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 31-year-old man with a complex, congenital heart disease diagnosed in the early childhood (transposition of the great vessels, tricuspid atresia, VSD, right ventricular hypoplasia). He has a history of multi-stage surgical correction: the Glenn shunt procedure, the Fontan operation, surgical PDA closure, reoperation after the Fontan Procedure with conversion to TCPC Fontan, transplantation of hepatic veins to the right atrium. He Has been hospitalized many times due to heart rhythm disorders, severe cyanosis and symptoms of heart failure. On admission to the Clinic the patient reported chronic fatigue, reduced exercise tolerance (NYHA class III) and recurrent respiratory tract infections. Physical examination revealed precordial systolic murmur, skin and mucous membranes cyanosis, clubbed fingers, varicose veins on both extremities, scoliosis and pectus carinatum. There were no signs of peripheral oedema and the liver edge was not palpable. Weight 54 kg, height 164 cm, BP 115/80 mmHg, HR- irregular of about 90 bpm, peripheral pulse not synchronized with heart rate, the blood saturation was 86% (on room air). Medical history: radiofrequency ablation (RFA) of atrial fibrillation and ectopic focus in anterior ventricular wall (04.2010), thrombocytopenia, secondary polycythemia, history of hepatitis B, amiodarone induced hyperthyroidism, history of upper and lower gastrointestinal bleeding episodes. The ECG revealed atrial fibrillation with ventricular rate of 90 bpm and ventricular ectopic beats. First Holter monitoring (2010) (pre – radio frequency ablation)showed atrial fibrillation with ventricular rate of 62 bpm (max-94 bpm, min 49 bpm), ventricular ectopic beats (1164), episode of VT lasting tree seconds with maximum rate of 150 bpm in the evening, episodes of bradycardia of 37 bpm at night. Post - radio frequency ablation Holter monitoring showed atrial fibrillation with ventricular rate of 87 bpm and 1161 ventricular ectopic beats without episodes of VT. Biochemical analysis showed high level of NT-proBNP- 1352 pg/ml (<125), Total bilirubin- 60,4 umol/L (<21,0), RBC- 6,03*10^6/ul (4,2-

6,00) and low level of platelet count- 71,0 K/uL (140-440). The transthoracic echocardiography revealed severely enlarged common atrium (85 cm2), severe AV-valve regurgitation assessed by vena contracta width (VCW)- 10mm, dilated AV-valve annulus diameter- 61mm, enlarged systemic ventricle with diameters 67x77mm, EF 45%, mild aortic regurgitation; IVC- 39 mm, no collapse on inspiration. The cardiopulmonary exercise testing (CPET) showed reduced exercise tolerance and a reduction in maximal oxygen consumption (VO2 max 19,2 ml/kg/min); carbon dioxide production (VE/VCO2 slope) was 38.6. We performed CT angiography which confirmed severely enlarged common atrium measuring 70×140 mm, large ventricular septal defect measuring 38x24mm. The anastomosis between the vena cava superior and the pulmonary trunk is patent. Moreover the dilated coronary sinus is connected with the common atrium (shunt). Bodypletysmography showed mild restrictive lung dysfunction and increased airway resistance. Abdominal ultrasound showed hepatosplenomegaly, abnormal liver with irregular contours, varying echogenicity and periportal fibrosis, dilatation of hepatic venous confluence and a small amount of fluid in the peritoneal cavity. Currently the treatment is: anticoagulants (LMWH), spironolactone 25 mg, furosemide 40mg, thiamazole 2,5 mg, digoxin 0,1 mg, sotalol 80 mg.

DISCUSSION

The Fontan operation, although without doubt a life-saving surgical strategy, is related to a variety of organ complications and unique morbidities that are recognized with increasing frequency as patients life-span increase. They most often manifest themselves in the second and third decades of life and beyond. Liver fibrosis, protein-losing enteropathy, chronic renal insufficiency, plastic bronchitis, chronic venous insufficiency and development of dangerous cardiac arrhythmias are consequences of a complex pathophysiology involving circulatory failure, inflammation and lymphatic derangement. These conditions often develop covertly over a long period of time. A better characterization of systemic long-term consequences of the Fontan circulation is necessary in order to develop specific methods of treatment.

EXPERT'S OPINION

At the moment the patient is disqualified from surgical treatment, because of very high operational risk (heart failure, hepatic cirrhosis, high risk of bleeding, chronic venous insufficiency, atrial fibrillation, hyperthyroidism. Close follow-up and optimal conservative treatment is necessary (adequate rehabilitation, regular laboratory tests, high protein diet, anticoagulation, diuretics and antiarrhythmic therapy.

CONCLUSION

At present due to very high surgical risk conservative treatment is recommmended. Close follow-up and regular check-up examinations are necessary.

REFERENCES

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