Introduction

Turner syndrome (TS) is the result of complete or partial absence of one X chromosome. In addition to short stature and ovarian failure, TS is associated with a wide range of abnormalities affecting nearly every organ system. Adults with TS have a reduced life expectancy because of a highly increased risk of atherosclerosis and structural abnormalities of the heart and aorta. The increased risk of atherosclerosis results from an increased prevalence of hypertension, diabetes mellitus and dyslipidaemia. TS patients may also have multiple comorbidities including hypothyroidism, skeletal problems, renal and gastrointestinal disease, hearing loss and psychosocial problems.

Girls with TS usually receive intensive medical care during childhood including treatment to promote growth and induce puberty. Thus, paediatricians must refer these girls to other physicians as appropriate. After completing puberty and reaching final height, TS patients should be transitioned from paediatric to adult healthcare. However, results of several studies suggest that medical care for adult women with TS is suboptimal, particularly with regard to cardiovascular health. Long-term oestrogen replacement is mandatory not only for optimal uterine development but also for cardiac and bone health. To cope with a range of overt and occult morbidities, women with TS are very likely to benefit from coordinated care provided by a multidisciplinary medical team.

In this workshop, Carolyn Bondy of the National Institute of Child Health and Human Development of the National Institutes of Health in Bethesda, Md., USA, discusses the relationship between fetal ultrasound findings

and survival of fetuses with a 45,X karyotype and presents evidence that a prenatal cytogenetic diagnosis of X chromosome monosomy without clinical correlation is a poor predictor of clinical outcome. In addition, Dr. Bondy summarizes current knowledge on the management of growth and puberty in girls with TS and considers the possible risks of early- and long-term growth hormone treatment. Treatment with low-dose oestrogen should be started at an age-appropriate time and continued throughout adulthood to ensure optimal pubertal, psychosocial, uterine and bone development. Dr. Bondy also discusses prevention of premature mortality from complications of congenital and acquired cardiovascular disease. At her institution, cardiac magnetic resonance images are used to screen for cardiovascular defects in girls and women with TS, and this technique has advantages over echocardiography. Finally, she stresses the importance of regular, long-term follow-up of the cardiovascular system in adult women with TS.

Disclosure Statement

S.M.P.F. de M.K.-S. declares no conflict of interest.

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