Top Ten Things to Know
Cardiovascular Health in Turner Syndrome

1. This Scientific Statement has been compiled to provide updated information for clinicians about the cardiovascular risks, diagnosis and treatment of cardiovascular diseases in Turner Syndrome (TS).

2. TS results from complete or partial absence of the second sex chromosome in either all or part of the cells of an individual. It is the most common chromosomal abnormality affecting females, occurring in 1 in 2,500 live-born girls. Characteristic clinical features include short stature, premature ovarian failure and lymphedema. Early morbidity and mortality are increased in TS patients compared with the general population and are mainly related to cardiovascular complications.

3. TS is mainly characterized by obstructive left-sided congenital heart defects (CHDs) that are rare in the general female population and should increase clinical suspicion for TS. CHDs that are commonly diagnosed in TS include coarctation of the aorta, bicuspid aortic valve (BAV), mitral valve anomalies, hypoplastic left heart syndrome, and partial anomalous pulmonary vein return. Genetic testing for TS should be considered in females with any of these abnormalities.

4. It is important for health care professionals to recognize that hypertension in children with TS, and coronary artery disease, myocardial infarction and stroke in adults with TS, are exacerbated by an underlying predisposition to metabolic abnormalities, including dyslipidemia, type II diabetes, obesity and hyperuricemia.

5. Due to the high prevalence of congenital and acquired cardiovascular disease in TS, noninvasive cardiac imaging is critical for diagnosis, management, and risk assessment.

6. All women with TS should be counseled about the increased cardiovascular risk of pregnancy. Women with aortic dilatation, BAV, elongation of the transverse aorta, coarctation of the aorta and/or hypertension should be advised that pregnancy carries a high risk of aortic dissection.

7. In addition to CHDs and aortopathy, girls with TS are at increased risk for obesity, abnormal triglycerides, diabetes, hypertension, stroke, and ischemic heart disease. More importantly, the majority of serious sequelae and increased mortality associated with these health risks present in adults with TS.

8. For all girls diagnosed with TS, a detailed transition strategy emphasizing an understanding of the cardiac status and the importance of lifelong care and prevention is critical for the patient and the family.

9. A heart-healthy lifestyle is essential and should be discussed with adolescents and young adults given the increased risk of obesity, abnormal triglycerides, diabetes, hypertension, stroke, and ischemic heart disease.

10. A better understanding of cardiovascular health in TS must overcome two fundamental challenges: (1) a vast array of cardiovascular diseases significantly affect girls and women with TS, and (2) TS is a rare condition making difficult the recruitment of large numbers of subjects for clinical studies. Accordingly, significant advances will require collaboration across all the cardiovascular subspecialties and will need to engage research centers throughout the world.