Cardiac involvement in paediatric patients with mitochondrial disease

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Introduction:
Mitochondrial diseases have a wide clinical spectrum, with multisystemic manifestations and can present at any age. Cardiac abnormalities are often present and include hypertrophic or dilated cardiomyopathy, heart conduction defects and ventricular pre-excitation. These manifestations may be late, subtle or can be the first presentation of the disease. The aim of this study was to review the cardiac manifestations and its impact in the outcome of the patients followed in a tertiary hospital.

Methods:
The authors made a retrospective analysis of clinical, laboratorial records and cardiac exams (EKG, Doppler echocardiography, Holter monitoring) of children with probable mitochondrial disease (MD), according to the modified Walker criteria (Bernier et al., 2002).

Results:
The study includes 22 patients, aged 1 day to 4 years at presentation of MD, with a median follow-up of 72 months (min 9 months, max 156 months). Eight patients (36%) had cardiac involvement, with a male-female ratio of 5:3. The cardiac diagnosis preceded the mitochondrial disease diagnosis in 2 cases presenting hypertrophic cardiomyopathy in the newborn period (one was asymptomatic but had left ventricular hyperechogenic foci in fetal ultrasound and the other developed respiratory distress syndrome and cyanosis after birth), while the other patients developed the cardiac abnormalities during follow-up (age of cardiac diagnosis varied from 7 days to 14,5 years). EKG abnormalities were found in all eight patients.

Five had primary myocardial disease: hypertrophic cardiomyopathy (n=5), and systolic dysfunction of the left ventricle (n=1); and two had conduction abnormality (atrioventricular block and WPW syndrome). One patient had implantation of pacemaker at the age of 14 years due to advanced atrioventricular block. Mortality was 12,5% in the cardiac group vs 7% in the non cardiac group.

Conclusions:
Cardiac abnormalities are often present in MD (36% of our children) but the patterns of heart involvement are very heterogeneous. They may occur as the principal clinical manifestation, may have an early or late onset, and carry a worse prognosis. The authors emphasize the need of a periodic cardiac evaluation and follow-up of these patients.