

## Cardiovascular Anomalies in Children and Young Adults with Ullrich-Turner Syndrome—The Erlangen Experience

THOMAS M.K. VÖLKL, M.D., KARIN DEGENHARDT, M.D., ANDREAS KOCH, M.D.,\* DIEMUD SIMM, M.D., HELMUTH G. DÖRR, M.D., HELMUT SINGER, M.D.\*

Divisions of Pediatric Endocrinology and \*Cardiology, Hospital for Children and Adolescents, University of Erlangen-Nuremberg, Erlangen, Germany

### Summary

**Background:** Females with Ullrich-Turner syndrome (UTS) have typical clinical features such as short stature, ovarian failure, visible dysmorphic stigmata, and abnormalities in different organs such as kidney or heart.

**Hypothesis:** The aim of the present study was to analyze the distribution, prevalence, and relative risk of cardiovascular anomalies (CVA) in females with Ullrich-Turner syndrome (UTS) seen at one single center compared with that of the regional Bavarian population.

**Methods:** The associations between CVA and karyotype were determined. In all, 117 girls and women with UTS, aged between 3 and 43 years (median 17.4 years) were studied retrospectively. The detailed cardiologic status including echocardiography was available in all patients. The prevalences of each cardiovascular anomaly were determined. On the basis of published epidemiologic data of CVA in Bavarian children, we assessed the relative risks of each CVA.

**Results:** Thirty-five (29.9%) girls with UTS had at least one CVA. In all of these CVAs, coarctation of the aorta and bicuspid aortic valve occurred most often (18.5% each). The aortic malformations represented over two-thirds of all CVA (72.8%), whereas anomalies of the septum (8.6%), mitral valve (6.2%), pulmonary veins (4.9%), and other locations together accounted for the other third. Bicuspid aortic valve and partial anomalous pulmonary venous drainage were associated with the highest relative risk (RR) (3,603 and 1,293, respectively) compared with the Bavarian population. The overall RR of CVA was 48.7. Of the 117 girls and women examined, 64 (54.7%) had complete monosomy 45 X.

**Conclusions:** Our data demonstrate that about every third female with UTS is affected with at least one CVA, mainly left sided and associated with aortic structures. Our results underline the necessity of thorough cardiologic evaluation.

**Key words:** Ullrich-Turner syndrome, congenital heart disease, cardiovascular anomalies

---

Address for reprints:

Professor Helmut Singer, M.D.  
Division of Pediatric Cardiology  
Hospital for Children and Adolescents  
Friedrich-Alexander-University of Erlangen-Nuremberg  
Loschgestrasse 15  
91054 Erlangen, Germany  
e-mail: [helmut.singer@kinder.imed.uni-erlangen.de](mailto:helmut.singer@kinder.imed.uni-erlangen.de)

Received: June 14, 2004

Accepted with revision: October 20, 2004