taussig shunt and a few years later operated using a classical Fontan procedure. In the further course it turned out that the patient had a patent ductus arteriosus, which was closed much later. Long term volume overload caused by anastomosis and leaving patent ductus arteriosus could affect the dysfunction of CPK. Late made classic Fontan procedure was subsequently replaced by the modified version without fenestration. In the presented case some of the data, such as mentioning the size of the common atrium or the hepatic vein transplantation suggest that one can think of the heterotaxy syndrome. Abdominal ultrasound has demonstrated however the presence of the spleen, but its presence or absence is not 100% proof of the occurrence of this abnormality. No information on the construction of the atrium appendages, as well as on the morphology of the lungs (bilateral three-lobes?), prevents an unambiguous classification of the identified lesions. The patient experienced most of the complications observed in patients 15-20 years after the Fontan procedure (increasing hypoxia and cyanosis, thromboembolic complications, supra- and ventricular arrhythmias, a decrease in exercise capacity and heart failure). There was no exudative enteropathy. However, this complication can occur even many years after the Fontan procedure. The patient was treated adequately to the identified changes. In my opinion to determine further management in-depth diagnosis is required including hemodynamic and angiocardiografic evaluation, at least MRI. In the treatment of heart failure Sildenafil may help, which beneficial effects on exercise capacity in children and young adults after Fontan has been demonstrated (7). It has not been determined till now what treatment should be used in the prevention of thromboembolic complications in patients after Fontan procedure (8). It should be noted that in case of no further options of pharmacotherapy referral to a heart transplant is required. Such therapy becomes necessary in

about 10% of patients 10 years after surgery, but 30% after 25 years (9).

CONCLUSION

Presented case of a patient with congenital heart disease: tricuspid atresia type II B (TA, D-TGA, VSD, PS) with a functionally single left ventricle illustrates the diagnostic and therapeutic problems in adults after final Fontan procedure. Advances in treatment resulted in a significant reduction in mortality in these patients and prolonged their survival. Still, there are factors resulting from the presence of only one ventricle of the heart and the nature of Fontan type circulation affecting increasing over time risk of death due to progressive cardiac dysfunction and systemic disorders (hypoxia, the development of collateral circulation, thromboembolic complications, supraventricular arrhythmias, dysfunction and liver cirrhosis. There may be iatrogenic disorders of thyroid function after treatment of persistent supraventricular arrhythmias with amiodarone, which also took place and was properly treated.

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