Assessment of cardiac functions in patient with Trisomy 21

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Introduction: Extra genetic material in Down syndrome (DS) may affect the function of any organ system. We wanted to evaluate cardiac functions using two-dimensional, M-mode and Doppler echocardiography even in absence of congenital and acquired heart disease in patients with Down syndrome.

Methods: 115 patients with DS between 6 and 13 years of age with clinically and anatomically normal heart and 55 healthy children were included in this cross-sectional. DS was diagnosed by a karyotype test. Patients with mosaic type were not included in this study. Systolic and diastolic functions were evaluated by echocardiography.

Results: Systolic pulmonary artery pressure was significantly higher in patients with DS than in healthy group (p<0.001). They had significantly higher left ventricular mass, ejection fraction, mitral annular plane systolic excursion values. Pulsed waved Doppler transmitral E (early diastolic velocity) /A (late diastolic velocity), tissue Doppler mitral annular Ea (early diastolic velocity)/Aa (late diastolic velocity), transtricuspid E/A and tricuspid valve annulus Ea/Aa, pulmonary venous Doppler S (systolic)/D (diastolic) wave ratio were lower in patients with DS than in control group (p=0.04, p=0.001, p<0.05, p<0.001, p<0.001 respectively). Mitral and tricuspid annular Ea were lower in patients with DS (p<0.001). The right and left ventricular myocardial performance index were higher in patients with DS than in controls (p<0.01).

Conclusion: Conventional and tissue Doppler echocardiography is important in determining the subclinic cardiac diastolic dysfunction in patient with DS. We think that these changes may be due to autonomic dysfunction, pulmonary hypertension, myocardial fiber structure.