

Turner Syndrome



A Guide for Families

TURNER SYNDROME: A GUIDE FOR FAMILIES

Patricia Rieser, CFNP and Marsha Davenport, MD



Patricia Rieser is a Family Nurse Practitioner who worked in Pediatric Endocrinology at the University of North Carolina, Chapel Hill, for twelve years. She helped form the Pediatric Endocrinology Nursing Society and has been involved with family support groups throughout her career.

Marsha Davenport, MD, is an Associate Professor of Pediatric Endocrinology at the University of North Carolina, Chapel Hill, where she directs the Turner Syndrome Clinic. She has been involved with TSS-US for many years.

TURNER SYNDROME: A GUIDE FOR FAMILIES

The authors would like to thank all the girls and families who have taught us so much about living with Turner syndrome; the staff of TSS-US for supporting this project; Martha Mallory (Spring, TX) for the Spanish translation; Greg Carter (Raleigh, NC) for graphic design; Ellen Jones, MEd (Buffalo, NY), for book suggestions; and Eli Lilly and Company, for providing funding and help with graphics.

We also appreciate the valuable input provided by these reviewers:

Dennis Agle is the father of a 14-year-old with TS and is a past Board member and newsletter editor of TSS-US. He lives with his family near Los Angeles, CA, where he is a bank consultant.

Tina Jones is the mother of a 12-yr-old daughter who has TS. Involved with TSS-US since 1990, Tina has served on the Board of Directors for many years, serving as Secretary, then President. Tina lives with her husband and four children in Casper, WY, where she is a commission stained glass artist and teacher.

Peggy LeBrun is the mother of two children, including a 12-year-old daughter who has Turner syndrome. She is a public relations consultant living near Milwaukee, WI, and she has been on the Board of TSS-US since 1997.

Jessica Lord is a woman with TS who has been a member of TSS-US since the late 1980s, currently serving on the Board. She is training to be a clinical pediatric neuropsychologist at the University of Connecticut and hopes to work with children who have developmental and medical disabilities.

Mary Gwyn Roper, MD, is a physician with TS receiving her training as a pediatric endocrinologist. She lives in Chapel Hill, NC, with her husband and has been involved with TSS-US since 1994.

David Sandberg, PhD, is a pediatric psychologist and researcher who specializes in helping children with endocrine disorders and their families. He works at the Children's Hospital of Buffalo and the University of Buffalo (NY) and has been involved with local and national family support groups for many years.

Merriott Terry has been Executive Director of TSS-US for two years, continuing a career in the non-profit, business and education sectors. She lives in a suburb of Houston with Jack, her husband of 32 years. They have two grown sons.

Louis Underwood, MD, is Professor of Pediatrics and former Division Chief of Pediatric Endocrinology at the University of North Carolina, Chapel Hill. He has been involved with several parent support groups in many capacities throughout his long career.

Introduction	2	How to use this guide
	2	The faces of Turner syndrome
The Basics	3	What is Turner syndrome?
	4	What causes TS?
	4	The nuts and bolts of Xs and Ys
	6	The genetics of TS
	7	How is TS diagnosed?
	7	Before birth
	8	During infancy
	8	During childhood and adolescence
Growth and Development	8	During adulthood
	9	Growth
	9	Growth in TS
	12	Growth hormone treatment
	14	Other physical features
	15	Sexual development
	15	Sexual development in TS
	16	Treatment with female hormones
	16	Fertility
	17	Intelligence and school performance
18	Social and emotional development	
Other Health Issues	20	Heart and blood vessels
	21	Kidneys
	21	Ears and hearing
	22	Eyes and vision
	22	Bones and teeth
	23	Immune system
	24	Metabolism
	24	Skin
Social and Emotional Support	25	General issues
	26	Health care issues
	27	Growth-related issues
	28	Social issues
	29	Sexuality and fertility Issues
	30	Educational and career issues
Wrapping Up	31	Summary
	32	Resources
	34	Glossary
	36	Appendix - Health care checklist

INTRODUCTION

How to use this guide

This booklet is intended for families who are affected by Turner syndrome* (TS). It contains a great deal of material, *some* of which will apply to most girls with TS and *much* of which will apply to *some* girls. TS is a highly variable condition. Every girl with TS is unique and no generalization will apply to every girl, no matter how accurate it is for the group. Most girls with TS are healthy and well-adjusted; some face special physical, emotional, social or learning challenges. All will benefit from good medical care, informed emotional support and careful screening for related conditions.

We suggest that you read this book cover-to-cover, then make a list of questions and concerns to discuss with your child's doctor or health care team. Share it with family members, friends, teachers, counselors and coaches. Together, you'll support your daughter's healthy growth in every way.

Words convey attitudes, so we have chosen to "put the person first" throughout this guide. We talk about girls with TS, not TS girls, to emphasize that people are not defined by a medical condition. When we talk about girls with TS, remember that we are generalizing and that every girl is unique. Thank you!

** Glossary words are bold and underlined the first time they are used.*

The faces of Turner syndrome



Sarah is a bright-eyed, alert newborn girl. Her parents and doctor notice right away that her hands and feet are unusually puffy and that she has extra folds of skin at the back and sides of her neck. Her doctor knows that these features are often signs of a condition called Turner syndrome, so she orders a blood test to find out if this is the case. Sarah's parents are worried, although the doctor assures them that Sarah is a strong, healthy baby.

Emily is 7 years old and has an ear infection. She has just seen a pediatrician for the first time in several years. When the doctor plots her height on a growth chart, he notices that she is shorter than most girls her age. He also observes that her fingernails are narrow and slant upward. He tells Emily's parents that he suspects she may have Turner syndrome and wants to refer her to a pediatric endocrinologist (a doctor who has special training in caring for children with growth and hormone problems).

Donna is 14 years old and is quite concerned because her chest is still as flat as her 8-year-old sister's. She is the second shortest girl in her class, but her height does not bother her as much as her lack of breast development. She is struggling in school and feeling lonely since her one good friend moved away. Her doctor is concerned, too, and draws a blood sample from her arm for a karyotype, the blood test used to diagnose Turner syndrome.



Martha is 24 years old and stopped having menstrual periods several months ago. Her gynecologist has checked for most of the common causes of amenorrhea (absence of menstrual periods), and the results of some of the blood work she's obtained are surprising – it appears that Martha's ovaries are becoming menopausal already. Martha is barely 5 feet tall and when the doctor examines her more carefully, she notices unusual fingernails and a high, arched palate (roof of the mouth). It occurs to her that Martha may belong to the group of women with Turner syndrome who go through puberty normally, only to have their ovaries fail in early adulthood. The results of a karyotype analysis confirm the diagnosis.

These four individuals do not appear to have much in common – they are different ages and do not look or behave at all alike – but each of them has Turner syndrome (TS). The purpose of this booklet is to answer some of the questions you and your daughter may have about this condition. Much of the information may be useful for adult women with Turner syndrome, as well, though it is beyond the scope of this booklet to address adult issues in depth. Your child's doctor is best qualified to answer specific questions about your child, but the general information you will find here may help you better understand this condition and what it may mean for your child and family.

THE BASICS

What is Turner syndrome?

A syndrome is a set of features or symptoms that often occur together and are believed to stem from the same cause. In 1938, Dr. Henry Turner published a report describing seven girls who shared a number of features, including short stature, lack of sexual development, cubitus valgus (arms that turn out slightly at the elbow), webbing of the neck and a low hairline in the back. The condition he described came to be known as Turner's syndrome or, as it is known today, Turner syndrome. Some people refer to TS as gonadal

dysgenesis because one of its main features is lack of development (dysgenesis) of the ovaries (female organs that store eggs and produce sex hormones; also known as gonads).

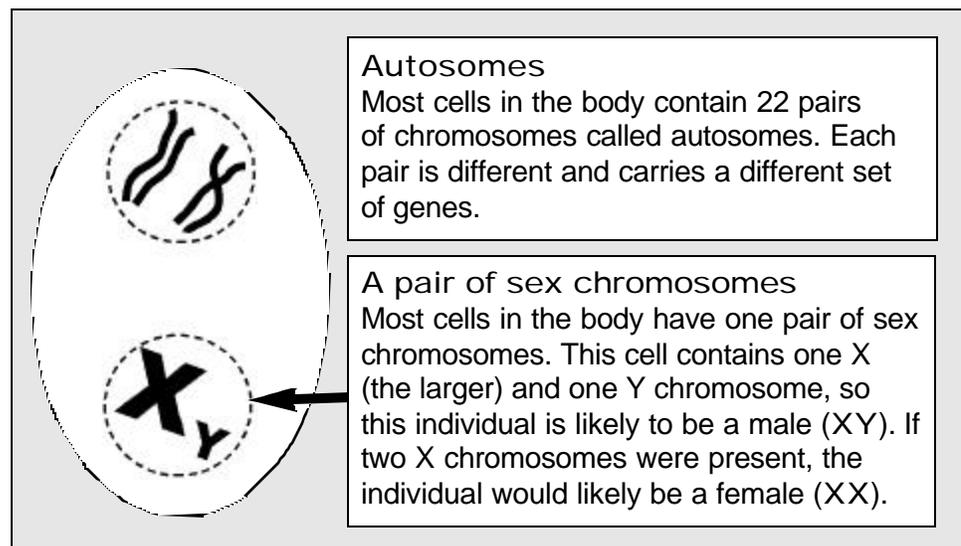
In 1959, Dr. C.E. Ford discovered that the cause of TS is a chromosomal (genetic) condition involving the sex chromosomes. He observed that most girls with TS he examined did not have all or part of one of their X chromosomes, and he suggested that the missing genetic material accounted for the physical findings observed in this condition. The diagnosis of TS is made on the basis of a blood test called a karyotype analysis, a picture taken under a microscope of the chromosomes in a person's cells. TS is one of the most common genetic conditions, affecting 1 of every 2,000 to 2,500 girls. Fortunately, most girls with TS can expect to lead healthy, productive and happy lives.

What causes TS?

The nuts and bolts of Xs and Ys

To understand the cause of TS, it will help to learn a bit about genes and chromosomes. Our bodies are made up of countless microscopic cells, each containing thin, rod-like structures called chromosomes. These chromosomes contain even smaller units called genes. Genes are like the computer programmers of our cells; they contain the messages that instruct the cells what to do and when to do it. Genes are responsible for the color of our eyes and hair, our height

Figure 1.
Genes in the
nucleus of a cell

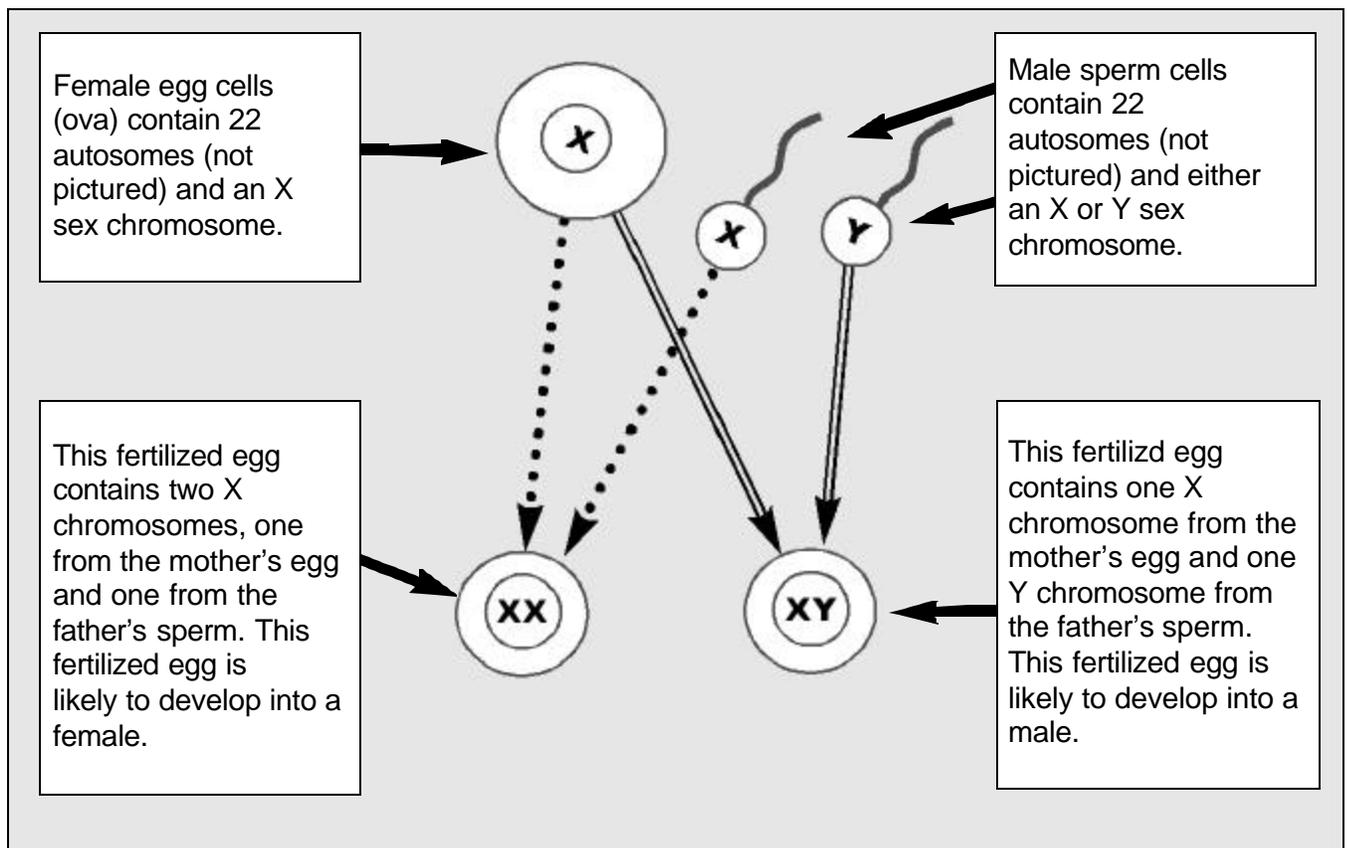


and body build, our growth and development – everything about us relates to our genes in one way or another.

Every normal cell contains 22 pairs of chromosomes called autosomes and one pair of sex chromosomes. A sex chromosome may be an X or a Y – usually, females have two X chromosomes and males have one X and one Y (Figure 1).

The reproductive cells (sperm in men and eggs/ova in women) contain only 23 chromosomes, one member of each of the 23 pairs. Each egg contains 22 autosomes and an X sex chromosome, and each sperm contains 22 autosomes and either an X or a Y sex chromosome. When the egg and sperm join at the time of conception, the normal zygote (fertilized egg at the earliest stage of growth) contains a full set of 46 chromosomes – 44 autosomes (22 from each parent) and two sex chromosomes (one from each parent). If two X chromosomes are present, the baby will be female (or 46XX in gene-talk); if one X and one Y are present, the baby will be male (46XY) (Figure 2).

Figure 2.
Boy or Girl?



The sex chromosomes (X and Y) contain many genes, a few of which are responsible for creating the different physical appearance and characteristics we associate with males and females. Some of these genes trigger the development of sex organs (ovaries in females and testes in males) which produce the hormones that result in sexual development. In 46XX females, most of either the mother's or father's X chromosome is randomly inactivated (has its genes turned off) in each cell in the body except for reproductive cells. However, a few genes that are similar to genes on the Y chromosome remain active. Two complete X chromosomes are needed for the normal growth and development of girls.

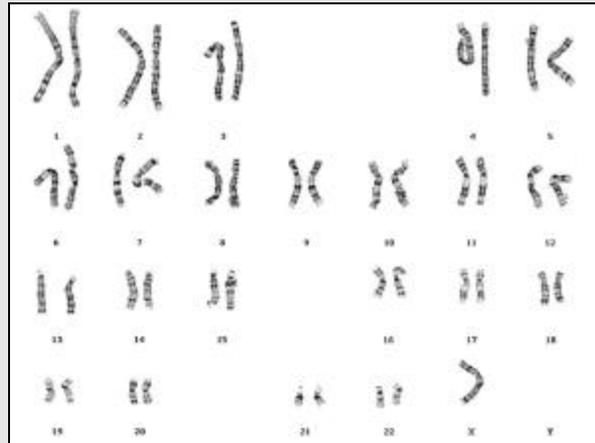
The genetics of TS

Turner syndrome results when all or part of one of the X chromosomes is lost before or soon after the time of conception. There is nothing that parents do to cause or increase the risk of TS in their daughters and nothing they can do to prevent it. In about half the cases of TS, one of the sex chromosomes is missing entirely, leaving 45 chromosomes instead of the usual 46. This karyotype is written as 45X (referred to in the past as 45XO). Girls with this

Figure 3. Karyotypes
 karyotype tend to have noticeable features of TS and often are diagnosed soon after birth (Figure 3).



Normal female karyotype



Turner syndrome karyotype

Turner syndrome occurs when only one normal X chromosome is present. The other X chromosome may be missing entirely in some or all body cells, or it may be present, but incomplete or altered.

About 30% of girls with TS have some cells that have the normal number of chromosomes (46XX) and others that are missing an X (45X); this is thought to result from the loss of an X chromosome in some cells after fertilization. The presence of more than one chromosome pattern in an individual is called mosaicism. 45X/46XX mosaicism can be hard to diagnose because many body cells have the usual 46XX pattern. These girls may have fewer features of TS than girls whose cells are all missing an X chromosome. A small percentage of girls with TS have some Y chromosome material. Girls with all or part of a Y chromosome in any of their cells are at increased risk of developing tumors in their poorly developed gonads even during childhood, so surgical removal of the gonads generally is recommended.

Another 20% of girls with TS have both X chromosomes, but one of them is incomplete or altered. Most of these alterations occur soon after conception and reflect a loss of genetic material from one of the X chromosomes. These include ring chromosomes (X chromosome is shaped like a ring, with ends joined), isochromosomes (X chromosome with two long arms instead of a long arm and a short arm) and deletions (part of the chromosome is missing).

For decades, scientists have tried to find out which genes on the X chromosome cause the features of TS. In 1997, the first of these genes, the SHOX (short stature homeobox-containing) gene, was discovered. Loss of this X chromosome gene seems to play a key role in causing the short stature of girls with TS. Because this gene is expressed during fetal development (before birth) in the limbs (especially elbow, knee and wrist), jaw bones and ears, it probably causes some of the other features of TS, as well. Other genes that are involved in TS may be discovered soon.

How is TS diagnosed?

Before birth

TS may be detected “accidentally” during pregnancy if a karyotype is performed on cells collected during amniocentesis or chorionic villous sampling (CVS); these tests often are ordered because the mother is older (which does NOT increase the risk of TS). TS also may be discovered when a prenatal ultrasound shows

that the fetus has physical conditions common to TS, such as cystic hygroma (a collection of lymph fluid around the neck) or heart defects, and a prenatal karyotype confirms TS. Miscarriage may occur if the problems are severe.

When the diagnosis is made early in the pregnancy, parents may struggle with the very personal and difficult decision about whether to have the baby. They may find it helpful to talk with families who have a child with TS, adults who have TS, a pediatric endocrinologist and their genetic counseling team. The Turner Syndrome Society of the US (TSS-US), listed under *Resources*, can help parents in this challenging situation to find support and sound medical information.



During infancy

Roughly half of the girls with TS are diagnosed at birth or during infancy, usually because of the presence of typical physical features, lymphedema (swelling due to excess lymphatic fluid, especially of the hands and feet) or a heart condition sometimes seen in TS. The baby's doctor should order a karyotype to confirm the diagnosis, even if one was obtained before the baby was born.

During childhood and adolescence

Some girls do not have obvious physical signs of TS at birth and may not be diagnosed until later in childhood, often because they have unexplained short stature or poor growth. Some girls may come to the attention of a health care provider because they do not go into puberty - that is, they do not develop breasts or their breast development begins, but they do not menstruate. An evaluation by a pediatric endocrinologist may uncover subtle signs of TS in these girls that were not apparent to their general health care providers.

If the results of a blood karyotype are normal in a girl with unexplained short stature or delayed puberty but the doctor remains suspicious of TS, a tiny piece of skin may be removed so that the chromosomes in cells different from those in blood can be examined. This test may be needed to confirm the diagnosis of mosaicism, in which some cells have normal chromosomes and others do not.

During adulthood

In a small number of women with TS, puberty occurs normally, but at some point, their ovaries stop functioning, hormones are not produced and menstruation ceases. Some of these women are

diagnosed when they see a gynecologist because their periods have stopped or become irregular or because they are having trouble getting pregnant. The doctor may order some hormone tests that can suggest TS, but a karyotype is the only way to diagnose the condition with certainty.

GROWTH AND DEVELOPMENT

Growth



As for any child, issues related to growth and development are central to the care of children with TS. This section will highlight three areas that are often of concern in girls with TS: statural growth, sexual development and the development of the skills required to do well in school and relationships.

Growth in TS

Short stature is the most common feature of TS. Girls with this syndrome generally are slightly small at birth – their average length is less than 18.5 inches (47 cm) compared to 20 inches (51 cm) for other newborn girls – and they tend to grow quite slowly during infancy and early childhood. About 50% of these girls fall below the 5th percentile in height by 1.5 years and 75% by 3.5 years of age. Girls with mosaicism vary more in their growth, but 50% still fall to below the 5th percentile by about 2 years of age. Growth continues to be slower than normal throughout childhood, so that the difference in height between a girl with TS and other girls the same age increases with time.

Many girls with TS have a delayed bone age, which means that their bones are more like those of younger girls. Puberty in girls usually begins when the bone age is about 11 years. Female hormones (estrogens) made by the ovaries during puberty (or taken by the child in pill or other form) speed up bone growth and maturation. Physical growth stops when the growth plates of the bones fuse together, which happens at a bone age of about 15 years. Most girls with TS who are not treated with any hormones will not have a pubertal growth spurt and may continue to grow at a slow rate until they are in their twenties (Figure 4).

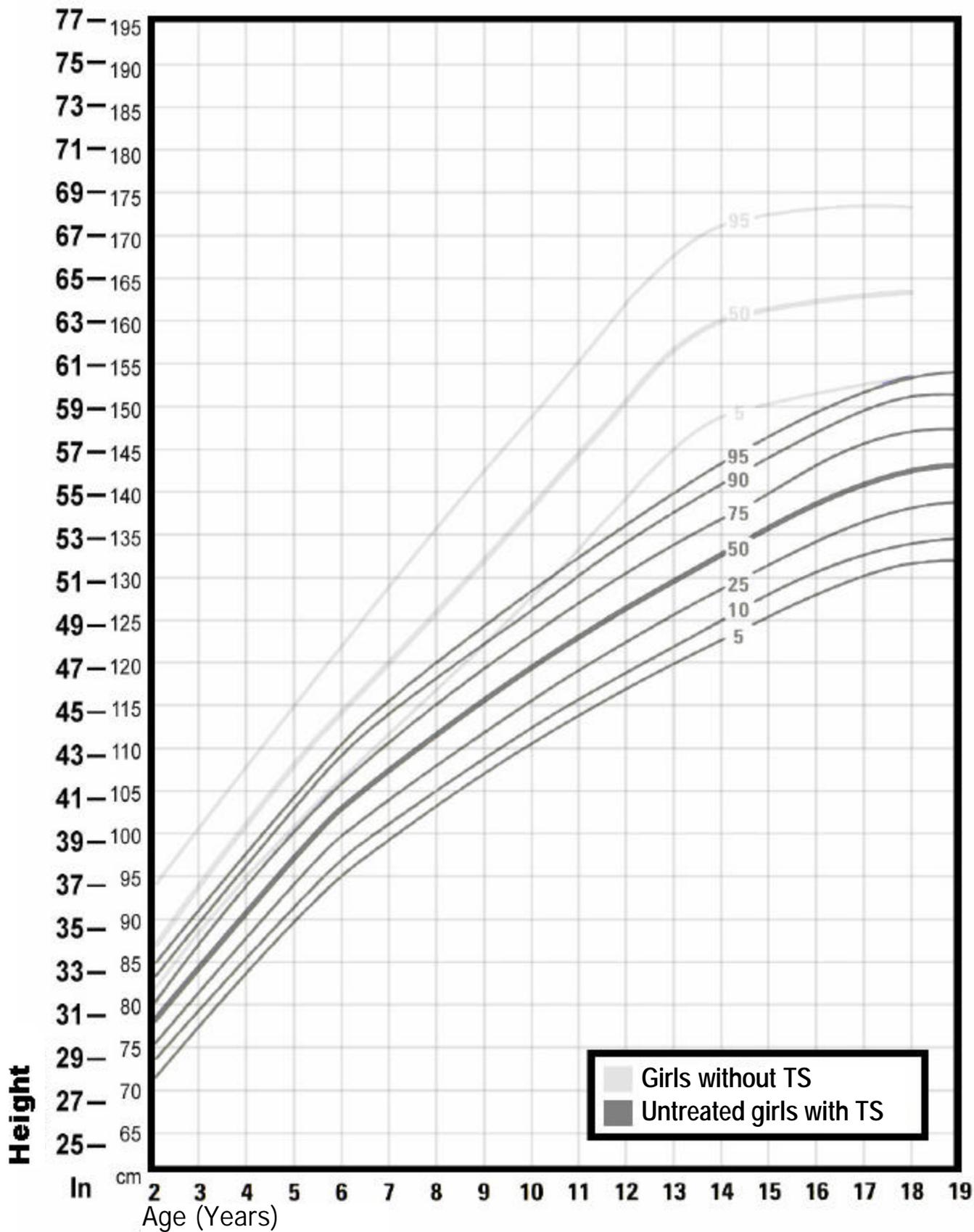
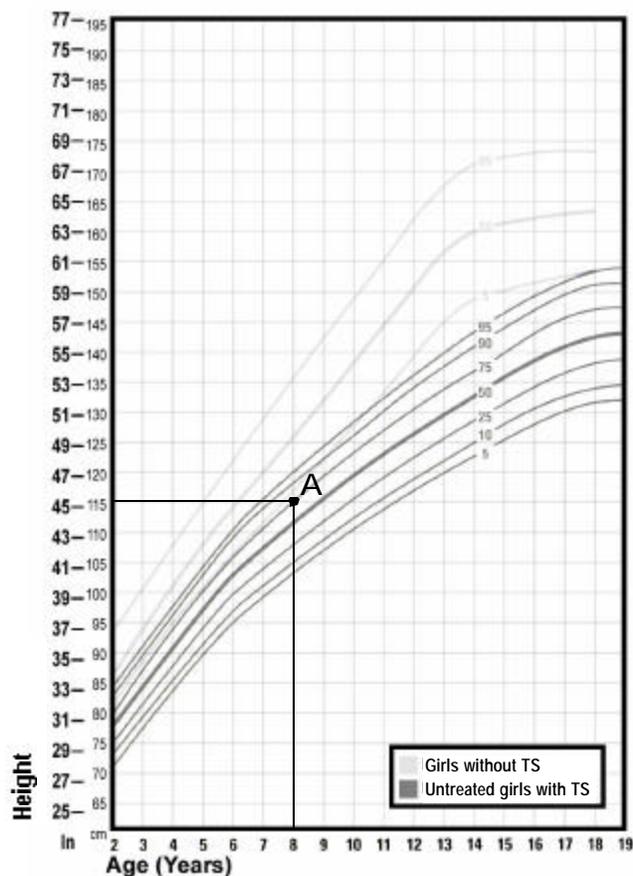


Figure 4.
Turner syndrome
growth chart



Growth Charts: Growth charts are used to compare the height of one child with the heights of other children the same age and sex. This growth chart shows the range of height for girls with Turner syndrome and the range of height for other girls.

The top light gray line marked “95” is the 95th centile line for normal girls. This means that a girl whose height is on that line is taller than 95% of girls her age. The 50th centile light gray line represents the average height for a given age.

Turner syndrome growth chart: The area between the top and bottom light gray lines represents the range of height for most girls. The white area between the top and bottom dark gray lines represents the range of height for most untreated girls with Turner syndrome.

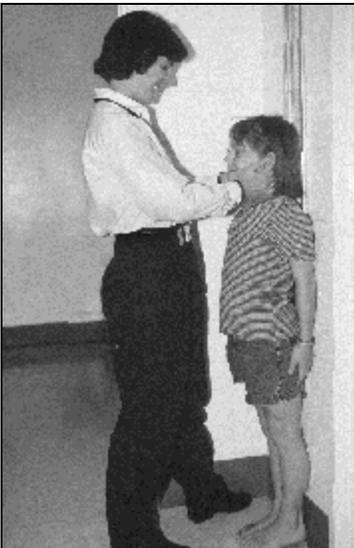
How to use a growth chart: To use a growth chart, find the child’s age along the bottom of the chart and draw a vertical line there. Find the child’s height in inches or centimeters along the side of the chart and draw a horizontal line there. The point where these two lines intersect is the child’s height for age.

Example: Nancy is an 8-year-old with Turner syndrome who is 45.5 inches (115.5 cm) tall (Point A on the growth chart). Her height is just below the 5th centile on the girls’ growth chart, which means that she is shorter than about 95% of normal girls her age. Her height is at the 75th centile on the Turner syndrome chart, which means that she is taller than about 75% of 8-year-old girls with Turner syndrome.

The average adult height of an untreated woman with TS is 4 feet, 8 inches (142 cm), although a few women reach 5 feet (152 cm). The heights of the parents affect the height of the daughter – a girl with tall parents is likely to be taller than one who has short parents. Girls with TS who are not treated with growth hormone end up about 8 inches shorter as adults than would have been predicted based on their parents' heights. All girls with TS should have their growth tracked carefully and plotted on the TS growth chart at regular intervals.

Growth hormone treatment

One of the major changes in the treatment of TS in recent years is based on the results of many research studies involving the use of biosynthetic growth hormone (GH) in these girls. The US Food and Drug Administration joined many other countries in approving the use of GH for girls with TS in 1996. Since then, GH has become a standard part of the treatment of TS. We now know that most girls with TS will grow faster and may reach a normal adult height (over 5 feet) if treated early enough and long enough with GH, although results for individuals vary greatly. Researchers are continuing to look for the safest and most effective ways to support normal growth and puberty in girls with TS, so treatment recommendations may change as more is learned. Participation in the TS Registry is one way to help researchers evaluate and improve treatments (see back cover).



Current guidelines suggest that GH treatment should be considered once the girl's height drops below the 5th percentile on the standard growth chart, even as young as 2 years of age. A higher standard dose is suggested for girls with TS than for GH-deficient patients, and some doctors increase the dose further if the girl's growth slows (while checking carefully for possible side effects).

Androgens (male-type hormones) normally are made by both boys and girls. In girls, the adrenal glands start to make androgens around the age of 8-9 years and the ovaries make more at the time of puberty. Together, these androgens promote normal growth of sexual hair, increase muscle mass and cause a growth spurt. The results of studies have shown that a low dose of androgens (often with a drug called oxandrolone), given along with GH, increases growth in girls with TS without causing signs of too much androgen (such as acne or facial hair). Androgens generally are given to girls with TS

over the age of 9 years, when androgen levels should be rising normally. Androgens should not be used alone (without GH) in girls with TS because they do not improve adult height by themselves.

Estrogens (female-type hormones) are not effective in enhancing growth. Once a girl begins taking estrogens to bring about puberty, her bone age advances more quickly and the clock ticks faster towards the time when her growth stops. If a girl does not begin puberty on her own, the decision of when to begin estrogens is one each girl and her parents must make, trying to balance continued growth and adult height with starting puberty at a close-to-normal time. The average girl without TS begins breast development between 10 and 11 years of age and has her first menstrual period between 12 and 13 years, although some do not start breast development until the age of 13.5 years.



Delaying the start of puberty until the time when most girls are ending it may affect a girl's self-image, her friendships with peers and her healthy sexual development. When to start estrogen is an important decision, and families should discuss the various aspects of it openly with each other and with the professionals who are part of the girl's support team. No one answer is right for every girl.

The best outcome in terms of increased adult height depends on several things:

- early diagnosis of TS
- good overall health and nutrition
- prompt treatment of poor growth with an appropriate dose of GH
- the addition of low-dose oxandrolone in late childhood (for some girls)
- relatively late start of estrogen therapy (this must be balanced with the importance of near-normal pubertal development).

All growth-related hormone treatments should be supervised by a pediatric endocrinologist, who will follow the girl closely to monitor her growth and check for rare, but possible, unwanted effects of treatment. Side effects from GH occur in fewer than 2 out of 1,000 girls, but include: severe headaches and vomiting caused by increased pressure in the brain; edema; insulin resistance and a rise in blood

sugar; worsening of scoliosis; damage to the growth plate in the hip (slipped capital femoral epiphysis or SCFE); and faster growth of moles (the latter three usually occurring during periods of rapid growth). Oxandrolone (androgens) given in high doses can cause acne, deepening of the voice, growth of facial hair, insulin resistance and rapid advance of bone age.

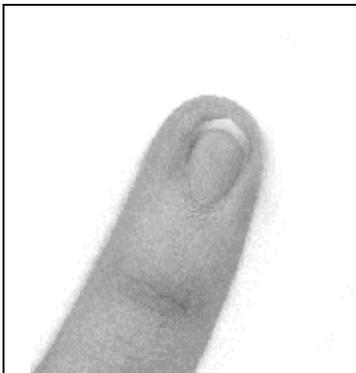
Other physical features

Many unusual physical features may be observed in girls with TS. A feature that is very noticeable in one girl may be almost invisible in another. Many girls have only a few of these features, and some may not have any obvious ones at all. Some of the features occur because of changes in the normal growth of bones before and after birth. Here is a list of some common features, including medical terms and descriptions:



- stocky appearance
- narrow, high-arched palate (roof of the mouth) and crowded teeth
- retrognathia (receding lower jaw)
- broad chest with widely-spaced nipples that may be inverted (turned inward instead of sticking out)
- cubitus valgus (arms that turn out slightly at the elbow)
- "short" ring finger (the hand bone that joins the ring finger is short, not the finger itself)
- pectus excavatum (funnel chest)

Other unusual physical features result from underdevelopment of the lymphatic system before birth. The lymphatic system carries fluid that has leaked into the tissues back into the veins. If this system is not working properly, the fluid collects in the tissues and is called lymphedema. In girls with TS, this fluid often collects around the neck before birth and may affect the appearance of the neck and ears. After birth, it often remains in the hands and feet. Some of these other features are listed below:



- thick, short neck, "webbed" appearance
- low hairline in the back of the neck
- prominent ears
- soft, narrow fingernails and toenails that point upwards

Some of these features may be barely noticeable, but others may cause self-consciousness or embarrassment for the girl. Some of them can be improved with treatment. For example, lymphedema may improve with manual compressive therapy (massage, exercises, support garments and skin care). A small number of girls have plastic surgery to improve the appearance of the neck and ears (though the surgeon should be aware of the risk of excess scar formation). A make-up and wardrobe consultant may be able to help the older girl or woman with TS look her best by drawing attention to her positive features.

Sexual development

Sexual development in TS



Lack of sexual development (breast development, feminine body contours and menstruation) during adolescence is another hallmark feature of TS. Sexual development is affected because the ovaries (female sex organs) often stop working early in life. Two major functions of the ovaries are to produce the female sex hormones, estrogen and progesterone, and to store eggs and release them on a regular basis once menstruation begins. If the ovaries do not produce their hormones, sexual development will not occur unless these hormones are replaced with medication. Few or no eggs are stored in the ovaries, so spontaneous pregnancy is rare and carries a high risk of genetic problems in the baby. The other female reproductive organs (fallopian tubes, uterus or womb and vagina or birth canal) are present and function normally.

Ten to 15 percent of girls who have a 45X karyotype and about one-third of girls with mosaicism have some signs of breast development in their early teens, and some will menstruate. In these girls, sexual development and menstruation usually stop sooner than usual. Some pubic and axillary hair grows in 10- to 12-year-old girls without puberty because their adrenal glands are producing normal amounts of androgens.

Treatment with female hormones

Fortunately, the missing female hormones can be replaced with medication. Because this treatment will cause bone age to advance more quickly, a girl may choose to delay puberty until she is close to the end of her growth potential. Most girls choose to begin estrogen treatment between 12 and 15 years of age, beginning with a low dose that starts the process of breast development, growth of the uterus (womb) and change in body contours. The dose of estrogen is increased over one to three years, and then progesterone is added to begin menstruation.

Estrogens are essential for a healthy body and should be continued at least until the usual time of menopause (around age 50 years). Estrogens help decrease a woman's risk of cardiovascular (heart) disease and stroke and are needed for bone health and strength. Women with TS who do not take replacement estrogen therapy through mid-life are at high risk for osteoporosis (weakened bones). The possible benefits and risks of continued estrogen therapy after 50 are unclear; each woman should discuss this with her doctor.

Sexual function is normal in women with TS who are receiving hormone treatment, and they should expect to have a healthy and satisfying sexual life. Teens and women with TS have the same risk for sexually transmitted diseases as anyone else and should learn how to protect themselves from this risk when they become sexually active.

As with any women on hormone replacement therapy, women with TS should have yearly gynecologic check-ups and promptly report any unusual symptoms to their gynecologist.

Fertility

Rarely, a woman with TS may conceive a baby without special medical treatment. Many of the same options for having children exist for women with TS as for other women with a fertility problem. Adoption is one common solution. Techniques that allow a woman to carry a pregnancy include oocyte donation (putting another woman's egg in the fallopian tube and trying to



fertilize it, called gamete intra-fallopian transfer or GIFT) and embryo donation (inserting a fertilized egg into the uterus, called in vitro fertilization or IVF). These techniques, combined with careful hormone therapy, have enabled many women with TS to carry a child through pregnancy and give birth.

Researchers now are focusing on the possibility of using preserved eggs or ovaries obtained in childhood to allow a woman with TS the chance of pregnancy with her own eggs later in life. Pregnancy in TS is always a high risk situation for mother and baby, though, and the pregnant woman should be followed very carefully by a team of well-trained specialists.

Intelligence and school performance



The intelligence of girls with TS varies just like that of the general population and usually is in the normal range. There are women with TS who are doctors, nurses, secretaries, day care workers, artists, teachers, lawyers, athletes – or a part of almost any profession that one can name. However, researchers have learned that girls with TS are more likely than other children to have specific learning disabilities, especially in nonverbal areas (sometimes called nonverbal learning disabilities or NLD). Commonly observed weaknesses include:

- difficulty imagining objects in relation to each other (visual-spatial processing; difficulty driving and poor sense of direction)
- trouble appreciating subtle social cues such as facial expressions (social cognition)
- problems with nonverbal problem-solving (math)
- clumsiness (psychomotor problems and poor manual dexterity).

These learning disabilities are common enough that some specialists suggest that girls with TS be screened for them as early as infancy; others suggest waiting to see if problems arise and testing them promptly at that time. Parents and teachers should have the same expectations of the girl with TS as of other children, while staying alert for signs of a learning problem.

Psychologists specialize just as physicians do, so it is important to find a psychologist who is trained to identify and meet your child's special needs. Pediatric psychologists focus on children who have medical conditions and their families, and neuropsychologists work with people who have conditions that affect their ability to learn. A psychologist with training in one of these specialties would be well-suited to become part of your child's support team. Ask your child's pediatric endocrinologist for suggestions or check the TSS-US website (see *Resources*) for more information about how, when, where and with whom to have your child screened for learning disabilities.

If a learning disability exists, appropriate learning activities and teaching strategies can be planned and put into action before the toddler falls behind in her motor development or the girl has serious problems at school. Occupational therapy (even for toddlers), academic tutoring and training in problem-solving also can help girls with TS cope with their visual-spatial and learning challenges, which, if present, will persist into adulthood. More information about these challenges and ideas for working with them can be found through TSS-US (see *Resources*).



Good career and vocational planning are important to young women with TS, especially if learning disabilities exist. Fortunately, there are an increasing number of successful women with TS who are involved in support groups such as TSS-US and can serve as role models for younger girls.

Social and emotional development

The results of early research on adults with TS show that they are less likely than other women to live independently, marry and be sexually active, in spite of similar education and employment backgrounds. This research involved women who, as a group, did not benefit from the types of hormone treatment and other support available today, so the current generation of girls who are receiving better care may be different. However, these findings point to the need for attention to social and emotional development in childhood and adolescence.



The results of recent research on the social and emotional adjustment of young girls with TS show that, compared with other girls, they tend to have more immature behavior, difficulty concentrating and problems with overactivity - all of which can affect their relationships with other children. Teenagers are prone to immaturity, anxiety, depression and social withdrawal and tend to begin dating and sexual activity at a later age than other girls.

Given these risks, it is important that families and professionals encourage and support these girls in establishing healthy friendships with other children the same age and participating in age-appropriate social activities (i.e., clubs, hobbies, Scouts, church activities, volunteer work, sports, etc.). Support of healthy, age-appropriate sexuality (in the context of personal family values) is important to create the foundation for dating and sexual relationships, including marriage.

Fostering independence throughout childhood and paying attention to career and vocational planning during the teen years will help create more choices for girls with TS and improve their quality of life in adulthood. Contact with other families through a support group such as TSS-US is very helpful in dealing with these issues, and the section on *Social and Emotional Support* contains ideas for supporting healthy inner growth and the development of social skills during childhood.

OTHER HEALTH ISSUES

There are a number of health concerns and medical problems that occur more frequently in girls with TS than in other children, so it is crucial that these girls receive good health care from a doctor who is familiar with the condition, such as a pediatric endocrinologist. TS clinics exist in some places and have teams of specialists who work to meet the needs of girls with TS and their families.

There is a range of severity in many of these conditions, with some girls being seriously affected, some mildly affected and others not affected at all. Many of the conditions can be corrected or controlled with treatment, and some of them can be prevented. This lengthy list is provided for completeness. No one girl will have all of these conditions! Your daughter's doctor or nurse is in the best position to answer specific questions about her care.

Heart and blood vessels



About 30% of girls with TS are born with a problem affecting the structure of the heart. The most common problems are a bicuspid aortic valve (the valve through which blood exits the heart into the aorta normally has three flaps, but in this case has only two) and coarctation (narrowing) of the aorta (the main artery leaving the heart). Although these conditions often are diagnosed at birth or in early childhood, all girls and women with TS, regardless of age, should have a thorough physical exam and an echocardiogram or MRI of the heart to look for heart conditions as soon as they are diagnosed with TS, during early adolescence (12-15 years old), and every three to five years during adulthood. If any problems are found, the child should be followed by a pediatric cardiologist (children's heart specialist) for ongoing care. In some cases, the problem may need to be corrected with surgery, and in others, the cardiologist may just follow the child carefully.

High blood pressure (hypertension) is another common condition in TS. Sometimes it results from a heart or kidney problem, but sometimes there is no known cause. High blood pressure may not occur until later in childhood or adulthood and may get worse with obesity and age. Women with TS should have their blood pressure checked regularly and follow their doctor's advice, which may include diet, exercise and medication.

Another heart problem, aortic root dilatation (enlargement of the aorta where it leaves the heart, often associated with weakness of its walls), is uncommon, but can have a devastating result – the layers of the weak walls of this major artery can separate from each other (dissection) or burst (rupture). This can cause internal bleeding, shock and even death if not diagnosed and treated quickly. The risk is greatest during adolescence and adulthood, and this can occur even in women who do not have any history of heart

problems or high blood pressure (although those factors increase the risk). All older girls and women with TS – and their families – must understand that severe chest pain, even if it doesn't last long, can be a symptom of this extremely serious problem. They should go to an Emergency Department immediately and have an MRI done to rule out aortic dissection. This condition is rare, and there is no need to live in fear of its occurrence – just a need to be aware of it so it can be diagnosed accurately and treated quickly if it occurs.

Kidneys

Between 25 and 40% of individuals with TS are born with a condition affecting their kidneys (the organs responsible for filtering blood and forming urine). In some, the kidneys are an unusual shape (horseshoe) or are in an unusual position, and in others, the internal structure of the kidneys is affected (double urine collecting system is most common). These conditions may not have any effect on the girl's health, but they may increase her risk of urinary tract infections, high blood pressure or other kidney problems. For this reason, all girls with TS should have an ultrasound exam of their kidneys soon after they are diagnosed. If any unusual findings are present, they should be assessed and treated (if needed) by a pediatric nephrologist (children's kidney specialist).

Ears and hearing



Ear infections and hearing loss are common in TS: in one study of girls with TS aged 4 to 15 years, 57% had some eardrum problem and 43% had hearing loss. Changes in the structure of the middle ear and eustachian tube (tube connecting the middle ear with the back of the throat) probably account for poor drainage and ventilation in the middle ear and easier movement of germs from the throat into the ear - all of which play a role in causing frequent middle ear infections (otitis media) and effusions (fluid in the middle ear), especially in early childhood. Middle ear effusions cause conductive hearing loss because sound does not move normally through an ear filled with fluid. Middle ear infections should be treated promptly with antibiotics. Placement of ear (tympanostomy) tubes should be considered if fluid in the middle ear has caused hearing loss for more than 3 months. Ear tubes drain fluid from the middle ear and restore hearing, decrease the number of ear infections and help prevent complications such

as growth of tissue in the middle ear (cholesteatoma) and infection of the mastoid bone (mastoiditis). Swollen adenoids can block the eustachian tube, cause fluid buildup in the middle ear and cause trouble breathing, so they may need to be removed (adenoidectomy).

Another type of hearing loss – sensorineural hearing loss – also is common in TS. This type of hearing loss is related to the function of the inner ear, where the vibrations caused by sound waves are transformed to nerve impulses that travel to the brain. In the study mentioned above, 58% of the girls also had some sensorineural loss. This hearing loss may begin in childhood and appears to worsen over time, so that more than 90% of women with TS in their forties have some degree of hearing loss, with more than 25% of them needing hearing aids.

Because of the high rate of ear and hearing problems and the effect they can have on a person's quality of life (including their speech), everyone with TS should have their hearing and middle ear function checked regularly throughout life. It also is important that girls and women with TS protect their hearing by avoiding loud noises and using ear protection in loud environments.

Eyes and vision



Several eye conditions are more common in TS than in the general population. These include strabismus (wandering or crossed eye), amblyopia (“lazy eye” with decreased vision), ptosis (droopy upper eyelid), red/green color blindness and congenital glaucoma (increased pressure inside the eye starting at birth). Of these, strabismus is most common, affecting about one-third of girls with TS. It usually becomes evident between 6 months and 7 years of age. If not treated promptly with special glasses or surgery, decreased vision will occur in the wandering eye(s) in 30-50% of affected girls. Girls with TS should have their eyes and vision checked by their primary doctor as part of each physical exam and by an ophthalmologist (eye specialist) at age 2 and then as needed.

Bones and teeth

Girls with TS may appear stocky because they have relatively broad shoulders and pelvis; they also tend to have large hands and feet. Infants with TS have an increased risk of congenital hip dislocation (which increases the risk of osteoarthritis in the hips of



older women). About 10% of girls with TS develop scoliosis (curvature of the spine) during early childhood or adolescence. Scoliosis also may become apparent or worsen during growth spurts related to GH treatment. The pediatric endocrinologist will check for these conditions at regular clinic visits and refer the girl to a specialist if needed.

Osteoporosis (“thinning” of bones) and fractures are more common in women with TS than in other women, but this may improve among girls with TS who receive growth hormone during childhood, start estrogens during early adolescence and take estrogens regularly during adulthood.

Girls with TS may have crowded teeth because of their small and receding lower jaw and narrow, high-arched palate. Their teeth may have shallow roots, placing them at risk for root resorption (“dissolving” roots). Good dental care and early referral to an orthodontist, at age 8 to 10 years, will allow coordination of orthodontic procedures with hormone treatments.

Immune system

Between 10 and 30% of girls and women with TS develop hypothyroidism (low thyroid hormone), which usually is caused by autoimmune disease (the body reacts to its own thyroid cells as if they were foreign and tries to destroy them; sometimes called Hashimoto’s thyroiditis). The risk of hypothyroidism increases with age. The symptoms of hypothyroidism (which may include constipation, low energy, dry skin and weight gain) often are subtle, so thyroid function should be checked at diagnosis and every year or two after that throughout life. Hypothyroidism is treated easily with medication.

Girls with TS seem to have a higher than normal risk of other immune system problems, including celiac disease (gluten sensitivity), inflammatory bowel disease and juvenile rheumatoid arthritis. Girls with digestive complaints or joint problems should be seen promptly by their doctor, who may refer them to a specialist if needed. Treatment may involve diet changes (for the bowel conditions) and medication.

Elevated liver enzyme concentrations are noted in up to 30% of women with TS; this usually is not related to any problems with liver function. In some women, it appears to be autoimmune, and in others, it is related to increased weight.

Metabolism

Girls and women with TS are at increased risk for a cluster of conditions sometimes called the “metabolic syndrome.” These include: high blood pressure; abnormal blood levels of lipids (such as cholesterol); non-insulin-dependent diabetes mellitus (NIDDM; also called Type II or late-onset diabetes); obesity; increased insulin secretion; and increased uric acid secretion (related to gout). Many of these conditions can lead to cardiovascular (heart and blood vessel) disease, so careful monitoring and prompt treatment of these problems (if they occur) throughout life is needed.

Obesity is a common problem in TS, especially during adolescence and adulthood. Extra weight on a short person is very noticeable and can lead to teasing by others and a poor self-image for the girl. Obesity is associated with high blood pressure and NIDDM, and since a girl with TS is at increased risk for these problems to begin with, it is important for her to maintain a healthy weight.



Diet and exercise are the keys to weight control in TS, as they are for everyone. Your child’s doctor or nurse can suggest a healthy diet and exercise program or make a referral to a dietitian for counseling. If you have a young daughter, help her develop good eating and exercise habits early in childhood – it’s much easier to stay lean than to lose weight.

Skin

Girls and women with TS are more likely than others to form exaggerated scars (hypertrophic scars or keloids) – even simple wounds like ear piercing and mole removal can heal with thick scar tissue. The surgeon and family should consider this before any surgery, including plastic surgery to improve the appearance of a webbed neck or prominent ears.

Benign moles (nevi) are common in TS and may increase in size and number throughout childhood, especially during GH treatment and adolescence. Although this does not appear to increase the risk of skin cancers, all moles should be watched for changes and evaluated by a dermatologist if they look suspicious. All children should use sunscreen to lessen their risk of skin cancers.



Hemangiomas (benign tumors made up of newly formed blood vessels) are more common in girls with TS than in other children. These may appear on the skin as birthmarks (port wine stains, “strawberry” or “raspberry” marks) or in internal organs such as the intestines, where they may burst and cause bleeding. A girl with TS who is anemic or having stools that contain fresh blood or look like coffee grounds should have a test to look for an intestinal hemangioma.

Seborrhea and eczema (atopic dermatitis) also are common and usually respond well to treatment.

SOCIAL AND EMOTIONAL SUPPORT

General issues

Growing up can be difficult for any child, and the child who is different by virtue of height or appearance may have more challenges than other children. Girls with TS may feel self-conscious and embarrassed about their condition, which may result in low self-esteem. The attitudes of parents, siblings and relatives are important in helping a girl with TS develop a strong sense of identity and self-worth. Parents may feel angry, guilty or disappointed when they learn that their child has a genetic condition. They may subtly pull back from her or overprotect and shelter her from the outside world. These are normal human reactions and are part of the expected process of adjustment that can lead to acceptance of the child and her condition.

Contact with other parents who have dealt with similar issues can be very helpful during the difficult time after diagnosis. The most important things parents can do for their children (and themselves!) are to accept and love them just as they are, recognize their strengths and potential and support them in developing independence and a strong sense of self.

Health care issues



The child's physician or nurse should provide the family with information about all aspects of TS either directly or by helping families locate good resources. The learning process will occur over months and years as different concerns and needs arise during infancy, childhood, adolescence and adulthood. Health care providers and parents should be open and honest with the girl and should include her in discussions about the implications and treatment of her condition. This is especially important as the girl becomes older and the time approaches to make decisions affecting her sexual development and growth.

Parents and older girls and women with TS may have to deal with health care providers who have limited knowledge about TS and the health risks related to it, sometimes even in emergency situations. It takes courage and assertiveness to demand that a health care provider consult with a physician who knows more about TS or listen to a knowledgeable parent or individual with TS. There may be times when such assertiveness may be necessary.

A tragic example of this is the recent death of a woman with TS in her mid-20s from an aortic dissection. She went to the Emergency Department when she started having severe chest pain, but no one knew that she was at increased risk of aortic dissection. When her pain eased, they sent her home without having done the appropriate test, an MRI of her aorta. She died soon after she left. Her death might have been avoided if she had known of this risk herself, told the doctors and demanded an MRI. Tragedies like this one will be avoided in the future as families and health care providers become more knowledgeable about TS.

Growth-related issues

Although GH treatment is making a difference in the growth of many girls with TS, short stature remains a concern in a society where great value is placed on height and even mild short stature is seen as a disadvantage. Family, friends and teachers may treat the short child according to size rather than age, and this may encourage immature behavior. Discussing this tendency with friends and teachers to alert them to it may be helpful.

Here are some hints for making life easier for a short child and supporting their independence:

- ❑ Make the physical environment at home as comfortable as possible for the child. Lower mirrors and closet rods and have steady footstools throughout the house.
- ❑ Teach your child ways of coping with the physical environment away from home. Act out situations that make your daughter feel uncomfortable in public and help her try out ways of dealing with them in the safety of your home first.
- ❑ Help your child learn social skills. Simple things like ordering food in a restaurant or asking for help in a department store may be intimidating if she is not quite sure how to do them. She can practice these skills at home and observe other children and adults in the same situations.
- ❑ Give your child responsibilities at home. Girls with TS should not be excused from sharing in household chores and jobs because of their size or condition. A footstool or arm extender will put most jobs within reach.
- ❑ Your child should dress according to age rather than size. This may mean having clothes altered or learning to sew. Styles that emphasize the bust and de-emphasize the waist and neck (such as the layered look and princess style dresses) may be most flattering for the older girl.
- ❑ Encourage your child to develop skills that allow for social interaction and competition with a variety of other children. There are many activities that help children develop special skills and learn to work and play with others – music, drama, dance, singing, 4H clubs, scouting and sports are just a few.



- ❑ Issues may arise for some girls around GH injections. Hints about dealing with stresses related to GH treatment are available in the materials provided by most of the companies that make GH and in a booklet distributed by the Human Growth Foundation (*Short & OK*; see *Resources*, Human Growth Foundation).

Social issues

The nonverbal learning difficulties that affect many girls with TS may affect their social development and relationships with others. Some of them have trouble “reading” and interpreting facial expressions and other nonverbal social cues, such as body language, touch and tone of voice. They may be easily overwhelmed by new situations and have trouble identifying their own emotions, as well as others’. They may not realize the effects their behavior has on other people, and sometimes they can be offensive without meaning to. Parents and teachers can help these girls to polish their social skills in several ways:



- ❑ Help your child to name and identify her own feelings – “You slammed the door and threw your backpack on the floor. What are you feeling inside? Are you angry right now?”
- ❑ Identify your own feelings as they arise in interaction with your child and help her learn the meaning of facial expressions and tone of voice. “I’m feeling happy right now – see, I’m smiling, my voice is high and sing-song and my body is relaxed.”
- ❑ Encourage social interaction with other children her age from early in life. Provide guidance about the “rules” of play – sharing toys, taking turns, etc.
- ❑ Talk with your child about social space (how far apart we stand from each other when we talk), eye contact (what it means to look someone in the eye) and the impact that smiling and complimenting people have on relationships.
- ❑ Help your child understand the effect her behavior has on others – “When you got up and walked away without saying anything, Brittany thought you were mad at her. Next time, tell her why you’re leaving. Then she’ll understand and her feelings won’t be hurt.”

- ❑ Encourage your child to practice or role-play specific social exchanges at home – what to do and say when you meet a new child or adult, for example, or how to order at a restaurant. Help her practice adapting these scripts to new situations.
- ❑ Teach your child ways of managing tension and stress – closing her eyes, relaxing her body and taking a few deep breaths, for example. There are many stress management techniques that work just as well for children as for adults – check your local library or ask your child’s health care provider for suggestions.

Sexuality and fertility issues



Many (perhaps most!) parents feel at least a little uncomfortable talking about sexuality and reproduction with their children. However, talking about all aspects of sexuality – differences between boys and girls, differences among girls, normal changes in puberty, how babies are made, how adults show their love for each other in respectful ways – is even more important when normal sexual development is affected by a medical condition. You’ll find many helpful books on talking with your children about sexuality and reproduction at your local bookstore or library; a few good ones are listed in the *Resources* section.

Doll-play is an important part of life for many young girls and gives parents a chance to introduce the idea that there is more than one way to become a mother while their daughter is very young. As she gets older, the girl with TS may have concerns about her sexuality and her relationships with boys. Reassure her that she is a woman in every way and can expect to have healthy, normal relationships with the opposite sex. She will experience the same feelings and confusion every teenage girl experiences as part of growing up. She will want to date boys, but may feel nervous because she sees herself as “different” from other girls. She will have to make decisions about how, when and with whom she wants to talk about her condition. Accepting infertility and the challenges she will face if she wants to have children may be difficult for a young woman with TS – and perhaps for her partner.

Talking with a parent, health care provider or trusted adult can help the teenager or young woman to feel more comfortable with herself and to make the decisions about these things that are right for her. Professional counseling or contact with other girls and women with TS often is very helpful. TS support groups exist in many areas; you can find out if one is close to you by contacting TSS-US (see *Resources*) or asking your child's doctor or nurse.

Educational and career issues

Parents and girls and women with TS should be well-informed about the learning problems associated with TS, as many individuals are affected by them, if only to a mild degree. These problems may interfere with school performance and limit career choices if not recognized and treated. They do not disappear with age, so early evaluation and promotion of coping skills are very important. Career counseling and preparation for leaving home (driving, managing finances, time management, etc.) should begin well ahead of time.

Here are some tips that may help your daughter deal with some of the challenges related to her learning style (they apply to many other children, too!):

- ❑ Encourage personal responsibility for her locker or cubby, her room, her notebooks, etc. Help her develop specific, explicit rituals for organizing things (notes from the teacher always go in this notebook pocket; lunch always goes on the top shelf of your locker, your jewelry goes on this shelf, etc). Reminder signs and notes may help.
- ❑ She may have trouble dealing with unexpected changes in routines. Give her early warning and explicit descriptions of what will happen, review step-by-step exactly what she'll need to do and arrange for a buddy, if possible.
- ❑ Help her with weekly clean-out and organization of her desk, locker, backpack, room (less over time, as she learns how to do it herself).



- ❑ Teach her to use a schedule book for recording assignments and color-coded folders for keeping her papers and homework organized.
- ❑ Give clear instructions for working on projects and homework: setting priorities, breaking a task into steps, developing a schedule and plan, tying up loose ends.
- ❑ Hand-written note-taking may be a problem. If so, it may be solved by using a tape recorder, getting a teacher outline, copying notes from classmates or using a word processor in class.
- ❑ Teach her structures and templates for various tasks – book reports, spelling assignments, etc.
- ❑ Coach your child to notice similarities and differences among tasks, find clues in a new task that relate to a previous task and adapt strategies for dealing with variations in tasks (generalizing, then adapting). “How is this assignment like the last one? Would it work to use a similar outline, or do you need to write a new one?”
- ❑ Help her to identify relationships among parts and the whole and among categories, sub-categories and the links between them.

SUMMARY

The outlook for girls and women with TS has improved significantly over the past two decades. Research into the genetic basis of the condition and its physical, psychological, emotional, social, and intellectual aspects has resulted in greater understanding of the condition and of ways in which the healthy growth and development of girls and women with TS can best be supported. With good medical and psychological care and the emotional support of friends and family, a girl with TS should look forward to a healthy and satisfying life!

RESOURCES

Support Groups

Information and support from others who have dealt with similar issues are invaluable for girls and women with TS and their families. The Turner Syndrome Society of the United States (TSS-US) is an organization whose membership has grown quickly into the thousands. TSS-US is a non-profit public service organization whose membership has quickly grown into the thousands. Its two-fold mission is:

- ❑ to enable innovations in health for Turner syndrome women by
 - working with health-care professionals to expand knowledge about the condition, its diagnosis, treatment and prevention through research
 - promoting the successful rearing, affirmation and support of individuals affected by the condition
- ❑ to enable innovations in learning for Turner syndrome women by
 - providing a public forum for communication of state-of-the-art information, exchange of ideas and social support
 - increasing public awareness of Turner syndrome, its effects and its possibilities.

More than 50 chapters and support groups are active across the country. In addition to general information about TS and TSS-US, the website contains links to other organizations and companies; an “ask the expert” section; chat rooms and bulletin boards for parent, teen, and adult members; an online store for ordering books, videos and other material for parents and professionals (including a CD-ROM for health care providers); research study opportunities and articles; information about the new TS Registry; and FAQs.

Website: www.turnersyndrome.org

Address: 1445 TC Jester, Suite 260, Houston, TX 77014

Phone: 1-800-365-9944

The Human Growth Foundation (HGF) helps parents of children and adults who have disorders of growth or growth hormone through education, research, support and advocacy. HGF has chapters across the country and distributes booklets and other resources about growth-related conditions, including TS. *Ready for School* and *Short & OK: A Guide for Parents of Short Children* are two that may be of special interest.

Website: www.hgfound.org.

Address: 997 Glen Cove Ave, Glen Head, NY 11545.

Phone: 1-800-451-6434.



The Major Aspects of Growth in Children Foundation (MAGIC) provides support and education regarding growth disorders in children and related adult disorders and has support groups all over the United States. MAGIC has a wide array of pamphlets on growth-related conditions (including TS) and other resources for families.

Website: www.magicfoundation.org.

Address: 1327 North Harlem Ave, Oak Park, IL 60302.

Phone: 1-800-MAGIC3.

Books and Websites

There are many wonderful books available in libraries, bookstores and online about helping children to develop a strong sense of themselves. These are especially useful for children who are “different” in some way. One classic is *Your Child’s Self-Esteem* by Dorothy Briggs; another is *Self-Esteem: A Family Affair* by Jean Illsley Clarke. Shel Silverstein’s *The Missing Piece* is a story for young children (of all ages) about a wheel that is missing a piece and how it learns to feel good about itself even though (or because) it is different from other wheels. *People* by Peter Spier is a book to help 4-8 year-olds understand and accept the differences and similarities among people.

Useful books about supporting the development of good social skills include: *Helping the Child Who Doesn’t Fit In* by Stephen Nowicki and Marshall Duke (1992) and *Teaching Your Child the Language of Social Success* by Marshall Duke, Elisabeth Martin and Stephen Nowicki (1996).

Two books by Kate Phifer are harder to find, but worth it for their common-sense, down-to-earth suggestions for dealing with the challenges of short stature: *Growing Up Small: A Handbook for Short People* (Eriksson Publisher, 1979) and *Tall and Small: A Book about Height* (Walker and Company, 1987). Your child’s doctor or nurse and other families who have a girl with TS probably will have other books and videos to suggest, so be sure to ask them what they’ve found useful.

You will find many helpful books about sexuality and reproduction at your local library or bookstore, but here are a few

worth mentioning specifically: *It's Perfectly Normal* (for children 10 years and up; 1994) and *It's So Amazing* (7 years and up; 2002) by Robie Harris; *What's Happening to Me?* (10-14 year olds; 1975) and *Where Did I Come From?* (6 years and up; 1973) by Peter Mayle; *The What's Happening to My Body?* and *My Body, Myself* books for teenage boys and girls (1987, 1983) by Lynda Madaras; and *The Underground Guide to Teen Sexuality* (older teens; 1997) by Michael Basso. Good sources of general health information for teenage girls are: www.youngwomenshealth.org and www.youngwomenshealth.com

Some valuable resources for more information about nonverbal learning disorders include: www.nldontheweb.org; www.nldline.com; *Nonverbal Learning Disabilities at Home* by Pamela B. Tanguay (2001) and *The Source for Nonverbal Learning Disorders* by Sue Thompson (1997). Two books by Byron Rourke (written and edited, respectively) are aimed at professionals, but may be useful for parents, too: *Nonverbal Learning Disabilities, the Syndrome and the Model* (1989) and *Syndrome of Nonverbal Learning Disabilities* (1995; contains a chapter on TS).

Websites that some parents may find useful for getting information about special education issues, testing for learning disabilities and parent rights are: www.exceptionalparent.com and www.Wrightslaw.com.

All the books mentioned in this section are available through Amazon.com; some are available through the TSS-US website.

GLOSSARY

Androgens – hormones that stimulate the development of male sexual characteristics; produced by the testes and adrenal glands (above the kidneys) in men; small amounts are produced by the ovaries and adrenal glands in women.

Autosomes – chromosomes other than the X or Y sex chromosomes; human cells contain 22 pairs of autosomes and one pair of sex chromosomes

Cell – basic unit of organization of living things; contains genetic material called chromosomes.

Chromosomes – rod-shaped structures in the nucleus (control center) of a cell; contain units of DNA called genes.

Estrogens – hormones that stimulate the development of female sexual characteristics and control the reproductive cycle in women; produced by the ovaries in women; small amounts are produced in fat tissue in men.

Genes – units of heredity containing DNA; determine traits passed from parents to child; also control reproduction and function of cells.

Gonads – sex organs; ovaries in females, testes in males.

Growth hormone – a hormone produced by the pituitary gland (located near the base of the brain); plays a key role in stimulating increased height.

Hormones – chemicals secreted into the bloodstream in small amounts by glands throughout the body; hormones “set in motion” many processes crucial to life – growth, puberty, reproduction, metabolism, self-preservation.

Karyotype – laboratory analysis of chromosomes.

Menstruation, menstrual cycle – female reproductive cycle starting at puberty; involves build-up of the lining of the uterus (womb) for conception, followed by shedding of this lining (menstrual period) if conception does not occur.

Mosaicism – in genetics, the presence of cells containing different sets of chromosomes (two or more cell lines, or populations) in one person; usually, all of a person’s cells contain the same set of chromosomes.

Ovaries – female reproductive organs located in the lower abdomen (pelvis) on either side of the uterus (womb); contain eggs and produce hormones that control sexual development and reproduction

Pediatric endocrinologist – specialist in children’s growth and hormone conditions.

Puberty – the stage of life during which the reproductive organs start to function and adult sexual characteristics begin to develop.

Reproductive cells – sperm in males and eggs (ova) in females.

Syndrome – a group of signs or symptoms that, when they occur together, are thought to stem from the same cause and suggest the presence of an underlying condition .

Sex chromosomes – chromosomes (X and Y) that determine the sex of a person.

APPENDIX

Here is a health care checklist for girls and women with TS. Everyone who is diagnosed with TS should be referred promptly to an endocrinologist (or other specialist in TS) and receive the listed evaluations. It is crucial to assess each of these systems to look for important conditions that could affect the person's health and to establish baseline assessments for future evaluations.

Your child's pediatric endocrinologist is in the best position to answer questions about your child, suggest appropriate tests and refer your child to other specialists for additional services and support.

At the time of diagnosis (any age)

Complete physical exam (including height, weight, pulse, blood pressure and assessment of lymphedema)

Screen for learning disabilities; assess social skills and behavior, development and school/work performance and family function

Contact with a support group (such as TSS-US)

Blood work to check thyroid function (TSH, T4)

Hearing evaluation (by a specialist)

Renal (kidney) ultrasound and urine culture

Complete cardiac (heart) evaluation by a cardiologist, including echocardiogram

At each visit, (at least yearly)

Complete physical exam, including height, weight, blood pressure, exam of heart and pulses and assessment of lymphedema

Careful screening for:

- Hip dislocation: infancy
- Ear infections (otitis media): infancy through childhood
- Strabismus: 4 months to 5 years
- Scoliosis, kyphosis: 4 years until growth is complete
- Thyroid function: 4 years onward, including thyroid function tests
- Skin moles: late childhood onward
- Pubertal delay: 10 years onward

Counseling about college/vocational/work plans and sexuality issues: early adolescence onwards

Yearly

Thyroid function: 4 years onward

Additional blood tests including liver and kidney function, fasting lipids and blood glucose: beginning around age 18

Every 3-5 years

Hearing evaluation: Begin at diagnosis

Cardiac echocardiograms or MRI: Begin repeat evaluations around age 12

Bone densitometry: Begin around age 15

At specific ages during childhood

Eye exam by pediatric ophthalmologist: around age 2

Evaluation by orthodontist: around age 8

HEALTH CARE CHECKLIST

These tests may be performed earlier and more often as needed. They are in addition to routine health care maintenance (such as lead screening in childhood and Pap smears and mammograms in adults).

	at diagnosis	each visit, at least yearly	yearly	every 3-5 yrs	at specific ages
Complete physical exam (including height, weight, pulse, BP, assessment of lymphedema)	x	x			
Screen for learning disabilities; assess social skills, family function; contact with support group	x	x			
Careful screening for: Hip dislocation Strabismus Ear infection/fluid Scoliosis/kyphosis Skin moles Pubertal development	x If age appropriate	x In infancy 4 mos - 5 yrs Infancy/childhood 4 yrs until growth is complete Childhood onward 10 yrs onward			
Hearing evaluation (by specialist)	x			x	
Renal ultrasound and urine culture	x				
Eye exam (by ophthalmologist)					at age 2
Thyroid function tests	x		x (Begin repeat exams ~ age 4)		
Orthodontic evaluation					at age 8
Counseling re: sexuality issues and vocation/ college or work plans		x (Begin ~ age 10)			
Cardiac evaluation; echocardiogram or MRI	x			x (Begin repeat exams- age 12)	
Bone densitometry				x (Begin ~ age 15)	
Blood tests: liver and kidney function, fasting lipids and blood glucose			x (Begin ~ age 18)		



Turner Syndrome Registry

If you've finished reading this guide, you may be struck by how much we have learned about Turner syndrome (TS) - and how much we still have to learn. The Turner Syndrome Society of the United States (TSS-US) is starting an exciting new program - an international Turner Syndrome Registry (TS Registry) to collect, study, and transmit information about TS. Data gathered with the help of women/girls with TS and their families will help researchers, health care professionals, educators, and others better understand the condition. This, in turn, will help them develop and test new approaches to and treatments for many of the conditions that often are related to it. Some of this information will be beneficial to all women, not just those affected by TS!

If you are interested in taking part in this ground-breaking effort, go to the TS Registry website (www.tsregistry.org). The TSS-US website (www.turnersyndrome.org) also has a direct link to the TS Registry.

The TS Registry has online forms that will walk you through the registration process. You would be notified by e-mail or mail about opportunities to take part in various research projects such as online surveys and clinical trials.

You will always be in control of your participation in specific projects. Your privacy will be carefully protected and the information you provide will not be shared with anyone without your permission. Please visit the TS Registry website or e-mail the TS Registry Director (director@tsregistry.org) for more information.

The results of these efforts will improve our understanding of TS and the quality of life of those who are affected by it. Please help us better help you!





The Turner Syndrome Society
of the United States

[www. turnersyndrome.org](http://www.turnersyndrome.org)
1445 TC Jester, Suite 260, Houston TX 77014
1-800-365-9944