

Turner Syndrome: Pediatric to Adult Transition



This department covers selected points from the 2009 Endocrine Update: A CME Day from the Division of Endocrinology and Metabolism at McMaster University and the University of Western Ontario.
Program Chairs: Aliya Khan, MD, FRCPC, FACP, FACE and Terri Paul, MD, MSc, FRCPC



Zubin Punthakee, MD, MSc, FRCPC; Lisa-Ann Fraser, MD, FRCPC; and Sultan Chaudhry, BHSc

Turner syndrome (TS) is a condition, affecting one in 2,000 to 5,000 live female births where all or part of one X chromosome is either missing or has a structural abnormality. Although TS can be identified by karyotype, the karyotype does not have a strong enough relationship with the phenotype to be used to predict clinical manifestations reliably.

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Due to lack of knowledge and understanding of the condition by many physicians, the transition from pediatric care to adult care is often a problem. A Belgian survey of 160 girls with TS (mean age of 23-years-old) found that 12.7% had no regular follow-up medical care and an additional 14.7% were being followed only by a GP. Of the girls surveyed, 40.2% were facing health problems including hypertension (10.7%) and

hypothyroidism (5.8%). Fourteen point five per cent of patients who had required pubertal induction in the pediatric clinic were no longer taking estrogen.¹ An Australian survey of 39 adult women with TS (mean age of 30-years-old) found 38% had no regular medical follow-up and 56% were receiving inadequate medical surveillance.²

Ninety per cent of TS patients experience gonadal failure and 70% have no spontaneous pubertal development (the majority have primary amenorrhea). Pubertal induction in the pediatric stage is started with estrogen at 12- to 13-years-of-age using one-eighth to one-tenth the usual adult dose. The dose is then increased to the adult dose over a period of two years.³ TS patients are then started on cyclic estradiol/estrogen and progesterone. Fertility has been reported in three of 352 TS patients (0.6%) in one published case series.⁴ However, when pregnancy is achieved, TS patients have an increased risk of pregnancy loss and fetal chromosomal abnormalities. Technology such as gamete intrafallopian transfer and oocyte or ovarian cryopreservation is available in specialized centres to help women with TS gain fertility.⁴ Full cardiac evaluation must be performed before pregnancy is considered due to increased risk of aortic dissection during pregnancy.

TS patients are also candidates for growth hormone therapy in childhood and experience

increased rates of hypothyroidism and celiac disease. Other medical problems include gonadoblastoma, inflammatory bowel disease, elevated liver enzymes and hearing loss.⁴

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One study has reported TS to be associated with an increased relative risk of 4.2 for premature death.⁵ Much of the mortality risk associated with TS is due to cardiac abnormalities and aortic dissection.^{5,6} ECHO is used to detect congenital cardiac abnormalities among infants and young girls with TS; however, for older girls and adults, MRI is often more informative.⁴ Arrhythmias, aortic dissection, hypertension, coarctation and dilation of the aorta, unusual angulation and elongation of the aortic arch and ischemic heart disease are

Dr. Punthakee is an Assistant Professor, Departments of Medicine and Pediatrics, McMaster University, Hamilton, Ontario.


Dr. Fraser is a Fellow, Division of Endocrinology and Metabolism, Schulich School of Medicine & Dentistry, University of Western Ontario, London, Ontario.

Dr. Chaudhry is an MD/PhD Candidate, the Michael G. DeGroote School of Medicine & Medical Sciences, McMaster University, Hamilton, Ontario.

some of the most serious consequences of TS. ECG can be used to detect T-wave abnormalities, QT prolongation, resting tachycardia and right axis deviation in some cases. Evaluation by an experienced cardiologist is recommended for all patients at the time of diagnosis and regular follow-up, counselling and cardiac monitoring should be a routine part of care.⁴

Increased fracture risk is also an issue in TS due to frail cortical bone (despite normal BMD), small bones/altered bone geometry, high risk of falls and trauma and higher risk of osteoporosis later in life.⁷ Maintaining adequate levels of vitamin D and calcium, exercise, monitoring of BMD, as well as estrogen replacement are important aspects of maintaining bone health.

For more detailed and complete information on the management of TS, please refer to Bondy, *et al's* 2007 guidelines.⁴ Recommended guidelines for adults include:

- estrogen (and progesterone) replacement,
- re-education on TS and pregnancy,
- BP and glucose monitoring,
- TSH and liver enzymes annually and
- celiac screening. 

References

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