Results We revealed nucleotide missen VUS: c.1259A>G, p. Q420R and c.2051T>C, p.I684T in the heterozygous state in LZTR1 gene. All variants were absent in HGMD professional and genome aggregation database.

Conclusion Child with severe hypertrophic cardiomyopathy and typical phenotype of Noonan syndrome was detected NS 2, caused by compound heterozygous missense variants c.2051T>C, p.I684T and c.1259A>G, p.Q420R in LZTR1 gene.

Tuberculous sclerosis complex (TSC) is a relatively rare autosomal dominant disorder characterized by hamartomatous lesions in a variety of organs.

Cardiac involvement remains within major criteria for diagnosing the syndrome, as it can initiate the TSC evaluation for some patients. Cardiac involvement is usually maximal at birth or early infancy and shows signs of regression during the first two years of life. The aim of this study was to evaluate the echocardiographic findings in our patients with tuberous sclerosis complex.

We present a case series of five patients diagnosed with TSC in our hospital within the last ten years. All patients underwent a complete cardiac examination at the time of diagnosis (clinical findings, electrocardiography and ultrasound examination of the heart) and were further monitored through controls.

All patients had a normal clinical findings. One patient had nonsignificant supraventricular and ventricular extrasystoles. Abnormal echocardiographic findings were seen in four of total five patients. Two patients had both atrial and ventricular cardiac masses with echo characteristics that raised suspicion of rhabdomyoma. Two other patients had multiple focal areas of increased intramyocardial echogenicity. There were no signs of outflow tract obstruction or impaired valvular function. Left ventricular size and function were normal. Close follow-up showed no signs of significant regression or progression in our echocardiographic findings.

The prognosis and growth potential of these masses are not known, but can be determined by longitudinal follow-up. Cardiac ultrasound should be considered for all patients with TSC regardless of physical findings.

We present a case of 14 year old boy after heart transplantation and care due to Uhl's anomaly diagnosed five years ago. Five years before this admission, child experienced exertional dyspnea and vertigo at school.

During transport to the hospital, child arrested and after a short resuscitation and defibrillation (initial rhythm VF), sinus rhythm occurred.

On the initial transthoracic echocardiography, dilated cardiomyopathy of right ventricle was observed. After the initial stabilization, MR was performed (May 2014.), where marked dilatation and impaired function of both right atrium and ventricle was noticed. The volume of right ventricle in diastole (EDV) was 245 ml (188ml/m2) with ejection fraction of RV of 19%.

The RV wall was thin, with almost complete absence of right ventricular free wall myocardium, without fat infiltration. Control MR, performed after one year, showed a progression in dilatation of right ventricle dimensions with impressive tricuspid regurgitation. The left heart structures, functionally, and morphologically, were within normal range. Differential diagnosis yielded two possibilities: Uhl's anomaly and arrhythmogenic dysplasia of right ventricle (ARVD). The recommended biopsy, as well as installation of implantable cardioverter defibrillator, were rejected by his parents. The metabolic diseases were excluded. From June 2018, child was dyspneic in everyday activities, had peripheral edema, was tired and sometimes complaining off tingling in the legs. After two rejections from his parents, heart transplantation was done in January 2019. Pathohistological diagnosis of Uhl's anomaly was confirmed. One month later, he was discharged from the hospital.

We give a question of differentiation between Uhl's anomaly and ARVD. In ARVD, the myocardium is progressively replaced by fibrofatty tissue, and has been linked to mutations in the genes encoding plakoglobin and desmoplakin.

Patients with Uhl's anomaly rarely survive to adulthood. There are two main surgical approaches for these patients; first one is based on the relief of right ventricle volume or excluding the entire right heart from circulation. This can be done by making total cavopulmonary connection, by closing tricuspid valve and making bidirectional Glenn procedure, or making a one-and-a-half ventricle repair with plication of the right ventricular free wall. Second option is the heart transplantation.