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

There were a case study of 46-year-old woman with symptoms of heart failure (NYHA I/II) and distal mild cyanosis. The medical history revealed a complex congenital heart defect (double-inlet ventricle; DIV) diagnosed early at the infancy, but without palliative surgery or radical repair. The patient was leading a near normal life doing all the house work and doing a physical labour. She had three healthy children delivered through a C-section without complications. The major routine laboratory tests abnormalities were: Hb-19.85g/dl, Ht-61.30%, TSH-7.27uIU/ml, FT4-12.97pmol/l, NT-proBNP-428 pg/ml. The performed six minute walk test was used to assess functional capacity showed a distance of 340 m with a desaturation (75 to 51%). Transthoracic echocardiography and cardiac magnetic resonance imaging showed a double-inlet left ventricle of normal systolic function with large septal defect and rudimentary right ventricle, d-transposition of great vessels and both pulmonary valve and supravalvular stenosis. There was also a mild-to-moderate regurgitation of both atrio-ventricular valves. The right heart catheterization procedure failed to directly assess pulmonary arterial pressure due to technical problems.

DISCUSSION

The double inlet left ventricle is rare congenital heart diseases. The right ventricle is small and both the mitral and tricuspid valve open into the enlarged left ventricle, This complex heart malformation is included into group of single ventricle (SV). The term describes a heart malformation associated with a functional SV chamber, the other being rudimentary, or - due to the absence of the interventricular septum – consisting of two ventricles forming one chamber. The Fontan operation (FO) has become the treatment of choice in patients with the single ventricle (1). The purpose of the operation in patients with SV is normalization of the volume load of the SV and separation of the pulmonary and systemic circulation to achieve a normal or near normal level of blood oxygen. The SV pumps blood into the systemic circulation through the aorta, whereas the systemic veins empty directly into the pulmonary artery. Univentricular heart is a severe congenital cardiac defect; 50% of patients are dead 14 years after diagnosis, a death rate of 4.8% per year. The most common causes of death were dysrhythmia, congestive heart failure and sudden and unexplained death (2). Adult patients with double-inlet left ventricle and perfectly balanced circulation may survive into the sixth decade with good functional capacity and preserved ventricular function. This should be considered before such patients are referred for a Fontan repair (3).

EXPERT'S OPINION

The patient with unoperated complex heart defect with normal single ventricular function, with no significant arrhythmia, with balanced circulation and good functional capacity should be qualify for clinical observation.

CONCLUSION

The patient should be qualify for systematic observation in centre specialised for adult with congenital heart diseases.

REFERENCES

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