Complete Summary

GUIDELINE TITLE

Health supervision for children with Turner syndrome.

BIBLIOGRAPHIC SOURCE(S)

Frias JL, Davenport ML. Health supervision for children with Turner syndrome. Pediatrics 2003 Mar;111(3):692-702. [72 references] PubMed

GUIDELINE STATUS

This is the current release of the guideline.

American Academy of Pediatrics (AAP) clinical reports automatically expire 5 years after publication unless reaffirmed, revised, or retired at or before that time.

COMPLETE SUMMARY CONTENT

SCOPE
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SCOPE

DISEASE/CONDITION(S)

Turner Syndrome and its associated conditions, including:

- Short stature
- Cardiovascular abnormalities
- Hypertension
- Hearing loss
- Strabismus
- Craniofacial abnormalities
- Obesity
- Glucose intolerance
- Urinary tract abnormalities

- Thyroid dysfunction and other autoimmune disorders
- Orthopedic problems
- Psychosocial needs

GUIDELINE CATEGORY

Counseling
Evaluation
Management

CLINICAL SPECIALTY

Endocrinology
Family Practice
Internal Medicine
Medical Genetics
Pediatrics

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

To assist the pediatrician in caring for the child in whom the diagnosis of Turner syndrome has been confirmed by karyotyping

TARGET POPULATION

Newborn, infant, child and adolescent females with Turner syndrome

INTERVENTIONS AND PRACTICES CONSIDERED

1. Referral to a geneticist, endocrinologist, and/or cardiologist, and performance of a renal ultrasound at the time of diagnosis
2. Anticipatory guidance, including early intervention services, stress support groups, and long-term planning
3. Ongoing medical evaluations, including prenatal evaluation for fetal growth retardation, growth measurements (height and weight) for infants and children, physical examinations, blood pressure measurements, thyroid screening, hearing screening, vision screening, skin assessment, and posture/spine evaluation
4. Addressing psychosocial issues, such as development and behavioral issues, school performance, socialization, and sexual issues
5. Assuring compliance with American Academy of Pediatrics recommendations for preventive pediatric health care (American Academy of Pediatrics Committee on Practice and Ambulatory Medicine. Recommendations for preventive pediatric health care. Pediatrics 2000;105:645)

MAJOR OUTCOMES CONSIDERED

Not stated

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

Not stated

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Not stated

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Not applicable

METHODS USED TO ANALYZE THE EVIDENCE

Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Not applicable

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Not stated

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The medical care of children with Turner syndrome requires ongoing assessment and periodic review of specific problems at appropriate ages. Specific recommendations are detailed below; refer also to Table 2 titled "Health Supervision Guidelines for Children with Turner Syndrome" in the original guideline document.

Prenatal Visit

When a prenatal diagnosis of 45,X Turner syndrome or another karyotype associated with Turner syndrome is detected, 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.

Medical evaluation for fetal growth retardation should be performed.

Health Supervision from Birth to 1 Month of Age: Newborns

1. 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 tissue samples 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 as described in the subsequent recommendations. (Note: for a complete list of clinical abnormalities in children with Turner syndrome, refer to Table 1 in the original guideline document.)
2. Examine the child's hips for dysplasia (Hall, 1989). Repeat the examination several times during early infancy.
3. Review results of the newborn hearing screening.
4. Obtain an initial consultation with a cardiologist with experience in pediatric cardiology for all persons with Turner syndrome. This includes patients who had "normal" results on prenatal ultrasonographic examination, because bicuspid aortic valve or coarctation could have been missed.
5. Check blood pressure and peripheral pulses during each physical examination. A careful comparison of arm and leg systolic pressure is important to evaluate for possible coarctation.
6. Perform a renal ultrasound (or repeat the ultrasound if it was done prenatally). If the patient is found to have an abnormality of the urinary tract, continue monitoring for urinary tract infections and renal function.

7. Distinguish the anomalies having major implications on the medical treatment (e.g., cardiac and renal anomalies) from those of primarily cosmetic and psychologic importance.
8. Inform the family that lymphedema may persist for months or longer and may recur.
9. Discuss the possibility of feeding problems. Some infants with Turner syndrome have inefficient sucking and swallowing reflexes because of impaired oral motor function (Mathisen, Reilly, Skuse, 1992).
10. Obtain an initial consultation with a pediatric endocrinologist to 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 women with Turner syndrome to have children (Hovatta, 1999).
11. Discuss subacute bacterial endocarditis prophylaxis if a cardiac anomaly is present.
12. Talk about how and what to tell other family members and friends.

Health Supervision from 1 Month to 1 Year of Age: Infancy

1. Assess the infant's weight, taking into account that many infants with congenital lymphedema lose weight during the first month of life because of diuresis.
2. Check blood pressure and peripheral pulses during each physical examination. A careful comparison of arm and leg systolic pressure is important to evaluate for possible coarctation.
3. Perform an ophthalmologic evaluation. Nonalternating strabismus may be present. Refer the infant to an ophthalmologist as soon as strabismus is suspected.
4. On every visit, check for serous otitis and otitis media and evaluate the child's hearing (objectively at 6 and 12 months of age and before 4 months of age if newborn screening was not performed). If sensorineural hearing loss is identified, recommend hearing aid evaluation, which can be conducted as early as 3 months of age (Barrenasa, Landin-Wilhelmsenb, & Hansonc, 2000; Roush, Davenport, & Carlson-Smith, 2000).
5. If a cardiac anomaly is present, have the patient followed up as frequently as recommended by her cardiologist. Provide prophylaxis for subacute bacterial endocarditis, although subacute bacterial endocarditis is rare in infancy.
6. 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.
7. Consider referring the infant to an appropriate pediatric specialist if renal or eye abnormalities are found.
8. If hypertension is present, treat it aggressively and perform a careful search for cardiovascular or renal causes.
9. Counsel the parents regarding environmental factors that may increase the risk of otitis media, such as bottle feeding, passive smoking, and group child care facility attendance. If otitis media is present, institute aggressive treatment. Discuss the option of tympanostomy tubes for persistent otitis media and consider referral to an otolaryngologist.
10. Refer the infant to developmental intervention programs if neuromuscular development is delayed.

Health Supervision from 1 to 5 Years of Age: Early Childhood

1. Follow the child's growth and have her evaluated by a pediatric endocrinologist if growth failure occurs. The age at which GH therapy is initiated varies but can be considered at as early as 2 to 3 years of age for girls who are below the fifth percentile for height in the growth chart for healthy girls of the same age. Early initiation of GH therapy may allow for greater gain and normalization of the timing of puberty. Plot growth on the Turner syndrome-specific growth curve starting at 2 years of age.
2. Evaluate the child's speech and refer the child to a speech therapist when appropriate. Consider hearing loss as a possible cause of speech delay.
3. If a cardiac anomaly is present, have the patient followed up as recommended by her cardiologist.
4. Check blood pressure and peripheral pulses during each physical examination. A careful comparison of arm and leg systolic pressure is important to evaluate for possible coarctation.
5. Evaluate the child's hearing and check for serous otitis and otitis media during every visit.
6. Continue to evaluate the child's renal status (urinalysis and culture, as indicated) if a renal anomaly is present.
7. Test for thyroid function by measuring thyroid-stimulating hormone and free or total thyroxine levels at 1- to 2-year intervals because of the increased frequency of hypothyroidism usually caused by autoimmune thyroiditis. In the absence of clinical signs, initiation of thyroid function testing may be delayed until after 4 years of age or later, because hypothyroidism is rare before then (Elsheikh, Conway, & Wass, 1999).
8. Evaluate the child for possible developmental delay and learning difficulties, particularly spatial perception problems. An 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.
9. If hypertension is present, treat it aggressively and perform a careful search for cardiovascular or renal causes.
10. Counsel the parents regarding environmental factors that may increase the risk of otitis media, such as bottle feeding, passive smoking, and group child-care facility attendance. If otitis media is present, institute aggressive treatment. Discuss the option of tympanostomy tubes for persistent otitis media and consider referral to an otolaryngologist

Health Supervision From 5 To 13 Years Of Age: Late Childhood

1. Discuss the diagnosis and treatment of Turner syndrome with the child as soon as she is able to understand as well as with the parents.
2. Continue to monitor growth carefully. In addition to GH, the endocrinologist may consider adding oxandrolone to the GH treatment regimen in older girls (9-12 years of age) with extreme short stature or those in whom the response to GH is not adequate (Rosenfeld et al., 1998).
3. Monitor the child for urinary tract infections if urinary tract abnormalities are present.

4. Check blood pressure and peripheral pulses during each physical examination. A careful comparison of arm and leg systolic pressure is important to evaluate for possible coarctation.
5. Evaluate the child's hearing and check for serous otitis and otitis media during every visit. Advise patients to protect their hearing by avoiding loud noises.
6. Check the child's dentition for malocclusion.
7. Continue testing for thyroid function at 1- or 2-year intervals.
8. Check for scoliosis yearly. Lordosis and kyphosis are also seen more frequently in girls with, than in girls without, Turner syndrome.
9. 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.
10. Discuss adjustment to short stature with the parents and separately with the child.
11. If hypertension is present, treat it aggressively and perform a careful search for cardiovascular or renal causes.
12. If otitis media is present, institute aggressive treatment. Discuss the option of tympanostomy tubes for persistent otitis media and consider referral to an otolaryngologist. Advise patients to protect their hearing by avoiding loud noises.
13. Counsel the family regarding the importance of optimizing bone density, and advise that the patient receive the recommended daily requirements for Vitamin D and calcium through diet or supplementation.

Health Supervision from 13 to 21 Years or Older: Adolescence to Early Adulthood

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 if they are rubbed by clothing. The risk of melanoma, which is low, does not appear to be increased by GH treatment (Gare et al., 1993; Wyatt, 1999).
2. Check blood pressure and peripheral pulses during each physical examination. A careful comparison of arm and leg systolic pressure is important to evaluate for possible coarctation.
3. Measure fasting lipid profile at least once during adolescence to screen for hyperlipidemia, especially in those who have a positive family history, have diabetes mellitus, or are overweight.
4. Evaluate the child's hearing and check for serous otitis and otitis media during every visit. If otitis media is present, institute aggressive treatment. Advise patients to protect their hearing by avoiding loud noises.
5. Check the adolescent annually for scoliosis and kyphosis.
6. Even for individuals with normal initial cardiac evaluation, refer to a cardiologist for a complete evaluation, including a close evaluation of the aortic root.
7. Continue to test the adolescent's thyroid function every 1 to 2 years.
8. Evaluate the adolescent for development of secondary sex characteristics. As many as one third of girls with mosaicism will enter puberty spontaneously. Measurement of luteinizing hormone (LH) and FSH concentrations may be helpful in assessing gonadal function (Pasquino et al., 1997). Increased LH

and FSH concentrations will help to confirm that the child is physiologically ready for initiation of estrogen therapy, and evidence of suppression of LH and FSH into the normal range may help to determine adequacy of the maintenance dose chosen.

9. Refer the adolescent to a pediatric endocrinologist for evaluation of sex hormone replacement. If gonadotropin levels are increased, initiate estrogen therapy and then begin cyclic therapy with a progestin at an appropriate age. Estrogen therapy may be initiated as early as 12 years of age for girls who are satisfied with their height. For others, estrogen therapy can be delayed until as late as 15 years of age to maximize height. Individualize treatment choices concerning the timing of feminizing therapy and length of growth enhancing therapy to the patient's psychosocial needs.
10. If hypertension is present, treat it aggressively and perform a careful search for cardiovascular or renal causes.
11. If otitis media is present, institute aggressive treatment.
12. If lymphedema is exacerbated by estrogen therapy, combined decongestive therapy that uses manual lymphatic drainage, low-stretch support garments, and exercises may be effective (Lerner, 1998). Some physicians also use diuretics.
13. Continue to monitor school function and behavior.
14. Discuss social adaptation. Girls with Turner syndrome tend to be socially immature for their age and need support in developing independence and social interactions. Support groups of girls with Turner syndrome are especially helpful. Provide psychosexual counseling.
15. Present information on reproductive options to bearing children, such as adoption and medically assisted reproduction.
16. Provide counseling regarding sexuality and sexually transmitted diseases.
17. Refer the rare girl with Turner syndrome who has sufficient ovarian function to ovulate and who may become pregnant for genetic counseling and prenatal diagnosis (if pregnant). These girls are at increased risk of having a fetus with chromosome abnormalities and having miscarriages (Tarani et al., 1998). Offer contraception advice when appropriate.
18. Facilitate transfer of the adolescent to adult medical care.

In addition to the recommendations detailed above, this guideline recommends that physicians ensure compliance with the American Academy of Pediatrics "Recommendations for Preventive Pediatric Health Care" (Pediatrics 2000;105:645).

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

REFERENCES SUPPORTING THE RECOMMENDATIONS

References open in a new window

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of evidence supporting each recommendation is not specifically stated.

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

Appropriate management of children with Turner syndrome

POTENTIAL HARMS

Lymphedema exacerbation caused by estrogen therapy

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

The recommendations in this report do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Living with Illness

IOM DOMAIN

Effectiveness
Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

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GUIDELINE DEVELOPER(S)

American Academy of Pediatrics - Medical Specialty Society

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American Academy of Pediatrics

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FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline.

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GUIDELINE AVAILABILITY

Electronic copies: Available from the American Academy of Pediatrics (AAP) Policy Web site.

Print copies: Available from American Academy of Pediatrics, 141 Northwest Point Blvd., P.O. Box 927, Elk Grove Village, IL 60009-0927.

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This NGC summary was completed by ECRI on August 18, 2003. The information was verified by the guideline developer on September 8, 2003.

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