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HYPERTROPHIC MYOCARDIOPATHY IN CHILDREN

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Introduction. Hypertrophic cardiomyopathy (HCM) is considered to be the most common genetic heart disease, posing significant morbidity and mortality risks. This myocardial disease affects 1 in 500 adults and is mostly related to sarcomere genes mutations. Pediatric HCM exhibits greater heterogeneity without reliable epidemiological data available, It is estimated that 40-60% of cases are primary (mutations in sarcomere genes), 35% of cases are secondary - associated with inherited metabolic disorders (e.g., glycogen storage diseases), neuromuscular variants, malformation syndromes, and mitochondrial diseases.

Objective: to assess possible manifestations and increase awareness for pediatric HCM.

Materials and methods. The case history, family anamnesis was studied in a seven-month-old male with hypertrophic cardiomyopathy, followed at the 2nd Pediatric Department (for cardiological patients) of 2nd Minsk Children's Hospital in 2024-2025.

Results and their discussion. Lev M., born prematurely at 35 weeks, has been diagnosed with HCM marked by ventricular hypertrophy unrelated to abnormal load volume since birth. A routinely performed echocardiogram revealed predominantly right ventricular HCM with significant obstruction of right ventricular outflow tract (HSD 112-125 mmHg). A contrast-enhanced CT scan showed congenital heart disease, moderate pulmonary artery valve stenosis, boundary dimensions of the pulmonary artery trunk, and a small interventricular muscle defect. The ECG indicated a sinus accelerated rhythm (heart rate 175 bpm), right deviation of the electrical axis, and RA hypertrophy.

From family history it was found out that uncle of the patient (mother's side), Alexander T., born in 2001, at the age of ten years during investigation for syncope was diagnosed with nonobstructive HCM. He was prescribed metoprolol. Soon after diagnosis was made he died of SCA. Mother of the patient is healthy. Lev's aunt (mother's side), Anastasia, born in 2017, is followed by cardiologist for persistent Arterial Hypertension since 5 yo, however multiple EchoCG didn't reveal any signs of HCM. Lev's grandfather (mother's side) died at the age of 43. Patient's greate-grandmother (mother's side) died at her mid-40 from SCD. At 7 months Lev gains weight and doesn't progress with heart failure. His treatment includes anaprilin to control heart rate and relief the RVOT.

Given family history genetic testing was performed and storage diseases, microdeletion syndromes ruled out, but results of testing for cardyopanel and Noonan syndrome – pending.

Conclusion. Given the family anamnesis with males affected and females being carriers X-linked type of inheritance can be suggested but exact mutations is still to be identified. Early testing and investigation should be performed in close relatives of HCM patients for appropriate genetic counseling and targeted treatments for affected families.