

Family form of the restrictive cardiomyopathy: clinical case

Slastnikova E.S.(1,2), Sadykova D.I.(1), Khabibrakhmanova Z.R. (1), Sabirova D.R. (1,2,3), Firsova N.N. (2), Kucheryavaya A.A. (2)

Kazan State Medical University (1); Children's Republican Clinical Hospital of the Republic of Tatarstan Ministry of Health (2); FSBEI HE Kazan SMU MOH Russia (3)

The aim is to optimize the early diagnostics of the restrictive cardiomyopathy, the family form.

Materials and methods: the description of the clinical case of the restrictive cardiomyopathy, the family form.

Results: the child K., 3 years old, was born from the first pregnancy with the risk of pregnancy interruption. The routine prenatal ultrasound examination didn't detect any fetal anomalies. The child arrived on the due date, from the "classic" vaginal delivery. In the family history: the child's grandmother died from the restrictive cardiomyopathy with the atrial fibrillation at the age of 45, the child's uncle died from the restrictive cardiomyopathy at the age of 16. The child's father is now 30 years old. His diagnosis is "Restrictive cardiomyopathy. Atrial fibrillation".

The child underwent echocardiography at the age of 19 days, no abnormalities were found. At the age of 8 months the echocardiography examination showed the left atrial dilatation. At this time the electrocardiography data revealed the both left and right atrial enlargement. At the age of 23 months the child was first hospitalized to the Children's Republican Clinical Hospital with the complaints of small increase in body weight, fatigue, exercise-induced dyspnea. Clinical examination showed paleness of skin integument, stunted physical growth, labored breathing, tachypnea up to 50 per minute, hepatomegaly.

Echocardiography examination showed both left and right atrial dilatation, inferior vena cava and hepatic veins distension, the restrictive form of both left and right ventricles diastolic dysfunction. Electrocardiography examination revealed biatrial enlargement, incomplete right bundle-branch block, ST-T changes. The ultrasound liver examination showed hepatomegaly and hepatic veins dilatation. The NT-proBNP level is significantly increased - 4000 pg/ml. Taking into consideration the family history, the patient was diagnosed with restrictive cardiomyopathy, the family form, NYHA II-III. The standard therapy of heart failure was prescribed. The genetic diagnostics in Moscow Medical Genetic Scientific Centre revealed the mutation c.4631C>A in gene FLNC, mutations in TTN, and the diagnosis was confirmed.

Conclusions: knowing the family history and genetic diagnostics data is incredibly important for the early diagnostics of the family form of the restrictive cardiomyopathy and for the early heart failure treatment.