

Turner Syndrome

A Guide for Families



Smile, Laugh, Live





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TURNER SYNDROME: A GUIDE FOR FAMILIES

This guide is published by the **Turner Syndrome Society of the United States** (TSSUS) for the purpose of providing a basic guide for understanding Turner syndrome (TS). Information and support from others who have dealt with similar issues are invaluable for girls and women with TS and their families. TSSUS is the largest grassroots non-profit organization in the world dedicated to supporting all persons touched by TS. We provide many resources related to TS throughout our website at www.turnersyndrome.org.



Resources TSSUS offers include:

- Information on specific topics such as the teen guide to TS, Coley's Story for girls, and the Clinical Practice Guideline for Turner Syndrome from the National Institutes of Health study group
- Local TSSUS groups across the country
- An online community forum for peer discussion topics
- An annual conference providing concurrent sessions to over 500 attendees, including girls and women with TS, their families and significant others
- An online store for purchasing books, awareness items and other material for parents and professionals
- Research study opportunities related to TS
- Event opportunities

Please contact us at 1-800-365-9944 or tssus@turnersyndrome.org, we enjoy helping the TS community.

The authors would like to thank all the girls and families who have taught us so much about living with Turner syndrome. Also, we thank all those involved with supporting this guide, from the initial stages to the current revisions.



Introduction	2	How to Use This Guide
	2	The Faces of Turner Syndrome
The Basics	3	What is Turner Syndrome?
	3	What Causes TS?
	4	The Nuts and Bolts of “X”s and “Y”s
	5	How Does Turner Syndrome Happen?
	6	How is TS Diagnosed?
	6	Before Birth
	6	During Infancy
	6	During Childhood and Adolescence
	7	During Adulthood
Growth and Development	7	Growth in TS
	9	Growth Hormone Treatment
	10	Other Physical Features
	11	Lymphedema
	11	Sexual Development in TS
	12	Treatment with Female Hormones
	12	Fertility
	13	Intelligence, Learning and School Performance
	13	Social and Emotional Development
Other Health Considerations	14	Heart and Blood Vessels
	15	Kidneys
	15	Ears and Hearing
	16	Eyes and Vision
	16	Bones and Teeth
	17	Immune System
	17	Metabolism
	18	Skin and Nails
Social and Emotional Support	18	General Issues
	19	Health Care Issues
	19	Growth Related Issues
	19	Social Issues
	20	Sexuality and Fertility Issues
	21	Educational and Career Issues
Wrapping Up	22	Summary
	22	Glossary
	24	Health Care Checklist



Introduction

HOW TO USE THIS GUIDE

This booklet is intended for families who are touched by Turner **syndrome*** (TS). It contains a great deal of information as TS is a highly variable condition. The purpose of this booklet is to answer some of the questions you and your daughter may have regarding TS. Much of the information may be useful for women with TS, though it is beyond the scope of this booklet to address adult issues.

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 even though 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. **Your child's doctor is best qualified to answer specific questions about your daughter**, but the general information you will find here may help you better understand this condition and what it may mean for your daughter and family.

We suggest that you read this booklet, and then make a list of questions and concerns to discuss with your daughter'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.

** 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 draws a blood sample from her arm for a **karyotype**, the blood test used to diagnose Turner syndrome. Sarah's parents are worried, although the doctor assures them that Sarah is a strong, healthy baby.

Maria 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 Maria'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).





Kristin is 14 years old and is quite concerned because her breasts are the size of 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.

Emily 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 Emily's **ovaries** are becoming menopausal already. She is barely 5 feet tall and when the doctor examines her more carefully, she notices many moles and a high, arched palate (roof of the mouth). It occurs to her that Emily may belong to the group of women with Turner syndrome who experience a typical **puberty**, 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 in the same way – but each of them has Turner syndrome (TS).



The Basics

WHAT IS TURNER SYNDROME?

A syndrome is a set of features or symptoms that often occur together. 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 angled away from the body at the elbow), webbing of the neck and a low hairline at the base of the neck. 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 abnormal development (dysgenesis) of the ovaries (female organs that store eggs and produce sex hormones; also known as **gonads**), but this term is not specific to Turner syndrome as there are other causes for gonadal dysgenesis.

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 did not have all or part of one of their X **chromosomes**, and suggested that the missing genetic material accounted for the physical findings observed in this condition. The diagnosis of TS is usually 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, effecting 1 of every 2,000 females. Fortunately, most girls with TS can expect to lead healthy, productive and happy lives.

WHAT CAUSES TS?

The Nuts and Bolts of “X”s and “Y”s

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

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 average 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 46,XX in gene-talk); if one X and one Y are present, the baby will be male (46XY).

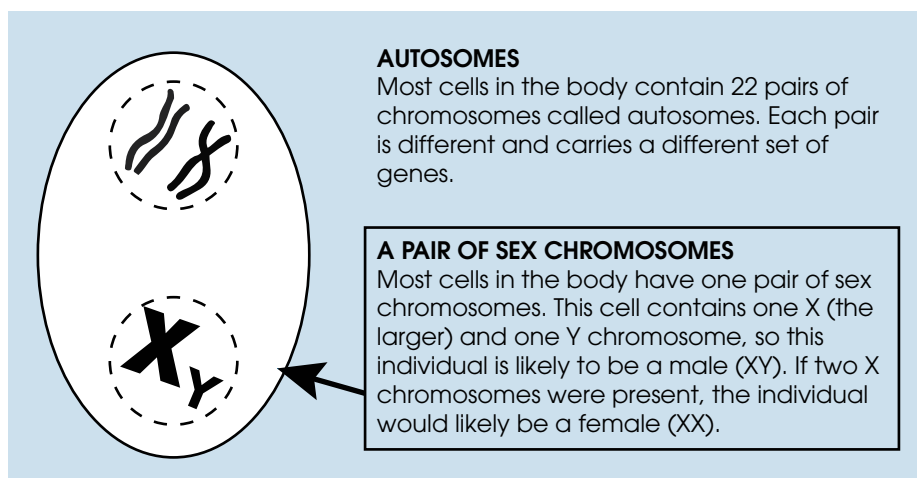


Figure 1.
Genes in the nucleus of a cell

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

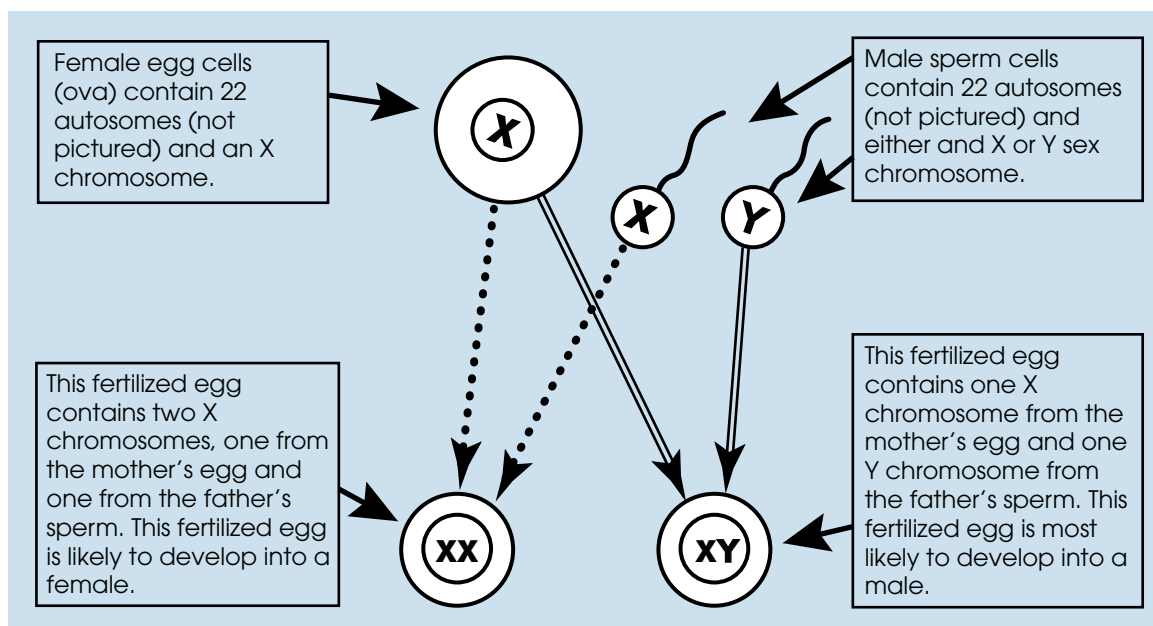


Figure 2.
Boy or Girl?



in sexual development. In 46,XX 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 natural growth and development of girls.

How does Turner syndrome happen?

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 either parent did before or at the start of the pregnancy that caused or increased the risk of TS for their daughter. The age of a parent, ethnicity, diet or other factors are unrelated to the conception of a child with Turner syndrome.

Turner syndrome occurs when there is one complete X chromosome and the second X chromosome is either missing entirely. Or, the X chromosome may be present in a form that is incomplete or different in structure. In addition, the X chromosome difference can be complete (present in every cell) or found only in a proportion of cells (called mosaicism). As you might guess, there are many variations of chromosome types ("karyotypes") which cause Turner syndrome.

Almost 50% of girls with TS are missing an X chromosome in every cell that is tested (which is usually blood). This is written 45,X.

About 30% of girls with TS have mosaicism, which means that some cells that have the complete number of chromosomes (written 46,XX) and the other cells are missing an X (45,X). We think this result is from the loss of an X chromosome in some cells after fertilization. . There are different types of mosaicism. Girls who have 45,X/46,XX mosaicism can be harder to diagnose as these girls usually (but not always) have fewer features related to TS because more body cells have the usual 46,XX pattern.

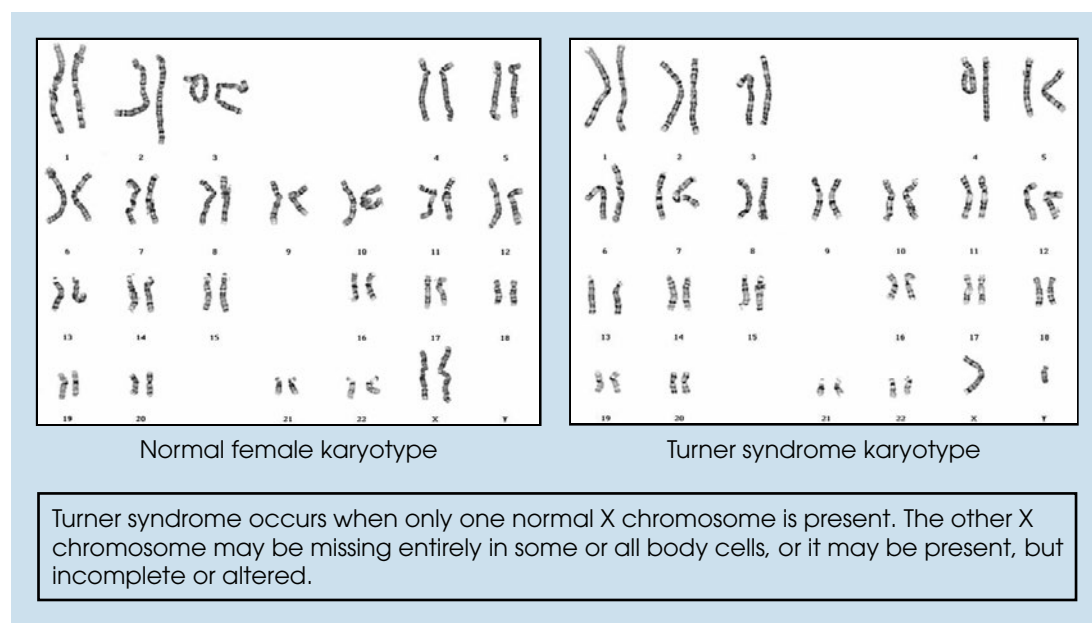


Figure 3.
Karyotypes

Another type of mosaicism is 45,X/46,XY and occurs in a small percentage of girls with TS. They have a little Y chromosome material which can increase the risk of developing tumors in their "gonads" (internal sex organ). Surgical removal of the gonads is generally recommended.



A rare type of mosaicism is 45,X/47,XXX in which there are 45,X cells and a second cell type which has an extra X chromosome. These girls are usually taller.

Some girls with TS have a combination of cell types, such as 45,X/46,XX/47XXX.

Finally, about 20% of girls with TS have two X chromosomes, but one of them is incomplete or different in structure. A “ring chromosome” means that the X chromosome is shaped like a circle, with the ends of the X chromosome joined. An “isochromosome” means that the X chromosome has two long arms instead of a long arm and a short arm. When the X chromosome is missing a little material we use the term “deletion”. These structural differences occur prior to conception during the formation of the egg and sperm (gametogenesis). A structurally abnormal X is often lost early in the development of the embryo and results in mosaicism. In other words, there is a structural abnormality of the X chromosome (ring, isochromosome, deletion) in some cells, but not all.

This review does not cover all possible forms and combinations. Talking to a geneticist, physician or genetic counselor can provide the details for you or your daughter.

HOW IS TS DIAGNOSED?

Before Birth

TS may be detected during pregnancy if a karyotype is performed on cells collected during amniocentesis or chorionic villus sampling (CVS); these tests often are ordered because the mother is older. Older parental age does not increase the risk of TS, but it is associated with other conditions so a prenatal test for another condition may detect TS.

TS also may be discovered when a prenatal ultrasound shows that the fetus has physical features common to TS, such as cystic hygroma (a collection of lymph fluid around the neck) or heart defects. A prenatal karyotype can confirm TS. Additionally, TS may be discovered when the mother undergoes a blood test for multiple markers of fetal abnormalities. Miscarriage may occur if the problems are severe.

When the diagnosis is made early in the pregnancy, parents may find it helpful to talk with families who have a daughter with TS, women who have TS, a pediatric endocrinologist or a medical geneticist/genetic counseling team. TSSUS can help parents find support and sound medical information.

During Infancy

Many girls with TS are diagnosed at birth or during infancy, usually because of the presence of typical physical features such as lymphedema (swelling due to excess lymphatic fluid, especially of the hands and feet) or a heart condition often 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 begin puberty, meaning, they do not develop breasts or their breast development begins, but they do not menstruate. An evaluation by a specialist may uncover



subtle signs of TS in these girls that were missed or not apparent to their general health care providers.

During Adulthood

In a small number of women with TS, puberty occurs naturally, but at some point, their ovaries may stop functioning because hormones are not produced and menstruation may cease. 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 _____

As for any child, issues related to growth and development is 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 (height), sexual development and the development of social skills.

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% fall below the 5th percentile around 2 years of age. Growth continues to be slower than average 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 as a supplement, 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 late teens. This delay in growth also applies to most girls with TS that have spontaneous (natural) puberty.

Growth charts: Growth charts are used to compare the height of one child with the heights of other children the same age and sex. Figure 4 shows the range of height for girls with Turner syndrome and the range of height for other girls.

The top pink line marked “95” is the 95th percentile line for typical growing girls. This means that a girl whose height is on that line is taller than 95% of girls her age. The 50th percentile line represents the average height for a given age. The lower pink line is the 5th percentile for typical girls.

Turner syndrome growth chart: The area between the top and bottom pink lines represent the range of height for most girls. The top and bottom blue lines represent the range of height for most untreated girls with Turner syndrome.

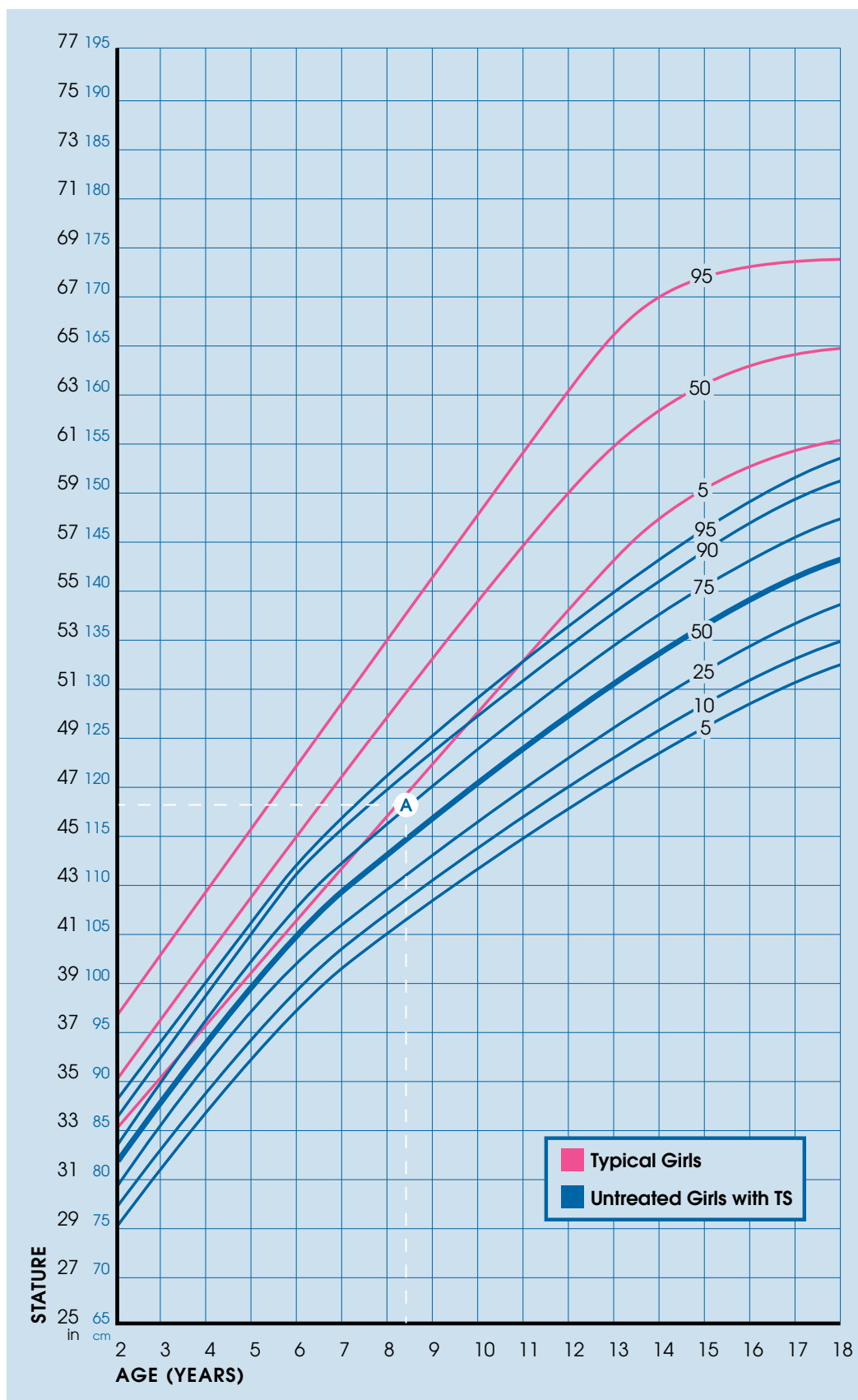


Figure 4.
Turner syndrome growth chart



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: Hannah 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 percentile on the girls' growth chart, which means that she is shorter than about 95% of typical girls her age. Her height is at the 75th percentile 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 a woman with TS that has not received **growth hormone** treatment is 4 feet, 8 inches (142 cm), although a few women reach 5 feet (152 cm). The heights of the parents may 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 (GH) 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 at regular intervals.** A standard growth chart is sufficient for tracking height in girls with TS, but a TS growth chart is advisable for charting the height of a girl when her height does not reach the minimum growth for the standard growth chart. Healthcare providers may also use condition specific growth charts as a means of comparison for a particular population.

Growth Hormone Treatment

Although girls with TS are not deficient in growth hormone, the US Food and Drug Administration joined other countries in approving the use of supplemental growth hormone (GH) for girls with TS in 1996 as studies showed final height was increased. 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 typical 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 growth and puberty in girls with TS, so treatment recommendations may change as more is learned.



Current TS guidelines suggest that GH treatment should be considered once growth failure is detected, which is a decrease in percentile on the standard growth chart, even in infancy. Although considered a standard part of care, every family must make their own decision whether their daughter uses a promoting therapy or not.

Research has shown that among women with TS, the quality of life is not related to either adult height or the height gained after receiving GH treatment. We know that short stature can be associated with some degree of psychosocial stress, even if it does not result in significant negative social and psychological consequences. Fortunately, studies of patients with TS and the general population show that youth with short stature function in their daily lives in ways comparable to their average-height or tall peers.



Androgens (male-type hormones) naturally 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. Androgens and GH together, promote natural 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 8 years, when androgen levels should be rising naturally. 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) in very low doses may enhance growth. Estrogen replacement therapy information is described within the sexual development section of this guide.

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 pubertal doses of estrogen therapy (A doctor can discuss balancing the importance or desire of some pubertal development using low dose estrogen)

All growth-related hormone treatments should be supervised by a pediatric endocrinologist, who will follow your daughter 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 a reversible condition of 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, delayed breast development and rapid advance of bone age.

Other Physical Features

Some unique physical features may be observed in girls with TS. Some of the features occur because of changes in the ordinary growth of bones before and after birth. Here is a list of some common features, including medical terms and descriptions:

- Wider upper body compared to narrow lower body
- 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 outward)
- Cubitus valgus (arms that turn out slightly at the elbow)
- Short 4th metacarpal (bone in hand under knuckle)
- Pectus excavatum (sunken appearance of the chest)

Lymphedema

Other unique physical features result from under development of the lymphatic system before birth. The lymphatic vessels carry 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 clear 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.

Additional unique features as a result of fetal lymphedema include:

- Thick, short neck, “webbed” or broadened appearance
- Low hairline at the back of the neck
- Ears protruding outward, and/or low set, and/or rotated toward neck
- Narrow fingernails and toenails that point upwards

Some features may be barely noticeable but may still make one feel self-conscious. Some of the symptoms of lymphedema 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 their neck and ears. As with any surgery, pros and cons should be discussed before a decision is made. Makeup, clothing and hairstyling are the most common ways in which those with TS choose to make themselves feel more confident.

SEXUAL DEVELOPMENT IN TS

Lack of sexual development (breast development, feminine body contours and menstruation) **during adolescence are hallmark features of TS.** Sexual development may be affected because the ovaries may not fully develop or may 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 dysgenetic gonads or ovarian streaks in TS, 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 45,X karyotype and about one-third of girls with mosaicism for 45,X/46,XX 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 average amounts of androgens.

Treatment with Female Hormones

If a girl does not begin puberty on her own, the missing female hormones can be replaced with medication. The decision of when to begin estrogens is one each girl and her family must make, trying to balance



continued growth [since estrogens hasten the closure of the bones and growth can stop earlier] with starting puberty. When to start estrogen and increase dosages are important decisions, and families should discuss the various aspects of it thoroughly. **Decisions will be different for each family.** Delaying the start of puberty may affect a girl's self-image, her friendships with peers, and her development. Most girls with TS 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.

Once estrogens reach high enough levels to bring about puberty (breast development), bone age advances more quickly and the clock ticks faster towards the time when growth stops. 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. The dose of estrogen is increased over one to three years, and then progesterone is added to begin menstruation. The standard level of estradiol (the major estrogen produced by the ovaries) can be achieved using transdermal skin patches.

Estrogens are essential for a healthy body and should be continued at least until the usual time of menopause in the general population (around 50 years old). Estrogen replacement therapy promotes cardiovascular health as well as bone health and strength and affects virtually every tissue in your body. 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.

While sexual function is often normal in women with TS, there can sometimes be problems with dyspareunia (painful intercourse) due to a small vagina or vaginal dryness, both of which can be treated. 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 all 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 exist for women with TS as for other women with fertility problems. Adoption is the most common solution at this time for women with TS. In-vitro fertilization and implantation with eggs from a donor are technically possible, but the availability has decreased among providers of Assisted Reproductive Technology because women with TS have an increased rate of maternal complications. First, because of their small size, many women with TS should deliver by cesarean section. Second, hypertension and diabetes are common. Most critically, the risk for dilatation and dissection of the aorta increases during pregnancy. Before contemplating spontaneous or assisted pregnancy, individuals with TS need a complete medical evaluation with particular attention to their cardiovascular system. For women with any significant abnormalities of the heart or great vessels, pregnancy is inadvisable because of the potential cardiovascular harm.

INTELLIGENCE, LEARNING AND SCHOOL PERFORMANCE

The intelligence of girls with TS varies just like that of the general population and usually is in the range of girls without TS. 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 be at risk for specific learning weaknesses, or disabilities, including problems in processing of nonverbal information** and in utilization of executive functions (i.e. management of cognitive and attentional skills). Commonly observed weaknesses include:

- Difficulty imagining objects in relation to each other (e.g., visual-spatial processing; difficulty driving and sense of direction)
- Problems with nonverbal problem-solving (particularly math)
- Clumsiness (coordination problems and poor hand/finger skills)
- Trouble appreciating subtle social cues such as facial expressions (social cognition)
- Problems with one or more aspects of managing processes of the brain (executive function) including working memory, sustained attention, problem solving, changing what you are thinking about and how you think about something (cognitive flexibility), task planning and execution

Although not every person with TS has challenges, these learning weaknesses or disabilities are common enough that some specialists suggest that girls with TS be screened for them as early as one or two years of age. **Parents and teachers should have the same expectations of a girl with TS as of other children, while staying alert for signs of a learning challenge.**

If a learning weakness or disability is identified, appropriate learning activities and teaching strategies can be planned and put into action before serious problems arise. For instance, explicit teaching of executive function skills is encouraged to increase overall cognitive abilities including those related to the visual-spatial domain. Occupational therapy (even for toddlers), academic tutoring and training in problem-solving also can help girls with TS strengthen their visual-spatial and learning weaknesses, which, if present, can persist into adulthood.

Persons with TS who have significant executive function-based attentional problems can sometimes benefit from medications used to treat Attention Deficit / Hyperactivity Disorder (ADHD). This determination should be made by a qualified mental health provider or doctor with expertise in ADHD.

Good career and vocational planning are important to young women with TS, especially if learning disabilities exist.

SOCIAL AND EMOTIONAL DEVELOPMENT

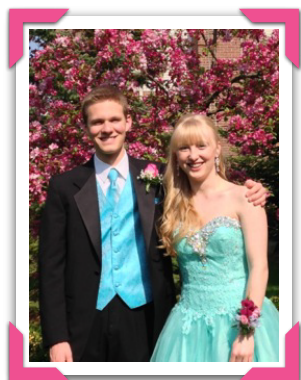
The results of research on the social and emotional adjustment of girls with TS show that, compared with other girls, they tend to have less age-appropriate social communication and interaction with others. It is believed that difficulties arise due to the lack of skills for processing social information, interpreting gestures, facial expressions, language tones/meanings, as well as having a poor sense of spatial awareness, difficulty concentrating and problems with over activity - all of which can affect their relationships with other children.

Given these observations, it is important that families and professionals encourage and support girls with TS in establishing healthy friendships with other children the same age and participating in age-appropriate social activities in supportive environments (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.



Therapy with a qualified mental health professional can also be of benefit for the child, teen or young adult with TS who is experiencing problems in social relationships and issues related to low self-esteem, particularly when accompanied by anxiety and depression. Such therapy often emphasizes a cognitive-behavioral approach and/or social skills training. Individuals with TS who have prominent symptoms of anxiety or depression that do not fully respond to behavioral therapies may benefit from adding treatment with medication. This determination should be made by a psychiatrist or other qualified mental health provider working closely with the person's primary care provider.

Other Health Considerations

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 Turner syndrome. TS clinics exist in some locations (refer to www.turnersyndrome.org) 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 50% of girls with TS are born with a problem affecting the structure of the heart. The most common problems identified by echocardiography (an ultrasound of the heart) include a bicuspid aortic valve (which refers to the valve through which blood exits the heart into the aorta having two leaflets, instead of the standard three) and coarctation (narrowing) of the aorta (main artery leaving the heart). These conditions may not be diagnosed at birth or in early childhood, therefore all girls and women with TS, regardless of age, should have a thorough physical exam and echocardiogram. If the diagnosis is made in infancy or early childhood, an MRI may be needed to define the structures outside of the heart (such as the aorta or pulmonary veins). A cardiac MRI or CT angiogram may be needed when a girl reaches an age when sedation is not necessary because certain cardiovascular structures have not been adequately visualized by echocardiography.

For young individuals without evidence of heart disease on their initial evaluation, a second evaluation should occur during early adolescence (12-15 years old). If problems are found, the child should be followed by a pediatric cardiologist (heart specialist for children) for ongoing care. In some cases, the problem may need to be corrected with an operation, and in others, the cardiologist may monitor only. In girls without evidence of cardiovascular disease an imaging study should be performed every 5-10 years. Often an echocardiogram is



adequate but in situations where the aorta (including the thoracic descending aorta) is not well delineated an MRI or CT scan may be necessary.

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. Both girls and women with TS should have their blood pressure checked regularly and follow their doctor's advice, which will include a healthy diet, exercise and possibly a medication.

Another heart problem, aortic dilation (enlargement of the aorta where it leaves the heart), commonly occurs. In a small number (~1%), there can be a devastating event when the layers of the weak walls of this major artery separate from each other (dissection) or burst (rupture). This can cause internal bleeding and even death if not identified and treated quickly. The risk is greatest during young adulthood. Nearly everybody who has an aortic dissection also has a heart malformation, high blood pressure or both. All girls and women with TS – and their families – must be aware that severe or persistent chest pain can be a symptom. **If there is a concern, an imaging study (echocardiogram, MRI, or CT scan) must be done immediately to rule out aortic dissection.**

KIDNEYS

Between 25 and 40% of individuals with TS are born with a condition affecting the structure of their kidneys (the organs responsible for filtering blood and forming urine). In some, the kidneys have 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 and do not cause kidney failure, 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 (kidney specialist for children).

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 naturally 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. Many women and girls with TS have some degree of hearing loss which increases over time, relying on hearing aids as hearing loss progresses.

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 pelvises for their height; they also tend to have large hands and feet for their size. 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 some degree of 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 your daughter 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. Adequate calcium and vitamin D are critical to bone health. Doctors must take into consideration the bone size of women with TS when testing for osteoporosis.

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 7 years old, will allow coordination of orthodontic procedures with hormone treatments. Growth hormone therapy tends to increase the growth of the lower jaw while having no significant impact on the growth of the upper jaw. The patient and orthodontist need to take this into consideration during orthodontia treatment.



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.

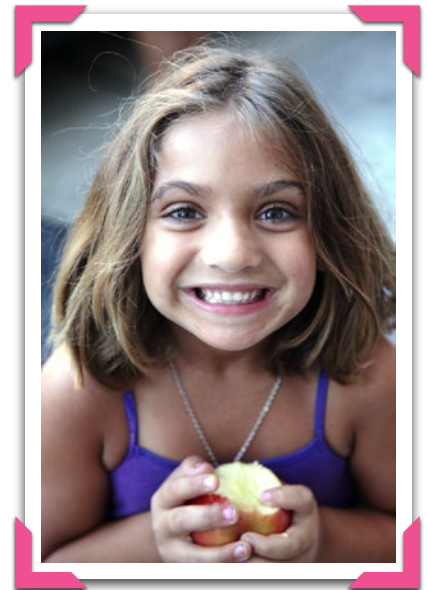
Girls with TS seem to have a higher than average risk of other immune system problems, including celiac disease (gluten sensitivity), inflammatory bowel disease and juvenile rheumatoid arthritis. Girls with digestive complaints or joint problems should be seen promptly by their doctor, who may refer 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. Possible causes of elevated liver enzymes include autoimmunity, excess weight and differences in how the liver was formed. This usually is not related to any problems with liver function. Elevated liver enzymes may improve with estrogen therapy.

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 and is associated with high blood pressure and NIDDM. 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 for her size. Diet and exercise are the keys to weight control. Your child's doctor or nurse can suggest a healthy diet and exercise program or make a referral to a dietitian for counseling if necessary. Good eating and exercise habits should be encouraged in early childhood.



SKIN AND NAILS

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 telangiectasis (small dilated blood vessels).

Any consideration of plastic surgery should take into account the risk of keloid formation (scarring), which is higher in the neck and upper chest.

In girls with a history of swollen feet at birth or those with very small upturned toenails, attention should be given to nail care to avoid infections. Careful pedicures may be recommended, and a consultation with a podiatrist can help those with more severe issues.

Social and Emotional Support _____

GENERAL ISSUES

Growing up can be difficult for any child, and a child who is different by virtue of height or appearance may have more challenges than other children. Girls with TS may feel self-conscious about their condition, which may result in low self-esteem. The support 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 common human reactions and are part of the expected process of adjustment.

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 guide 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 about health care issues** and should include the person with TS in discussions about the implications and treatment of TS. This is especially important as your daughter becomes older and the time approaches to make decisions affecting her sexual development and growth.

You 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 confidence 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 is necessary.

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 a short child according to size rather than age, and this may encourage the development of age-inappropriate social communication and interaction habits.** Discussing this tendency with friends and teachers may be helpful.

Here are some hints for making life easier for a short child and supporting her 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.
- 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.
- Dress according to age rather than size. This may require having clothes altered or learning to sew.
- 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.

SOCIAL ISSUES

The learning and behavior difficulties that affect many girls with TS may affect their social development and relationships with others. Some girls 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 behaviors have on other people, and sometimes they can offend others without meaning to. Some children with TS may require specific social skills training/therapy by an expert typically a licensed psychologist. Speech Language Pathologists help develop oral and non-verbal communication skills.



Parents, teachers and therapists can help a girl with TS to polish her social skills in a variety of ways:

- Teach social skills by role-playing/practicing situations or discussing scenarios and identifying appropriate behavior. Some examples include ordering food and asking for help. She can practice these skills at home and actively observe other children and adults in the same situations. Help her practice using learned skills in new situations.
- Talk 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.
- Provide guidance about the “rules” of play – sharing toys, taking turns, etc. – by engaging your child early in activities where they can learn to work and play with others. Music, drama, dance, singing, 4H clubs, scouting and sports are just a few.
- Identify your own feelings as they arise 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 cheerful and my body is relaxed.”

- Help her 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.”
- Teach 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.

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, natural changes in puberty, how babies are made, how adults show their love for each other in respectful ways – is even more important when sexual development may be affected by a medical condition. There are many helpful books on talking with your children about sexuality and reproduction.

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, a girl with TS may have concerns about her sexuality and her relationships. Reassure her that she is a woman in every way and can expect to have healthy relationships. She will experience the same feelings and confusion every teenage girl experiences as part of growing up. She will want to date 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. When the time is appropriate, talking with a parent, health care provider or trusted adult can help her feel more comfortable with herself so that she can make decisions that are right for her. Professional counseling or contact with other girls and women with TS often is very helpful. Local TS groups exist in many areas, you can find out if one is close to you by contacting TSSUS or asking your child’s doctor or nurse. Young women with TS need a caring gynecologist to support them during their first internal examination and to provide them with the opportunity to discuss sexuality.

EDUCATIONAL AND CAREER ISSUES

Parents, 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 is 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 routines 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.

- Give her early warning and explicit descriptions of changes in routines if unexpected changes are stressful.
- 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 voice recorder (often a feature included with smart phones), getting a teacher outline, asking to copy 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.



Wrapping Up

SUMMARY

Research into the genetic basis of the condition and its physical, psychological, emotional, social, and intellectual aspects has resulted in greater understanding of ways in which the healthy growth and development of girls and women with TS can best be supported.

Many parents agonize, worry and fret about how to tell their daughters about their diagnosis and what it may mean to them. A few research studies concluded that girls feel betrayed if their diagnosis is not disclosed to them in a timely manner. Internet resources for gaining knowledge are useful but they can also offer misinformation, so discuss TS with your daughter before she gains access to information about it on her own. Together your family can deal successfully with difficult conversations with age appropriate answers.

A diagnosis of Turner syndrome is a piece of medical information like any other medical information. It



is not secret or shameful and you may ask, “Is information about TS relevant to this particular relationship/situation, and if so, what information is important to share?” Social media allows children to share information easily so you may choose to talk to your daughter about whom she may want to share information with, how much information to disclose, and how she will share it.

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!

Dr. Melver (who has TS), her husband and their adopted daughter (who also has TS) enjoy the TS conference.

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.

Aorta – the main artery of the body, supplying oxygenated blood to the circulatory system. In humans it passes over the heart from the left ventricle and runs down in front of the backbone.

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 (chromosome analysis).

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

On the following page is a health care checklist for girls and women with TS. Everyone who is diagnosed with TS should be referred promptly to an endocrinologist and receive the listed evaluations. It is crucial to assess each of these systems to look for important conditions that could affect your daughter’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.



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	At Each Visit, At Least Yearly	Yearly	Every 3 - 5 Years	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	If age appropriate	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		Begin repeat exams at age 4		
Orthodontic evaluation					At age 7
Counseling re: sexuality issues and vocation/ college or work plans		If age appropriate			
Cardiac evaluation; echocardiogram or MRI	X			Begin repeat exams at age 12	
Bone densitometry				Begin repeat exams at age 15	
Blood tests: liver and kidney function, fasting lipids and blood glucose	X		Begin repeat exams at age 18		As recommended



Notes or Personal Health Information

Lined area for notes or personal health information, consisting of 30 horizontal blue lines.

Whether your daughter is 2 days old or 18 years old, hearing that you daughter has Turner syndrome may seem scary. Everyone generally responds with "What is Turner syndrome, I've never heard of that?" Take a deep breath and exhale. Your mind is probably spinning with questions and concerns and we'll address the main ones in this booklet as best we can. Then we hope you'll contact us for more personal information.

TSSUS has supportive and knowledgeable volunteers that want to share their experiences with you as well as help you on your journey of learning about what TS means to your family. Please contact our office at 800-365-9944 or email: tssus@turnersyndrome.org and a volunteer will contact you directly.



"I have never used my TS as an excuse, if anything I used it as a springboard to prove people wrong and give me the drive to succeed. We need to strive for whatever we want because we are strong!"
- Nicole

"I'd be different without Turner syndrome but not better. Everyone deals with something."

- Trudy

Friends
Forever

