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Cardiovascular Abnormalities in Williams Syndrome; 20 Years' Experience in Istanbul

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Introduction: Williams syndrome (WS) is a microdeletion syndrome affecting cardiovascular and connective tissue as well as the endocrine and central nervous systems in 1 in 10,000 live births. This study aims to identify and evaluate cardiovascular abnormalities (CVAs) in 45 WS patients.
Patients and Methods: We retrospectively reviewed a cohort of WS patients that were followed at our institution from January 1, 1990 through December 31, 2010. WS was clinically diagnosed by an experienced medical geneticist and confirmed by fluorescence in situ hybridization. CVAs were assessed using electrocardiography, echocardiography or cardiac catheterization.
Results: Twenty-seven patients (60%) were male; 18 were female (40%). The mean age at presentation was 4.6±3.1 years (3 months-13 years); the follow-up period was 6.9±4.4 years (6 months-18 years). CVAs were found in 86% of patients, the most common one being supravalvar aortic stenosis (SVAS) in 73% (isolated in 48%), peripheral pulmonary artery stenosis (PAS) in 42%, and mitral valve prolapse (MVP) in 22%. Less common were aortic insufficiency (15%), ventricular septal defect (11%), valvular pulmonary stenosis (11%), and aortic arch hypoplasia (8%) and coarctation (2%). Hypertension was present in 22% of patients. Surgical or catheter-based interventions were performed in 22% of cases. Two patients were lost in the postoperative period.
Conclusion: CVAs were found in more than four out of five patients, the most common ones being SVAS and PAS. Although surgery was performed in more patients with SVAS than with PAS, SVAS was minimal or mild in most patients and improved in few cases.