Health Supervision for Children With Turner Syndrome

Committee on Genetics

This set of guidelines is designed to assist the pediatrician in caring for the child in whom the diagnosis of Turner syndrome has been confirmed by karyotype. Although the pediatrician’s first contact with the child is usually during infancy, occasionally the pregnant woman who has been given the prenatal diagnosis of Turner syndrome will be referred for advice. Therefore, these guidelines offer advice for this situation as well.

Turner syndrome, as used here, refers to a condition in which there is short stature and ovarian dysgenesis in females because of the absence of a normal second sex chromosome. Nonchromosomal gonadal dysgenesis is excluded. The birth prevalence of Turner syndrome has been estimated to be from 1:2000 to 1:5000 female live births. About 1% to 2% of all conceptuses have a 45,X chromosome constitution. Of these, the majority (99%) spontaneously abort, usually during the first trimester of pregnancy. With the more frequent use of ultrasound, it is recognized that some pregnancies with a fetal 45,X chromosome constitution progressing into the second trimester are associated with nuchal cysts, severe lymphedema, or hydrops fetalis. These pregnancies are associated with a high frequency of fetal death.

PHENOTYPE

Pediatricians are most familiar with the clinical findings that prompt the diagnosis in children, namely, short stature and the classic Turner syndrome features such as lymphedema, webbed neck, low posterior hair line, and cubitus valgus. A wide range of clinical abnormalities may be found (Table 1). Turner syndrome, however, is not always accompanied by distinctive features and most often is not diagnosed in infancy. Later in childhood, Turner syndrome may be suspected primarily because of short stature. Other prominent presenting features in teenage years are delayed puberty and delayed menarche, and in adult women, anovulation and infertility. A girl with 45,X Turner syndrome may manifest an X-linked recessive disorder because she has only one X chromosome. When a girl has an X-linked recessive disorder, therefore, the possibility of Turner syndrome caused by 45,X or a structural abnormality of an X chromosome should be considered.

Growth in children with Turner syndrome is characterized by a slight intrauterine growth retardation, relatively normal growth velocity for the first several years of life, a progressive deceleration of growth later in childhood, and the lack of a pubertal growth spurt. Because of delayed epiphyseal closure, relatively small gains in height may occur even after 20 years, although normal height is rarely achieved, except with mosaicism (the presence in an individual of two or more chromosomally different cell lines, both originating from the same zygote). The anticipated adult height is about 143 cm and rarely exceeds 150 cm.

Contrary to earlier reports, most persons with Turner syndrome are not mentally retarded, although they may have learning disabilities, particularly with regard to spatial perception, visual-motor coordination, and mathematics. As a result, the nonverbal IQ in Turner syndrome tends to be lower than the verbal IQ.

Cytogenetics

Turner syndrome most likely results from haploinsufficiency (the presence in the cell of one set of genes rather than the usual two sets) for specific genes located on the X chromosome. In healthy 46,XX female embryos, inactivation of one X, referred to as lyonization, occurs in every somatic cell shortly after fertilization. The genes involved in Turner syndrome, however, seem to escape inactivation selectively. Thus, the healthy 46,XX female embryo has a functional diploid set of the genes in question. These genes also seem to have homologs on the Y chromosome, which accounts for the normal growth and development in XY male embryos.

The exact location of the genes on the X chromosome involved in Turner syndrome has not been determined. At present, evidence exists that there is a locus for stature on the distal portion of the short arm; there are loci for normal ovarian function on both the short and long arms; and there are loci contributing to fetal viability on the long arm of X.

In about 80% of girls with 45,X, the single remaining X chromosome is inherited from the mother, and in the remaining 20% it is from the father. Imprint-
<table>
<thead>
<tr>
<th>Clinical Abnormalities in Turner Syndrome*</th>
<th>Approximate Incidence (%)†</th>
</tr>
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<tbody>
<tr>
<td>Short stature</td>
<td>100</td>
</tr>
<tr>
<td>Gonadal dysgenesis with hypoplasia or aplasia of germ cells</td>
<td>&gt;90</td>
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<tr>
<td>Edema of hands and feet</td>
<td>&gt;90</td>
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<tr>
<td>Broad chest with inverted or hypoplastic nipples</td>
<td>&gt;80</td>
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<tr>
<td>Unusual shape and rotation of ears</td>
<td>&gt;80</td>
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<tr>
<td>Narrow maxilla including palate</td>
<td>&gt;80</td>
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<tr>
<td>Micrognathia</td>
<td>&gt;70</td>
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<tr>
<td>Inner canthal folds</td>
<td>&gt;40</td>
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<tr>
<td>Low posterior hairline with appearance of short neck</td>
<td>&gt;80</td>
</tr>
<tr>
<td>Webbed neck</td>
<td>50</td>
</tr>
<tr>
<td>Cubitus valgus or other elbow anomaly</td>
<td>&gt;70</td>
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<tr>
<td>Knee anomaly, eg, tibial exostosis</td>
<td>&gt;60</td>
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<tr>
<td>Short metacarpals or metatarsals, usually 4th</td>
<td>&gt;50</td>
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<tr>
<td>Nail dysplasia (eg, narrow, hyperconvex, deeply set, with soft, upturned tips)</td>
<td>&gt;70</td>
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<tr>
<td>Pigmented nevi</td>
<td>&gt;50</td>
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<tr>
<td>Abnormal dermatoglyphics</td>
<td>&gt;40</td>
</tr>
<tr>
<td>Cardiac anomalies</td>
<td>&gt;20</td>
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<tr>
<td>Mostly bicuspid aortic valve, coarctation of aorta, aortic valve stenosis, also hypoplastic left heart, mitral valve prolapse, dissecting aortic aneurysm (rare)</td>
<td>60</td>
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<tr>
<td>Renal anomalies</td>
<td>&gt;60</td>
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<tr>
<td>Mostly horseshoe kidney, duplicated renal pelvis, ectopic or malrotated kidney, or vascular anomalies</td>
<td>&gt;50</td>
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<tr>
<td>Central nervous system</td>
<td>&gt;60</td>
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<tr>
<td>Hearing loss</td>
<td>&gt;50</td>
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<tr>
<td>Occasional abnormalities</td>
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<tr>
<td>Dysplastic hip</td>
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<tr>
<td>Madelung deformity (radial deviation of hand because of abnormal ulnar or radial growth)</td>
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<td>Scoliosis</td>
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<td>Kyphosis</td>
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<td>Vertebral fusion</td>
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<td>Ptosis</td>
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<td>Strabismus</td>
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<td>Blue sclerae</td>
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<td>Cataract</td>
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<tr>
<td>Hemangiomas, rarely of intestine</td>
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<tr>
<td>Tendency to form keloids</td>
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<tr>
<td>Tendency to obesity</td>
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<tr>
<td>Idiopathic hypertension</td>
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<td>Diabetes mellitus</td>
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<td>Abnormal glucose tolerance</td>
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<td>Crohn disease</td>
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<td>Thyroid disorders</td>
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<td>Ulcerative colitis</td>
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* Modified from Jones KL.15
† Incidence figures in published studies vary with source of data and population characteristics.
‡ Controversial. See text.

ing (an alteration in the expression of a gene or a chromosome, depending on whether the genetic material is inherited from the mother or father) does not seem to operate, because the phenotype of an individual with 45,X does not vary according to the parental origin of the X chromosome.10

When the diagnosis of Turner syndrome is suspected, the appropriate test is chromosome analysis. A wide range of karyotypic abnormalities exists in Turner syndrome. When conventional karyotyping is done from lymphocyte cultures, about 50% of patients show a 45,X chromosome constitution. Other karyotypes found with Turner syndrome are mosaicism of 45,X with other cell lines such as 46,XX, 46,XY, or 47,XXX. Structural anomalies of an X chromosome, such as isochromosomes (an abnormal chromosome with equal arms originating from a transverse division of the centromere during cell division, instead of the normal longitudinal division), deletions, rings, or translocations, also may be found in Turner syndrome. Structural X anomalies are often mosaic with 45,X or 46,XX cells. Although mosaicism with a 46,XX line (which is the most frequent mosaicism found with 45,X) tends to be a more "normal" phenotype on the average, the clinical findings cannot be predicted in an individual case.

In fact, mosaicism in Turner syndrome may be more common than previously thought. When two tissues (lymphocytes and fibroblasts) were examined, about 80% of patients with 45,X Turner syndrome were found to be mosaics.11 Mosaicism in live-born girls with Turner syndrome is more frequent than that in fetuses with Turner syndrome, suggesting that a second sex chromosome (or a critical portion of a second sex chromosome) may be necessary for fetal survival and that most, or perhaps all, individuals with Turner syndrome are mosaics.9,11,12

Girls with 45,X Turner syndrome should have an adequate cytogenetic and DNA examination for covert Y chromosome mosaicism or a covert Y cell line. When the karyotype shows a marker chromosome
unknown origin, molecular studies using Y chromosome identified by conventional cytogenetic methods) of Turner syndrome mosaicism also should be investigated thoroughly if clitoromegaly or masculinized genitalia are present at birth or if virilization occurs at puberty. When Y chromosome mosaicism is present, there is an increased risk, estimated from 15% to 25%, for developing gonadoblastoma and dysgerminoma in the dysgenetic mosaicism is present, there is an increased risk, estimated from 15% to 25%, for developing gonadoblastoma and dysgerminoma in the dysgenetic gonads,\textsuperscript{13,14} and prophylactic gonadectomy is recommended.

Buccal smears for X chromatin bodies (Barr bodies) when Turner syndrome is suspected do not have sufficient sensitivity and specificity, even for screening. Individuals with mosaicism or a structural X abnormality may be X chromatin positive. Also, a Y chromosome would not be detected in a routine buccal smear with examination only for X chromatin.

**MEDICAL TREATMENT**

The medical care of children with Turner syndrome requires ongoing assessment and periodic review at appropriate ages (Table 2), including the following:

1. Check the child's blood pressure and peripheral pulses during each physical examination. Although idiopathic hypertension is found in Turner syndrome, a careful search for cardiac or renal causes should be made.
2. Specifically check for serous otitis and otitis media on every visit, and, if present, institute aggressive treatment. Evaluate the child's hearing, especially if otitis has been present. Hearing loss, which is common in Turner syndrome, may be conductive or sensorineural.
3. If the child's features are significantly dysmorphic, consider plastic surgery for the neck, face, or ears to improve appearance before the child enters school and thereafter, as indicated. Some individuals with Turner syndrome have a tendency to form keloids,\textsuperscript{15} which must be taken into account when surgery is considered.
4. Discuss diet and exercise for weight control, because obesity may be a problem in Turner syndrome.\textsuperscript{16}
5. Perform routine urinalysis annually to monitor for diabetes mellitus. Glucose intolerance occurs more frequently in persons with Turner syndrome than in the general population; however, frank diabetes mellitus seems to be rare.\textsuperscript{17,18}
6. Review the psychological support available to the child and family to optimize the child's psychosocial adjustment.
7. Encourage family support by referral to individuals with Turner syndrome, parents of children with Turner syndrome, or a Turner syndrome support group. Supply the family with literature and referral when indicated by history or examination findings; (6), see “discussion”; \bullet, to be performed; S, subjective, by history; O, objective, by a standard testing method.

**TABLE 2. Health Supervision Guidelines for Children With Turner Syndrome**

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Prenatal</th>
<th>Infancy (1 mo–1 y)</th>
<th>Early Childhood (1–5 y)</th>
<th>Late Childhood (5–13 y)</th>
<th>Adolescence (13–21 y)</th>
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<tbody>
<tr>
<td>Karyotype review (1)</td>
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<td>Review family support</td>
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<td>Thyroid screening (6)</td>
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<td>Echocardiogram (6)</td>
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<td>Renal sonogram (6)</td>
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<td>Serum gonadotropins (LH and FSH)</td>
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<td>Development and behavior (5)</td>
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<td>School performance (5)</td>
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<td>Socialization</td>
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<tr>
<td>Sexual issues</td>
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</table>

* Assure compliance with the American Academy of Pediatrics Recommendations for Preventive Pediatric Health Care. (1), at time of diagnosis; (2), discuss referral to specialist; (3), give once in this age group; (4), according to state's screening program; (5), objective testing when indicated by history or examination findings; (6), see “discussion”; \bullet, to be performed; S, subjective, by history; O, objective, by a standard testing method.
THE PRENATAL VISIT

When a prenatal diagnosis of 45,X or another karyotype associated with Turner syndrome is made, counseling is ordinarily provided for the family by a medical geneticist, a pediatric endocrinologist, or another physician with special knowledge of Turner syndrome. Sometimes because of a previous relationship with the family, the pediatrician may be asked to review the information and to assist the family in decision making.

Prenatal diagnosis of Turner syndrome may have been made or suspected because of ultrasonographic evidence of fetal edema or nuchal cystic hygroma, or an abnormal karyotype discovered when fetal chromosome analysis was done for other reasons, such as advanced maternal age. Results of maternal serum screening with multiple markers (maternal serum alpha-fetoprotein, human chorionic gonadotropin, and unconjugated estriol) projecting an increased risk for Down syndrome also have detected some fetuses with Turner syndrome. Ultrasonography showing a left-sided cardiac anomaly (Table 1), growth retardation, or relatively short limbs also may suggest Turner syndrome. If an abnormality associated with Turner syndrome is made, discuss with the geneticist whether further cytogenetic studies should be performed. Chromosome analyses from peripheral blood or other tissues may be indicated depending on the adequacy of the prenatal study and the possibility of mosaicism, especially for the Y chromosome. Evaluate the child for typical features of Turner syndrome (Table 1) as follows.

1. Examine the child’s hips for dysplasia. Repeat the examination several times during early infancy.
2. Obtain an echocardiogram. Give special attention to the possibility of left-sided cardiac anomalies (Table 1).
3. Obtain an initial consultation with a pediatric cardiologist for all persons with Turner syndrome who have abnormal echocardiograms.
4. Obtain a renal sonogram (or repeat the sonogram if it was done prenatally). Although some of the renal anomalies listed in Table 1 are not clinically significant, others may predispose the child to urinary tract infections, hydronephrosis, and hypertension.

Anticipatory Guidance

Discuss the diagnosis of Turner syndrome, the phenotype, and the variability of the phenotype. Both parents should be present whenever possible. They need to know that short stature and infertility are likely, mental retardation is unlikely, some congenital anomalies may be present (Table 1), and some learning difficulties are expected.

Discuss the treatments and interventions available, such as sex hormone replacement and growth-enhancing therapy, and emphasize that with medical supervision and psychosocial counseling and support, girls with Turner syndrome may lead healthy, satisfying lives. In cases of early prenatal diagnosis, however, some parents may decide to terminate their pregnancies.

The risk for having a fetus or child with Turner syndrome is not related to advanced maternal age. Most often, Turner syndrome is a sporadic event, and the risk of recurrence is not increased for subsequent pregnancies. There may be some rare exceptions, however, such as inheritance of a structural X anomaly and inherited mosaicism.

HEALTH SUPERVISION FROM BIRTH TO 1 MONTH: NEWBORNS

Examination and Laboratory Studies

Confirm the diagnosis of Turner syndrome and review the karyotype. If a prenatal diagnosis was made, discuss with the geneticist whether further cytogenetic studies should be performed. Chromosome analyses from peripheral blood or other tissues may be indicated depending on the adequacy of the prenatal study and the possibility of mosaicism, especially for the Y chromosome. Evaluate the child for typical features of Turner syndrome (Table 1) as follows.

1. Examine the child’s hips for dysplasia. Repeat the examination several times during early infancy.
2. Obtain an echocardiogram. Give special attention to the possibility of left-sided cardiac anomalies (Table 1).
3. Obtain an initial consultation with a pediatric cardiologist for all persons with Turner syndrome who have abnormal echocardiograms.
4. Obtain a renal sonogram (or repeat the sonogram if it was done prenatally). Although some of the renal anomalies listed in Table 1 are not clinically significant, others may predispose the child to urinary tract infections, hydronephrosis, and hypertension.

Anticipatory Guidance

1. Distinguish the anomalies requiring careful medical treatment (eg, cardiac and renal anomalies) from those of primarily cosmetic and psychological importance.
2. Inform the family that lymphedema may persist for months or may recur.
3. Discuss the possibility of feeding problems. Some infants with Turner syndrome have inefficient sucking and swallowing reflexes because of impaired oral motor function.
4. Discuss the current status of endocrine therapy for growth and for the development of secondary sex characteristics. Indicate that infertility is almost always present, although assisted reproduction techniques may enable infertile couples to have children.
5. Discuss subacute bacterial endocarditis prophylaxis if a cardiac anomaly is present.
6. Talk about how and what to tell other family members and friends.

HEALTH SUPERVISION FROM 1 MONTH TO 1 YEAR: INFANCY

Examination and Laboratory Studies
1. Assess the infant’s weight, taking into account that many infants with congenital lymphedema lose weight during the first month because of diuresis.
2. Perform an ophthalmologic evaluation. Nonalternating strabismus may be present. Refer the infant to an ophthalmologist as soon as strabismus is diagnosed.
3. Obtain an echocardiogram, even if one was obtained in the newborn period, because abnormalities of the aortic valve may not have been identified.

Anticipatory Guidance
1. Provide prophylaxis for subacute bacterial endocarditis if a cardiac anomaly is present. Subacute bacterial endocarditis is rare in infancy.
2. If urinary tract abnormalities are present, perform a urinalysis and culture when indicated for possible urinary tract infections. Ultrasonography is also advised if urinary tract infections recur or hypertension develops.
3. Consider referring the infant to an appropriate pediatric specialist if cardiac, renal, or eye abnormalities are found.
4. Consider referring the infant to developmental intervention programs if neuromuscular development is delayed.

HEALTH SUPERVISION FROM 1 TO 5 YEARS: EARLY CHILDHOOD

Examination and Laboratory Studies
1. Follow the child’s growth on the Turner syndrome growth curve, starting at 2 years (Figure).
2. Evaluate the child’s speech and refer the child to a speech therapist when appropriate.
3. Continue to evaluate the child’s cardiac status with an echocardiogram or magnetic resonance imaging scan at yearly intervals. A magnetic resonance imaging scan may be preferable for examination of the aorta. Because aortic dissection is a serious complication, the aortic root should be evaluated carefully for dilatation. About 9% of patients with Turner syndrome had unrecognized aortic root dilatation in one study.24 These patients are at substantial risk for aortic dissection. Therefore, it is recommended that the child be referred to a pediatric cardiologist if dilatation of the aortic root is suspected or confirmed to be present.
4. Continue to evaluate the child’s renal status (urinalysis and culture, as indicated) if a renal anomaly is present.
5. Test for thyroid function at 1- to 2-year intervals because of the increased frequency of hypothyroidism, usually caused by autoimmune thyroiditis.25

Anticipatory Guidance
Evaluate the child for possible learning difficulties, particularly spatial perception problems. A special psychological assessment of the child before entering a preschool program may benefit the child and parents as well as school personnel. Information about testing and evaluation resources may be obtained from the school or from state and regional programs for persons with developmental disabilities.

HEALTH SUPERVISION FROM 5 TO 13 YEARS: LATE CHILDHOOD

Examination and Laboratory Studies
1. Discuss the diagnosis and treatment of Turner syndrome with the child, as soon as he or she is able to understand, as well as with the parents.
2. Monitor the child for urinary tract infections.
3. Check the child’s dentition for malocclusion.
4. Continue to obtain an echocardiogram each year if a cardiac anomaly is present, otherwise obtain an echocardiogram at 2-year intervals. The pediatrician may prefer to have the child treated by a pediatric cardiologist.
5. Continue testing for thyroid function at 1- or 2-year intervals.
6. Check for scoliosis yearly. Lordosis and kyphosis are also seen more frequently in girls with Turner syndrome.

Anticipatory Guidance
1. Watch for potential school problems, such as specific learning disabilities, attention deficits, hyperactivity, and difficulty in developing social skills. Refer the child for educational evaluation and intervention, as indicated. Encourage parents to interact with school personnel.
2. Discuss adjustment to short stature with the parents and separately with the child.
3. Refer the child to a pediatric endocrinologist for consideration of hormonal growth-enhancing therapy. Ordinarily such therapy is begun before 10 years of age and may be advised when the child’s height falls below the fifth percentile for healthy children that age. Biosynthetic human growth hormone alone or in conjunction with androgen (oxandrolone) increases the rate of growth in most girls with Turner syndrome without advancing the bone age.26 Although preliminary results suggest that adult height in Turner syndrome may be increased by this therapy, the effect of growth hormone on final height and possible long-term side effects are unknown.27

HEALTH SUPERVISION FROM 13 TO 21 YEARS OR OLDER: ADOLESCENCE TO EARLY ADULTHOOD

Examination and Laboratory Studies
1. Examine the adolescent for pigmented nevi, which may not be prominent in young children but tend to increase in frequency in adolescence and older ages. Pigmented nevi have primarily cosmetic significance. Advise removal of the nevi
Figure. Turner syndrome growth curve. ▲, healthy girls. Percentiles are derived from the National Center for Health Statistics. ●, untreated patients with Turner syndrome. Turner percentiles are from Lyon et al.²

if they are rubbed by clothing. Melanoma has been rare.²⁸

2. Check the adolescent annually for scoliosis.
3. Obtain an echocardiogram for aortic dilatation every 2 years or annually if a bicuspid aortic valve is present.
4. Continue to test the adolescent’s thyroid function every 1 to 2 years.
5. Monitor luteinizing hormone and FSH levels. Luteinizing hormone and FSH may be normal in childhood but are significantly elevated by 10–11 years in Turner syndrome. Evaluate the adoles-
Anticipatory Guidance

1. Consider referring the adolescent to a pediatric endocrinologist or tertiary care center for medical treatment of sex hormone replacement. The protocol for hormone replacement therapy needs to be designed in consultation with a pediatric endocrinologist.

2. Provide estrogen therapy, then cyclic therapy at an appropriate age, after checking to be certain that gonadotropin levels are elevated. If possible, wait until the adolescent is about 15 years of age to begin estrogen therapy to maximize height. Treatment, however, should be individualized to the patient’s psychosocial needs, as well as coordinated with growth-enhancing therapy when elected.

3. If lymphedema is exacerbated by estrogen therapy, suggest support hose and diuretics, which are sometimes helpful.

4. Orthopedic leg-lengthening procedures are sometimes recommended for patients with fused epiphyses.

5. Continue to monitor school function and behavior.

6. Discuss social adaptation. Girls with Turner syndrome tend to be socially immature for their age and need support in developing independence and social, particularly heterosexual, interactions. Support groups composed of girls with Turner syndrome are especially helpful. Provide psychosexual counseling.

7. Present information on reproductive options to having children, such as adoption and medically assisted reproduction.

8. Refer for genetic counseling and prenatal diagnosis the rare girl with Turner syndrome who has sufficient ovarian function to ovulate and who may become pregnant. These pregnancies are at increased risk for fetal chromosome abnormalities and miscarriage.29 Offer contraception advice when appropriate.

9. Facilitate transfer of the adolescent to adult medical care.

REFERENCES


24. Lin AE, Lippe BM, Geffner ME, et al. Aortic dilation, dissection, and...

SUGGESTED READINGS

Rosenfeld RG, Grumbach MM, eds. Turner Syndrome. New York: Marcel Dekker, Inc; 1990

BIBLIOGRAPHY AND RESOURCES FOR PARENTS
Turner Syndrome Society. c/o Lynn-Georgia Tesch, 15500 Wayzata Blvd, No. 768214, 811 12 Oak Ctr, Wayzata, MN 55391; 612/475-9944 or 800/465-6744.
Health Supervision for Children With Turner Syndrome
Committee on Genetics
*Pediatrics* 1995;96;1166

Updated Information & Services
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