Prevalence of Bicuspid Aortic Valve in Turner Syndrome and its Correlation to Karyotype: a Multicentre Prospective Magnetic Resonance Study

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Introduction: Congenital heart disease affects approximately 50% of individuals with Turner syndrome (TS), and significantly contributes to an overall increased mortality in TS. Bicuspid aortic valve (BAV) represents the most common congenital heart defect in TS patients. It proved to be one of the most important risk factor for aortic dilatation and dissection.

Objectives: To determine the correlation between the particular karyotypes and the prevalence of BAV in TS individual.

Methods: 67 TS patients aged 6.6 to 32.5 years (median 14.0 years) followed in three tertiary centres underwent cardiac magnetic resonance imaging study. They were divided into four cytogenetic subgroups: 45,X karyotype; 45,X/46,XX mosaicism; structural abnormalities of the X chromosome; and 45,X/structural abnormality of the X chromosome mosaicism. Prevalence of BAV and odds ratio (OR) compared to the general population in the whole study group; and statistical comparison of the prevalences of BAV among the individual subgroups were determined.

Results: BAV was found in 28.4% of all patients (OR 208.3; p-value 0.0001). Statistically significant difference between the prevalences of BAV in patients with any 45,X cell line in their karyotype compared to the patients with only structural abnormality of the X chromosome was proved (p-value 0.05).

Conclusion: our data confirms the hypothesis that prevalence of bicuspid aortic valve differs between karyotype subgroups of TS. 45,X karyotype is associated with the highest prevalence of BAV. Also, the presence of the 45,X cell line in the mosaic karyotype increases the probability of BAV.