Evaluation of the incidence and echocardiographic findings of congenital heart diseases in children with Down Syndrome in the Middle Anatolia of Turkey

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Objectives: Down syndrome is the most common chromosomal disorder in all countries and congenital cardiac disease is the greatest cause of mortality and morbidity in these patients. In the recent study, incidence and types of congenital heart diseases of children with Down syndrome were evaluated in the Middle Anatolia of Turkey.

Methods: Data of 239 patients diagnosed as Down syndrome who were consulted to our pediatric cardiology department between March 2007 and April 2011 were retrospectively reviewed. History, physical examination, echocardiographic and cardiac catheterization findings of all patients were listed. Chromosome analysis was performed in our genetic laboratory.

Results: Congenital heart diseases were detected in 86 (% 36) of 239 patients with Down syndrome. Congenital heart diseases were detected in 48 (%33.8) of 142 patients who were admitted during neonatal period while this ratio was % 39.2 (38/97) after neonatal period (p=0.041). Evaluation of all single or multiple cardiac defects revealed that endocardial cushion defect (% 37.2) was the most common congenital heart disease. During the study period only two patients died due to the cardiac operation. These patients were diagnosed as complete endocardial cushion defect in older ages and cardiac operation was performed with high risk.

Conclusions: In our study the prevalence of congenital heart diseases in neonates with Down syndrome was lower than the current literature. To determine the real prevalence of congenital heart diseases in neonates with Down syndrome, prospective and multicenter studies were needed in our country. Appropriate-time-planned cardiac operations will decrease the high incidence of mortality and morbidity in Down syndrome with large ventricular septal defect or endocardial cushion defect.

Key words: Down Syndrome, Infant, Newborn, Congenital Heart Defects