

## CLINICAL OBSERVATIONS

### Deficient Medical Care for Adults with the Turner Syndrome

*Background:* The Turner syndrome is due to complete or partial absence of 1 sex chromosome in a phenotypic female, occurring in approximately 1 in 2500 live female births (1). Individuals with the

## Letters

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disorder are at increased risk for congenital heart defects, including bicuspid aortic valve, aortic coarctation, and dilation of the aorta (2, 3). These cardiovascular defects require timely diagnosis, monitoring or treatment to prevent aortic valve deterioration, endocarditis, and aortic dissection (2). Many patients have renal defects and progressive hearing loss. Good clinical practice recommends screening for these problems with echocardiography, renal ultrasonography, and audiology at the time of diagnosis (4). The risk for deterioration of the aorta in the Turner syndrome mandates regular echocardiographic monitoring over time (4). There is little information on the extent to which these practice guidelines are implemented.

*Objective:* To investigate the state of medical care received by girls and women with the Turner syndrome who were participating in a National Institutes of Health (NIH) protocol.

*Methods and Findings:* In this retrospective, cross-sectional study, we surveyed 126 adults (mean age, 36 years [SD, 11]; range, 18 to 62 years) and 52 girls (mean age, 12 years [SD, 3]; range, 7 to 17 years) with the Turner syndrome and their parents regarding 3 screening tests: echocardiography, renal ultrasonography, and audiology (4). Participants were recruited through NIH Web site notices for an institutional review board–approved study on the Turner syndrome, conducted at an NIH Clinical Research Center between 2001 and 2005. All participants or parents of minors signed informed consent forms. Ninety percent of participants were white; the remainder were Asian, African-American, and Hispanic. Inclusion criteria and study design have been described elsewhere (5). Participants answered specific questions regarding echocardiography, renal ultrasonography, and audiology on a written survey, with verification by personal interviews. Of girls with the Turner syndrome, 41 of 52 (79% [95% CI, 68% to 90%]) had all recommended tests (echocardiography, 90%; renal scan, 87%; audiology, 85%) while only 46 of 126 adults (36.5% [CI, 28% to 45%]) had all 3 tests (echocardiography, 69%; renal ultrasonography, 47%; audiology, 88%). More important, 39 of the adults (31% [CI, 24% to 40%]) had never had echocardiography even though, on average, at least 20 years had elapsed since the Turner syndrome was diagnosed. In contrast, only 5 of the girls (10% [CI, 8% to 18%]) had not had echocardiography, and all 5 had received diagnoses very recently. The mean age at diagnosis for adults was 12.3 years (SD, 8.7). The adults were relatively well-educated (mean years of formal education, 15.4 [SD, 2.3]), and 81% had medical insurance. Comparison of demographic characteristics for those who had had all 3 tests as recommended ( $n = 46$ ) and those who had had none or at most 1 of the recommended tests ( $n = 34$ ) showed no differences in age at diagnosis, time since diagnosis, years of education, or medical care coverage.

*Conclusion:* Most girls with the Turner syndrome are receiving recommended care, including cardiac, renal, and hearing evaluation. In contrast, many adult women with this disorder are not receiving adequate care. It is especially concerning that almost one third of adults with the Turner syndrome had never had cardiac imaging, despite the fact that these women are at high risk for life-threatening complications of aortic dissection and endocarditis. Fewer than half had received screening for renal abnormalities. The relatively high

rate of audiology screening seems to be due to patient-driven concern about hearing loss. The demographic characteristics of our adult study sample do not offer any obvious explanation for this deficiency in medical care. The women were generally well-educated and were health-conscious; most had health insurance and were receiving regular medical care. We conclude that there is a need for increased awareness among health care providers for the specific risks and health needs of adult women with the Turner syndrome.

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