P1-383 Cardiovascular Anomalies and Association with Karyotypes in Turner syndrome in Taiwan: one medical center experience

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BACKGROUND

Turner syndrome (TS) is characterized by growth failure, primary ovarian failure, cardiac anomalies, and other anomalies. Cardiovascular abnormalities such as bicuspid aortic valve (BAV), coarctation of the aorta (CoA), aortic stenosis (AS), and aortic dilation (AD) account for some TS-related early mortality. In this study, we aimed to investigate the correlations between cardiovascular phenotypes and karyotypes in TS.

METHODS

We conducted a retrospective cohort analysis on 105 local TS patients, aged 6–43 years, between January 1994 and December 2018 and a literature review on 2578 TS patients to ensure results representability. They were categorized into two groups of complete monosomy X (45, X) and other X chromosome abnormalities. Most of the patients underwent echocardiography (n = 88, 83.8%), cardiac computed tomography (CT) angiography and/or cardiovascular magnetic resonance imaging (MRI) (n = 58, 55.2%). We use independent Student's t test, chi-square test or Fisher's exact test, the log rank test to compare differences in continuous data, proportions, and Kaplan–Meier survival analysis between two TS groups.

RESULTS

45, X was most common karyotype (n = 47, 44.8%). Phenotypically, cardiovascular malformations were found in 29 TS patients (27.6%). BAV (n = 6), CoA (n = 3), AS (n = 2), ASD (n = 1, 2.5%), and PAPVR (n = 1, 2.5%) were only found in the 45, X group. The mean age at AD onset was 25.55 ± 5.78 years (mean ± SD). 45, X group had significantly more AD (n = 13 vs 3, p = 0.02) and earlier onset by survival analysis (p = 0.042).

Table 1. Age and cardiovascular malformations in 105 local TS patients according to karyotype

	KARYOTYPE		TOTAL	P Value
	Monosomy X (n = 47)	Others (n = 58)	(n=105)	
Current age (mean and range [yr])	26.23±7.38 (7.43-43.03)	22.86±7.30 (6.38-36.93)	24.36±7.41 (6.38-43.03)	0.021
BAV	6/40(15.0%)	0/48(0.0%)	6/88(6.8%)	0.007
CoA	3/40(7.5%)	0/48(0.0%)	3/88(3.4%)	0.090
AS	2/40(5.0%)	0/48(0%)	2/88(2.3%)	0.204
AD	13/40(32.5%)	3/48(6.2%)	16/88(18.2%)	0.002
Aortic root dilatation	9/40 (22.5%)	3/48 (6.2%)	12/88 (13.6%)	0.033
Ascending aortic dilatation	9/40 (22.5%)	1/48 (2.1%)	10/88 (11.4%)	0.005
Both of aortic root and ascending aortic dilatation	5/40 (12.5%)	1/48 (2.1%)	6/88 (6.8%)	0.088
MVR	2/40(5.0%)	4/48(8.3%)	6/88(6.8%)	0.685
TVR	2/40(5.0%)	4/48(8.3%)	6/88(6.8%)	0.685
AR	1/40(2.5%)	2/48(4.2%)	3/88(3.4%)	1.000
MVP	1/40(2.5%)	1/48(2.1%)	2/88(2.3%)	1.000
ASD	1/40(2.5%)	0/48(0.0%)	1/88(1.1%)	0.455
VSD	0/40(0%)	1/48(2.1%)	1/88(1.1%)	1.000
Right-sided aortic arch	0/40(0%)	1/48(2.1%)	1/88(1.1%)	1.000
PAPVR	1/40(2.5%)	0/48(0.0%)	1/88(1.1%)	0.455

CONCLUSION

Cardiovascular abnormalities, such as BAV, CoA, AS, and AD, are common and potentially progressive in TS patients, especially 45, X. They should receive immediate cardiological assessments upon diagnosis, regular assessments and carefully control blood pressure, even with no apparent congenital heart disease.