CARDIOVASCULAR ANOMALIES IN TURNER SYNDROME

INTRODUCTION

Turner syndrome (TS) has several defects affecting different organs. Heart defects are the most common. They can be symptomatic (Heart murmur, high blood pressure) or diagnosed systematically. The chromosomal profile affects the nature of the anomalies encountered.

OBJECTIVE

Report cardiac abnormalities in TS

POPULATION, METHODOLOGY

This is a retrospective study of 60 TS patients identified in 20 years. Mean age was 16 ± 0.4 (3 months-17) They were given a complete physical examination, a cardiovascular evaluation (ECG, Doppler ultrasound and cardiac MRI) and a karyotype at diagnosis. Annual Cardiovascular revaluations (ECG, Doppler ultrasound) and at the slightest sign were made

RESULTS

Heart abnormalities are found in 45% of cases. The most frequent abnormalities are bicuspid aortic valve (30%) and coarctation of the aorta (10%). Interatrial communications , pulmonary valve or blood abnormalities ((stenosis and / or mitral disease (prolapse) were noted in 5% of cas. Cardiac abnormalities were present at diagnosis . Revaluations have noted the appearance of a hypertension in 10% associated with e hypercholesterolemia (6%) and diabetes mellitus in 4%. There was a genotype-phenotype correlation. Heart abnormalities are present in 70% of subjects with monosomies 45 X0; they are less common (15%) in case of chromosomal formulas mosaic. Aortic coarctation xas more common in formulas 45 X0, pulmonary stenosis in mosaics X0 45 - 46 XX

DISCUSSION

Fifty to 75% of Turner patients are carriers of cardiovascular defects. Congenital heart defects are described in approximately one third of patients, the most common are the bicuspid aortic valve (16%) and aortic coarctation (11%). However, cardiovascular complications can occur at any age (aortic aneurysm, dissection, hypertension) and warrants regular cardiac monitoring in childhood and specialized care. Turner patients mortality is three times that of the general population, cardiovascular complications are the main cause of early mortality with a life expectancy could be reduced by more than ten years.

A complete cardiovascular evaluation must be undertaken in search of a bicuspid or dilatation of the ascending aorta due to the high risk of aortic dissection.

The chromosomal profile affects the nature of the anomalies encountered. Congenital heart defects are present in 40% of subjects with homogeneous monosomies 45 X0 (against 0.8% in the general population); they are less frequent (about 11 to 22%) in case of chromosomal mosaic formulas . Aortic coarctation is more common in formulas 45 X0, and pulmonary stenosis in mosaics X0-46 45 XX. Finally, in the formula normocytic X (X chromosome the two arms are identical), we do not find heart defects but frequently isolated hypertension.

Turner women have an increased risk of atherosclerosis, exacerbated by the higher prevalence of hypertension, diabetes, dyslipidemia, obesity and estrogen deficiency. Thus, the risk of coronary artery involvement is multiplied by 2, and one stroke by a factor of 2.7. The higher frequency of diabetes mellitus affects cardiovascular outcomes (mortality 11 times higher)

In all cases, cardiovascular evaluations should be undertaken. They should include an annual cardiac examination with measurement of blood pressure, echocardiography repeated every three to five years, and an MRI if identified anomaly. Two-dimensional transthoracic echocardiography with color Doppler search aortic malformations (bicuspid aortic valve, coarctation of the aorta shape abnormalities) and any abnormalities in venous return and tracks aortic acquired diseases (aneurysms, dilatation). It serves as a reference test for future monitoring.

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