P-299

Asymptomatic hypertrophic cardiomyopathy and elevated troponin levels as first indications of Friedreich ataxia.

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INTRODUCTION:
Friedreich ataxia is a neurodegenerative disease with autosomal recessive heredity. Myocardial involvement usually occurs several years after the onset of the neurological symptomatology in adolescence and early aduldhood.
PURPOSE:
We present three cases with first manifestation of the disease the unusually inflamed myocardium.
MATERIAL AND METHODS:
Three children one boy and two girls, aged 10,12 and 11 years old girl underwent pre-athletic screening test by electrocardiography and echocardiography. Because of ECG and echocardiography findings, troponin levels were measured and cardiovascular magnetic resonance (CMR) was performed in all of them. Neurological evaluation and genetic tests followed.
RESULTS: All children were asymptomatic. The ECG showed T wave inversion in leads II, III, AVF, V4-V6 and LV hypertrophy using the Sokolow-Lyon index: SV1+RV6>3.5mV in all of them. Echocardiography showed moderate cardiac concentric hypertrophy, (intraventricular septum thickness/lateral wall thickness, IVS/LA: 12.66±0,57mm/ 12,5±0.7mm) with normal left ventricle without obstruction and normal right and left ventricular function. Measurements of hs-cTnT assay were elevated in all patients vs controls ($p<0.005$). The CMR tissue characterization revealed evidence for oedema, perfusion and fibrosis in all cases. Due to a slightly peculiar walking, neurological assessment was requested and a molecular genetic examination confirmed Friedreich ataxia. Children were referred to a neuromuscular disorders unit.
CONCLUSION: To conclude a combination of ECG and imaging parameters can reveal early FA-CM and motivate early risk stratification and start of cardiac treatment.