Journal of Pediatric Nursing (2010) **25**, 66–68

PENS COLUMN

Column Editor: Terri H. Lipman, PhD, CRNP, FAAN

Turner Syndrome: Do Not Miss This Diagnosis

Rosemary Scales RN, MSN*, Colleen Weber RN, BSN

Pediatric Endocrinology Nursing Society, University of Utah, Salt Lake City, UT

As a pediatric nurse, have you ever wondered whether you're missing the diagnosis of a patient? Have you cared for a young girl who has short stature and frequent ear infections or an adolescent girl without breast tissue? Would you be concerned? If so, would you consider the diagnosis of Turner syndrome? It may be problematic for the pediatric nurse to identify this condition due to the wide array of presentation and physical characteristics.

Turner syndrome (TS) occurs in about 1:2000 live female births; the diagnosis requires the presence of the typical phenotypic features in a female and the complete or partial absence of a second sex chromosome (Postellon, 2008). Karyotyping (testing to identify and evaluate the size, shape, and number of chromosomes in a sample of body cells) is required for the diagnosis and may depict one of three possible chromosomal structural anomalies. The most severe phenotype, occurring in approximately 50% of these girls, result in a 45,XO karyotype, demonstrating the absence of an X chromosome. The second possibility is known as a partial X deletion in which both X chromosomes are present but one arm of one X chromosome is missing or malformed; this anomaly occurs in 30% of girls with TS. The third genetic form, mosaicism, is consistent with the presence of 2 or more cell lines that differ in their chromosomal makeup; some cells may have 46 chromosomes, whereas others have an altered number. Those with Turner mosaicism usually have the less severe phenotype and may enter puberty spontaneously before developing gonadal failure. The American College of Medical Genetics recommends the standard 30-cell karyotype for diagnosis. The condition may also be established prenatally by amniocentesis, although chromosomes should be reevaluated postnatally to confirm the diagnosis (Bondy, 2007; Davenport & Rosenfeld, 2003; Postellon, 2008).

Almost all girls with TS have short stature and ovarian failure, although other physical findings and anomalies vary. At birth, the diagnosis of TS may be made after the identification of edematous hands and feet. A webbed neck, ear deformities, broad chest, and cardiac or renal malformations resulting from the congenital lymphedema may also be apparent. Growth failure, lack of pubertal development, and/or amenorrhea usually brings these girls to medical attention after the newborn period (Moshang, 2005). There are numerous common physical characteristics and abnormalities that may occur in TS; girls may exhibit one or several of these anomalies. It must be emphasized that some girls will present with short stature and/or signs of ovarian failure, whereas the other syndrome features may be mild or absent causing a delayed diagnosis. Refer to Table 1 for other physical characteristics and abnormalities that may be exhibited.

Short stature, the most common visible characteristic, occurs in 98% of girls with TS; they typically have intrauterine growth retardation, a subnormal growth

* Corresponding author: Rosemary Scales, RN, MSN.
E-mail address: rjscales@comcast.net (R. Scales).

0882-5963/$ – see front matter
doi:10.1016/j.pedn.2009.10.001

Table 1 Other Possible Abnormalities in TS

Facies	Narrow maxilla, wide mandible
	Narrow and high-arched palate
	Micrognathia and/or retrognathia
Skin/nails chest	Broad chest with widely spaced nipples, inverted nipples
	Short webbed neck, nuchal folds as an infant, low posterior hairline
	Lymphedema of hands and feet as an infant
	Increased body hair growth, bushy eyebrows
	Hyperconvex uplifted or underdeveloped/ hypoplastic nails, absent toenails
	Increase incidence of pigmented moles, telangiectasis
Opthalmic	Ptosis, strabismus, amblyopia, myopia
	Epicanthal folds
	Cataracts more common
Ears	Low posterior rotated ears, deformed auricles
	Inner ear defects, increased risk of ear infections
	Hearing problems, sensorineural hearing loss
Cardiac	Left-sided cardiac malformations occurs in 30% of girls including coarctation of the aorta, bicuspid aortic valve, stenosis of aortic isthmus, aortic dilatation/aneurysm, hypoplastic left heart, anomalous pulmonary venous return
Orthopedic	Congenital hip dislocation in infants
	Cubitus valgus (increased carrying angle for the elbows)
	Short fourth and/or fifth metacarpal
	Increase incidence of scoliosis, spongiose bone structure, and/or osteopenia
	Knock-knees, madelung deformity
Renal	Kidney malformations occurs in 30% of patients
	Defect in collecting system
	Abnormal positioning
	Variation in number and position of renal arteries
Neuropsych issues	Visual—spatial and temporal processing difficulties, difficulty with math, reading maps, following directions
	Visual–Motor difficulties—clumsiness, difficulty with fine motor skills
	Difficulty with social cognition—difficulty with nonverbal learning and social skills

velocity during infancy and childhood, and an absence of the pubertal growth spurt. These girls are also at higher risk for skeletal anomalies; 10%–20% of these girls develop scoliosis and kyphosis. Short stature may include a disproportionate increase in weight and may affect the lower limbs more than the upper limbs, resulting in shorter legs than trunk. The average adult height of a woman with TS is 142–147 cm (4 ft 8 in; Bondy, 2007; Raine, Donaldson, Gregory, Savage, & Hintz, 2006; Raine, 2007). TS is an approved indication for human growth hormone (hGH) treatment, although girls with TS are not deficient in hGH. In childhood, it is standard for an endocrinologist to treat girls with TS with hGH with the goal being to obtain as normal height for age as possible. It is recommended that consideration of hGH should be as soon as growth failure is demonstrated; in the United States, growth hormone therapy is initiated at the Food and Drug Administration indicated a dose of 0.375 mg/kg per week (Bondy, 2007; Postellon, 2008). Girls with TS treated long term with appropriate dose of hGH will have a mean final height gain of approximately 7–9 cm (2.7–3.5 in; Raine, Donaldson, Gregory, Savage, Hintz, 2006).

Gonadal or ovarian dysgenesis, a hallmark of partial or complete X chromosome loss, refers to abnormal or incomplete development of the ovary and occurs in more than 90% of TS girls. Lack of estrogen and progesterone results in the absence of breast development, feminine body contours, and menstruation, with accompanying infertility. During adolescence, most girls with TS will have an increased level of follicle-stimulating hormone values, indicative of ovarian failure. Up to 30% of these girls may enter and achieve partial puberty; some may achieve menarche, but ovaries eventually will cease functioning. Those who achieve spontaneous puberty are at risk for premature menopause. Fertility occurs in less than 2%–5% of these females. Estrogen/Progesterone replacement therapy is indicated to induce pubertal development and maintain feminization; the form, dosing, and timing should reflect the normal process of puberty (Bondy, 2007; Moshang, 2005).

The clinician should be prompted to obtain a karyotype on any girl who presents with unexplained growth failure; once the diagnosis of TS is confirmed, the patient should be referred to a pediatric endocrinologist. At that point, the diagnosis would be discussed thoroughly so that further testing and treatment can begin. Necessary radiologic procedures would include an echocardiogram and/or a cardiac MRI, a renal/pelvic ultrasound, and a bone age. The pediatric endocrinologist should follow up these patients for treatment with growth hormone, sex steroid replacement, and possibly thyroid medication. Other recommended procedures may include an ophthalmologic examination, an ENT/audiology examination, blood pressure monitoring, scoliosis exam, Dual Energy X-ray Absorptiometry (DEXA) scan, an orthodontic evaluation, and comprehensive psychosocial and academic assessments. Dietary/Exercise counseling may be necessary. Surgical repairs may be required for cardiac and renal anomalies as well as cosmetic procedures for a webbed neck and perhaps breast augmentation. Scoliosis treatment may also be required. Speech therapy, educational interventions, and a psychosocial plan may also be necessary (Bondy, 2007; Ross, St. Dennis-Feezle, Weber, 2001).

Because few characteristics may be obvious, the diagnosis of TS may be easily missed; a delayed diagnosis may lead to obstacles in health care (Davenport & Rosenfeld, 2003). As the pediatric patient ages, it is

particularly important to emphasize that transition to adult care must occur in a timely fashion. These patients require frequent monitoring as children and adults to achieve a productive, normal lifestyle.

References

Bondy, C. (2007). Clinical practice guidelines. Care of girls and women with Turner syndrome: A guideline of the TS Study Group. *J Clin Endocrinol Metab*, *92*, 10−25.

Davenport, M., & Rosenfeld, R. (2003). Turner syndrome. In S. Radovick, & M. MacGillivray (Eds.), *Pediatric endocrinology a practical clinical guide* (pp. 117−137). New Jersey: Humana Press.

Moshang, T. (2005). *Pediatric endocrinology the requisites in pediatrics.* Elsevier Mosby.

Postellon, D. (2008). Turner syndrome. emedicine.medscape.ecom.

Raine, J., Donaldson, M., Gregory, J., Savage, M., & Hintz, R. (2006). *Practical endocrinology and diabetes in children* (2nd ed., pp. 50−51). USA: Blackwell Publishing Ltd.

Ross, J.L., St. Dennis-Feezle, L.K, Weber, C., (2001). Turner Syndrome, *Toward Early Recognition and Improved Outcomes*, CD by University of California, Irvine College of Medicine, Deborah Wood and Associates & Eli Lilly and Company.