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Case report: Leopard syndrome in a newborn with severe hypertrophic cardiomyopathy

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We present rare form of RASopathy and myotonic dystrophy 1 with familial incidence. Our patient is a boy born at gestational age of 38 weeks by cesarean section to polymorbid tertigravida tertipara. After delivery there were signs of perinatal asfyxia, the prompt resuscitation was started, due to the absence of spontaneous breathing activity endotracheal intubation was performed. APGAR score was 2-4-7, an umbilical cord pH was 7,24. The neonate had clinical signs of fetal hydrops (ascites, pleural effusion, skin edema), the other clinical findings were gothic palate and equinovarus feet. Cardiac examination at 1st day of life revealed extreme hypertrophy of interventricular septum, decreased systolic and diastolic function. The heart function as well as pulmonary hypertension was improving according to regular cardiac examinations, ventilatory support was terminated after 18 days. 21st day of life an attack of ventricular tachycardia occured in the newborn, terminated spontaneously. The beta blocker therapy was initiated, with no recidive of arytmia. Based on the progressive ventricular hypertrophy the prognosis of the baby was uncertain. Leopard syndrome and myotonic dystrophy 1 were diagnosed prenatally. The patient was dismissed at age of 2 months in stabilized health condition, due to the severe cardiomyopathy he received DNR.

Biography

Tereza Pomahačová has completed her MD at the age of 25 years from Charles University – Faculty of Medicine in Pilsen. Since graduation in 2017 she has ben working at Department of Neonatology in Faculty hospital in Pilsen. Supported by a Grant from Ministry of Health of the Czech Republic – Conceptual Development of Research Organization Faculty Hospital in Pilsen – FNPI 9608.

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