



Vascular Anomalies And Aortic Dilatation in Turner Syndrome

Study In A Large Cohort Of Young-Adult Patient



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Introduction and Objectives

Patients with Turner Syndrome (TS) are at high risk for congenital heart disease (CHD), aortic dilatation and dissection with increased mortality and morbidity.

Thoracic gadolinium-enhanced MRI angiography allows clear imaging of all great vessels and revealed a spectrum of silent vascular anomalies (VA), both venous and arterial, undetected at Echo.

The aim of this study is to evaluate retrospectively the prevalence of VA and aortic dilatation in TS, in particular in patients without CHD.

Methods

A cohort of 115 patients (mean age 24.4 years; range 16 - 42,6 years) with genetically confirmed TS and no evidence of CHD at Echo and MRI was analyzed.

Echocardiography and MRI evaluations were collected in order to obtain a complete cardiological evaluation. Aortic diameters were evaluated at MRI and the values were indexed for BSA and compared with the reference values for aortic dilatation validated by Roman et al..

Results

Vascular anomalies were detected in 44 patients (38,3%) (Fig 1).

Vascular Anomalies (%)

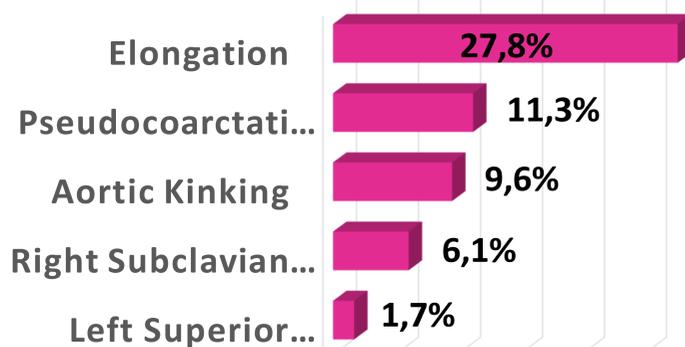


Fig. 1

Aortic dilatation was detected in 17 patients (14,8%) and was significantly associated with VA (OR=4; p=0,026; 95%CI 1,2-13,8) and age (OR=1,1; p=0,028; 95%CI 1-1,7).

The subjects with VA showed a greater prevalence of aortic dilatation both at Sinuses of Valsalva and Ascending Aorta levels than the subjects without (Fig 2).

Aortic Dilatation	No VA	VA	χ^2 , p-value
Sinuses of Valsalva	9 (7.8%)	13 (30.9%)	13.66 p<0.001
Ascending aorta	12 (10.4%)	16 (38.1%)	16.06 p<0.001

Fig. 2

The mean BSA-indexed diameters were significantly higher in subjects with VA than in subjects without (Fig.3).

Aortic Dilatation	No VA	VA	t; p-value
Sinuses of Valsalva	1.8±0.2	2.0±0.5	-4.1, p<0.001
Ascending aorta	1.6±0.2	1.8±0.5	-4.6, p<0.001
Descending aorta	1.1±0.2	1.2±0.2	-4.7, p<0.001
Aortic arch	1.2±0.2	1.2±0.3	-0.2, p=0.862
Syno-tubular junction	1.4±0.2	1.6±0.3	-3.0, p=0.003

Fig. 3

Elongation of Transverse Arch (ETA) (Fig. 4) was the VA with the greatest influence on aortic dilatation (OR=4,5; p<0,015, 95% CI 1,3-15).

No significant association was found between aortic dilatation and karyotype, phenotype severity, renal anomalies, GH and estrogen replacement therapy.

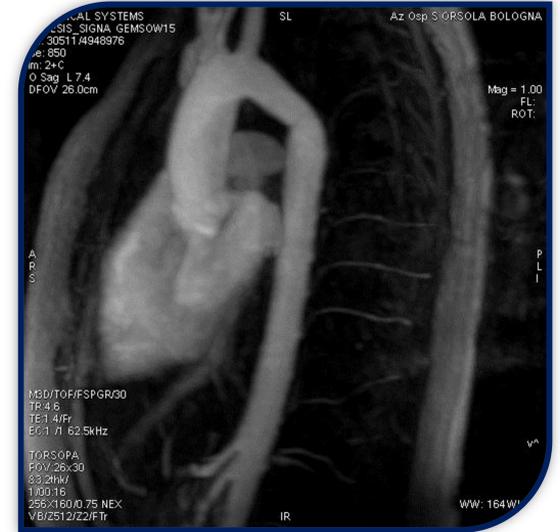


Fig. 4

Conclusions

Our study demonstrates that aortic dilatation in TS can occur without CHD and can be significantly associated with silent VA, detected at MRI.

In particular, ETA was found to be associated with a high risk of dilatation, both at ascending aorta and sinuses of Valsalva levels.

Given the high prevalence of unexpected VA in patients with TS and the correlation with aortic dilatation, it is mandatory to recommend a thoracic MRI, even in absence of CHD, in order to better estimate their cardiovascular risk, in particular before seeking to become pregnant.

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