

## Turner syndrome *at a Glance*

Turner syndrome (TS) results when all or part of one of the X chromosomes is missing in females. Short stature and lack of pubertal development are seen in most girls with TS. Heart defects, regulations of the amount of fluid in the tissues (lymphatic system), endocrine problems and psychosocial problems are also common. TS can also affect kidney development and hearing.

TS affects about 1 in 2500 females.

### Learn More

#### Consistent findings: Occur in 90-100% of girls with TS

##### Short stature

The most common feature of TS is short stature

- In TS, growth failure begins at birth. Without extra growth hormone, girls usually won't have a pubertal growth spurt. They may grow at a slower rate until their 20s.
  - The average adult height of a woman with TS (not treated with growth hormone) is 4 feet, 8 inches (142 cm). However, a few women will reach 5 feet.
  - Females treated with growth hormone may reach a low but normal adult height.

##### Premature ovarian failure

Although the ovaries develop normally at first, they undergo changes similar to menopause before birth causing the ovaries to be non-functioning at birth. That is they do not produce eggs or estrogen. This means that there is insufficient estrogen to cause the changes associated with puberty.

##### Sexual development

- Lack of sexual development during adolescence is another hallmark feature of TS.
  - Breast development
  - Feminine body contours
  - Menstruation
- Pubertal delay may exacerbate the negative psychosocial effects of later ovarian

## development

- Excessive shyness
- Social anxiety
- Delayed sexual development
- Sexual development is affected because the ovaries often stop working early in life.
- Ten to 15 percent of girls 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 under arm hair grows in 10-12-year-old girls without puberty because their adrenal glands are producing normal amounts of other hormones, leading to an imbalance.
- When a girl isn't developing with her peers she may become self-conscious about her appearance. This delayed start of puberty may affect a girl's self-image, her friendships with peers, and healthy sexual development. It may make her feel different from her peers
- The ovaries can no longer produce eggs so most women with TS do not ovulate and are infertile.
- The uterus and vagina are normally formed and therefore it is possible to achieve pregnancy with a donated egg and hormonal support.

## **Frequent findings: Occur in at least 30% of girls with TS**

### **Intelligence**

- Most girls are of normal to slightly below normal intelligence.
- Girls may have difficulty with spatial processing (imagining objects in relation to another), non-verbal memory, and attention. Can lead to problems with:
  - Math
  - Sense of direction
  - Manual dexterity
  - Social skills

### **Physical Features of TS**

About a third of girls with TS will have features related to the poor development of the lymphatic system before birth. Lymph drains from the tissues into the blood. This system carries fluid that has drained from the tissues into the blood

If not working properly, fluid collects in the tissues. This is called lymphedema.

- In girls with TS, this fluid often collects around the neck before birth. This may affect the appearance of the neck and ears.
  - After birth, fluid often remains in the hands and feet.
    - May lead to small narrow fingernails and toenails that turn up
    - Hands and feet may also be “puffy”
  - It may lead to a thick, short neck, “webbed” appearance, and/or low hairline.
  - These features may be barely noticeable
    - However, may cause self-consciousness or embarrassment for some girls.
    - Some girls use make-up tricks or clothing to disguise these features.
  - Some of the features can be improved with treatment.
    - Lymphedema may improve with manual compressive therapy
      - Massage
      - Exercises
      - Support garments
      - Skin care
  - A small number of girls have plastic surgery to improve the appearance of the neck and ears
- 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.
  - Good dental care is important.
- Short ring finger (the metacarpal is short, not the finger itself)
- Pectus excavatum (caved or sunken in chest wall)
- Eyes
  - Slight droop to eyelids
  - Strabismus (lazy eye)
  - Red-green color blindness
- Broad chest
  - Not as obvious after breast development
- Arms may not fully extend at the elbows
- Scoliosis (curvature of the spine)
- Flat feet

## Heart abnormalities

- 1/3 of girls with TS have a heart abnormality

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- Heart defects include:
  - Coarctation (narrowing) of the aorta
  - Bicuspid aorta valve
- Higher risk for hypertension/high blood pressure
- Should be aware of symptoms of dissection of aorta (rare complication)
  - Sudden, sharp, stabbing, tearing, ripping chest pain
  - Intense anxiety
  - Rapid pulse
  - Profuse sweating nausea and vomiting
  - Dizziness, fainting
  - Shortness of breath

### **Kidney abnormalities**

- 25-40% have kidney abnormalities
- Kidneys may be unusual shaped or in a unusual position
- Kidney abnormalities may not cause medical problems
  - May result in increased urinary tract abnormalities and increased blood pressure.

### **Thyroid abnormalities**

- Hypothyroidism (low level of thyroid hormone) occurs frequently in individuals with TS.

### **Ear infections and hearing loss**

- Ear infections are extremely common in TS girls particularly in infancy and early childhood.
- The majority (~60%) of TS women will also develop early sensorineural (nerve) hearing loss
  - Woman may require hearing aids earlier than the general population.

### **Autoimmune disorders**

- Girls with TS seem to have a higher than normal risk of other autoimmune disorders.
  - Celiac disease (gluten sensitivity)
  - Inflammatory bowel disease
  - Juvenile rheumatoid arthritis

- Girls and women with TS are at increased risk for a cluster of conditions sometimes called the “metabolic syndrome.”
  - Clinical signs include:
    - High blood pressure
    - Abnormal blood levels of cholesterol
    - Non-insulin-dependent diabetes mellitus
    - Obesity
    - Increased insulin secretion
    - Increased uric acid secretion (related to gout).
    - Many of these conditions can lead to cardiovascular (heart and blood vessel) disease,
    - Careful monitoring and prompt treatment of these problems is needed.

## Obesity

- A common problem in TS, especially as teens age.
- Extra weight on a shorter person is more noticeable and can lead to a poor self-image.
- Obesity is associated with high blood pressure and non-insulin requiring diabetes mellitus.
  - Girls with TS are at increased risk for these problems to begin with and it is important to maintain a healthy weight.
- Diet and exercise are the keys to weight control in TS

## Scars

- Girls and women with TS are more likely to form exaggerated scars
- Even simple wounds like ear piercing and mole removal can heal with thick scar tissue, known as keloid formation

## Intelligence

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  - Sense of direction
  - Manual dexterity
  - Social skills

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## ***Things to Think About***

### **1. Dietary / Medical Needs**

#### **What you Need to Know**

No special diet is required for TS although a well balanced diet is important

The list of possible medical problems in TS can be extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.

#### **Learn More**

School age children with TS may have annual doctor and specialist visits to monitor medical conditions.

#### **What you can do**

- A yearly check up and studies as needed should occur in the child's Medical Home.
- Regular eye and hearing exams
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.

### **2. Education Supports**

#### **What you need to Know**

The intelligence of girls with TS varies just like that of the general population. It may be slightly lower than normal overall. Cognitive issues with Turner's syndrome include difficulties in visuospatial and executive skills, visual working memory, and mathematics. Girls with TS tend to excel at verbal skills.

Girls may have what is known as a "Turner neurocognitive phenotype." This means that

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complete or partial absence of the X chromosome impacts the development of the nervous system. This often leads to difficulty in nonverbal areas or performance based tasks (i.e. visual-spatial, intuitive, organization, evaluative, and processing functions.) These are sometimes referred to as nonverbal learning disabilities or NLD. Girls with TS tend to score lower on performance compared to verbal sections on intelligence tests.

### **Commonly observed challenges 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)
- Clumsiness (psychomotor problems and poor manual spatial awareness)
- Understanding time
- Math
  - Understanding:
    - Place value
    - Signs
    - Borrowing
    - Division
  - Sequencing needed to solve problems
  - Difficulty with abstract concepts
  - Hard time visualizing 3D object in a 2D format

#### **Writing**

- Spelling
- Punctuation
- Capitalization
- Drawing
- Copying
- Ordering
- Handwriting

#### **Reading comprehension requires spatial skill**

- Grammar
- Writing and drawing

### **Learn more**

#### **Executive function problems**

- Hard time planning and structuring tasks/work

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- Self organization may be a challenge

### **Motor problems**

- May be improved by hormonal therapy at puberty time

### **Difficulty in concentration**

- Repetition
- Short amount of information at a time
- Quiet calm environment away from distractions

### **Memory**

- Short term memory problems
- Long term memories usually good

### **Difficulty following instructions**

- Be clear
- Simple language
- Specific and exact directions

### **Fine motor skills**

- Trouble with tasks requiring dexterity
- Getting changed may be hard with buttons

### **A rare cause of Turner Syndrome may lead to increased risk of mental retardation**

- Small ring X chromosome

## **What You Can Do**

### **Help with weekly clean-out**

- Organization of desk, locker, backpack, room

### **Teach to use a schedule book for recording assignments**

- Color-coded folders for keeping papers and homework organized

### **Give clear instructions for working on projects and homework.**

- Set priorities
- Break task into steps
- Developing a schedule and plan
- Teach structures and templates for various tasks
  - Book reports
  - Spelling assignments, etc.



### **Help to notice similarities and differences among tasks.**

- Find clues in a new task that relate to a previous task
- Adapt strategies for dealing with variations in tasks (generalizing, then adapting).

### **Help to see relationships among parts and the whole structure**

### **Provide outline of day to allow to feel more confident.**

- Use visual aids
- Handouts
- Realistic target settings
- Use reward systems and praise

### **Hand-written note taking may be a problem.**

- Use a tape recorder
- Have teacher provide outline
- Copy notes from classmates
- Use a word processor in class.

### **Other**

- Routine is important
- Seat child facing teacher
- Make eye contact
- Keep background noise to a minimum or provide a quiet spot
- Detailed instructions and repetition
- Untimed tests because of slower processing time

### **Possible treatments for Non verbal Learning Disabilities (NLD) include:**

See <http://www.turnersyndromefoundation.org/learning.html#1>.

### **Auditory integration therapy**

- Improves auditory processing and the way the brain processes information, particularly in normally noisy environments
- May be an improvement in reading, writing, speech, and language development, and ability to handle noisy environments and social situations.
- May be improvements in comprehension and increased development of neural pathways that could improve handwriting and other fine and gross motor skills

### **Cognitive behavioral therapy**

- Guided therapy that enables people with NLD to perform better on everyday tasks. For example:
  - Concentrating and focusing on tasks
  - Making appropriate choices in social situations
- Begins with an assessment of cognitive/thought patterns
  - Uses this information to create a plan

### **Sensory integration therapy**

- Helps with organizing and interpreting sensory signals in the environment.
  - particularly for those who are overwhelmed by the many sensory signals that come from the environment such as smells, tastes, and sights

### **Social skills groups**

- Helps increase social success
  - gives tips and strategies for confronting the challenges of being socially successful

### **Homework performance can be improved by following a few strategies**

- Plan study time with a schedule
- Prioritize with a to-do list that includes
  - Homework that needs to be done
  - Activities
  - Chores
  - Plans that have been made
- Organize each day and
  - Make sure everything has been packed for school.
  - Binders could be used to organize work for each subject
- Set up a workspace that has good lighting, plenty of space, no clutter, and little or no noise.
  - Establishing “rules” such as avoidance of loud music, TV, and text messaging while doing homework.
- Encourage the student to ask for help from the teacher
- Consider the use of a study buddy or homework partner
  - Can be supportive of friend’s needs resulting from her challenges
  - Helpful in organizing assignments, task division, and time management

## **3. Behavioral and Sensory Support**

## What you Need to Know

Girls may have subtle difficulties in behavior function. These can include difficulties in:

- Nonverbal skills
- Slowed response times
- Increased rates of attention deficit disorder
- Increased risk for social isolation
- Maturity
- Depression
- Anxiety
- Higher rate of ADD
- Shyness, social anxiety, and reduced self-esteem related to the premature ovarian failure and fertility issues.
  - These things affect relationships with other children
  - Individuals tend to have more immature behavior
  - Difficulty concentrating
  - Problems with over activity
- Social problems related to spatial understanding
  - Spatial disorders may affect social relationships. May be rigid and inflexible with regard to behavior of others
  - May invade personal space
  - May not be able to understand the effects on others of things they may have said/done
  - Struggle with order in environment and may try to maintain rigid and inflexible order
- Teenagers are prone to
  - Immaturity
  - Anxiety
  - Depression
  - Socially withdrawal
  - Tend to begin dating and sexual activity at a later age than other girls.
- Psychosocial implications of NLD (Non Verbal Learning Disability). The nonverbal learning difficulties that affect many girls with TS may affect their social development and relationships with others.
  - Difficulties in adapting to new situations may include:
    - Inappropriate behavior in a new situation
    - Issues with social skills and making new friends

- Difficulty gauging how to act in a social environment
- Possible anxiety and depression
- Possible decrease in physical activity levels
- Reading and interpreting facial expressions and other nonverbal social cues.
  - Such as body language, touch and tone of voice.
- Easily overwhelmed by new situations and trouble identifying their own emotions, as well as others’.
- May not realize the effects their behavior has on other people
- May be offensive without meaning to be
- Girls with TS may feel self-conscious and embarrassed about their condition,
  - May result in low self-esteem.

## What You Can Do

- Help her practice situations that feel uncomfortable in public and help her try out ways of dealing with them.
- Help her learn social skills.
  - Simple things like ordering food in a restaurant or asking for help in a department store may be intimidating.
  - Practice these skills at home
    - Observe/talk about other children and adults in the same situations.
  - Dress according to age rather than size.
    - This may mean having clothes altered or sew on own.
  - Encourage development of skills that allow for social interaction and competition with a variety of other children.
    - Teach “rules of play”
      - Taking turns
      - Sharing
    - Social space
    - Eye contact
    - Teach how to read facial features and other social cues.
- There are many activities that help children develop skills and learn to work and play with others.
  - Music
  - Drama
  - Dance
  - Singing

- 4H clubs
- Scouting
- Sports
- Help child understand the effect her behavior has on others.
  - Role play
- Teach stress reduction techniques

### **Maintaining and making new friends**

- Social interactions with new or established friends can start with more structure and one-on-one play.

### **Peer pressure**

- Teachers and parents can be more aware of interactions between students and try to support social skill development.

### **Bullying**

- There are anti-bullying programs in many school districts and many schools have provisions in their codes of conduct about bullying and its consequences.
- Talk to the child if you think that she is being bullied at school and intervene, if necessary.
- Teasing may be misunderstood and taken more seriously than intended so adult can help clarify.
- Stop the bullying before it gets worse.

### **Adapting to new situations**

- Go to the new environment beforehand and walk through it.
- Provide a list of activities that will occur
  - Helps her feel prepared

### **Anxiety and depression**

Anxiety and depression may be seen in those with NLD. To deal with these major issues, here are some tips for coping at home and at school:

- Exercise regularly with a set routine.
- Learn to relaxation techniques
- Establish a regular sleep schedule.
- Schedule “down” time

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- Eat healthy.
- Have a support network available that can help.
- Learn time management and organizational skills.
- Use praise.
- Cognitive-behavioral therapy or mindfulness techniques can help with depression issues as can talking to a therapist, psychologist or psychiatrist.
- Transition: proactive measures can help.
  - Preparing in advance for any major life transitions and guiding the child with NLD through each step of the transition to give a clear sense of direction on what is to occur throughout this new stage of life.

## 4. Physical Activity, Trips, Events

### What you need to know

- Exercise is important and should be encouraged. Eligibility for competitive sports should be determined by a cardiologist.
  - Gross motor skills
    - Coordination may be problem and make individuals a target for teasing.
    - Practice can help.
    - Hand- eye coordination is often poor
  - May do better with individual sports vs. team sports.
    - Swimming
    - Dance
    - Gymnastics
    - Track and Field
  - Lack of stamina and energy levels can be problem
    - Individuals may not know how to pace themselves or when to rest.
- Field Trips
  - May have trouble dealing with unexpected changes in routines.
  - Give warning and descriptions of what will happen
    - Review step-by-step exactly what she'll need to do.
  - Arrange for a buddy or assign an aide/chaperone
  - Provide a list of activities so the child will know what is going on and be prepared.

## 5. School Absences & Fatigue

### What you need to know

Children with Turner may be absent due to illness and/or medical appointments. Help to make transitions in and out of school as seamless as possible

Individuals with Turner should not have excess fatigue.

## 6. Emergency Planning

### What you need to know

- Develop an emergency plan if necessary, depending on the needs of individual children.

## 7. Resources

### Turner Syndrome Society

The Turner Syndrome Society of the United States provides many resources for parents and women in addition to support and event announcements, research, and advocate opportunities. <http://turnersyndrome.org>

### Genetics Home Reference

Consumer-friendly information about human genetics from the US National Library of Medicine <https://ghr.nlm.nih.gov/condition/turner-syndrome>

### Turner Syndrome Foundation

The Foundation leads an advocacy effort to identify the gaps those with Turner Syndrome face as they transition from pediatric to adult care. <http://www.turnersyndromefoundation.org>

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## Turner Syndrome Article

The article titled “The cognitive phenotype of Turner syndrome: Specific learning disabilities” discusses the need for a detailed assessment of the child’s cognitive difficulties prior to the onset of formal schooling. <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2742423/>

## Turner Syndrome Support Society (UK)

TSSS offers support, advice, and information to women and girls with Turner Syndrome and their families. Some publications of particular interest may be “A Teenagers Guide to Turner Syndrome”, and “How to help your child survive and succeed at school”. <http://tss.org.uk/>

## Guide to Primary Ovarian Insufficiency (POI)

This guide, titled "Moving Forward" developed by Children's Hospital Boston provides information for parents and women with primary ovarian insufficiency. While the guide is targeted to women with galactosemia, it may provide useful information for dealing with POI.

[https://newenglandconsortium.org/brochures/Your\\_Guide\\_to\\_Galactosemia\\_and\\_POI.pdf](https://newenglandconsortium.org/brochures/Your_Guide_to_Galactosemia_and_POI.pdf)

## Classroom Accommodations for Students with Visual Issues

The Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C., in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

<https://www.bouldervt.com/>

## 8. Meet a Child

### Flying with Ellie

*GEMSS would like to thank Ellie and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!*





(The butterfly is a logo for the Turner syndrome groups and Ellie is seen with her wings in these pictures.)

Ellie loves to snuggle each night in her Pennsylvania (USA) home with each one of her 4 older brothers and sisters and get hugs and kisses from all of them. In fact, one recent night when her brother went to a sleepover, Ellie cried herself to sleep because she missed their nightly ritual. Then each morning, when her sibs get on the bus, the same hugs and kisses are passed between them. She is also very affectionate, extra friendly, and very caring, especially of animals and babies. Her mother Jordi says that no one needs to ask Ellie to help, she just automatically notices and says 'Can I help you?' "She has amazing compassion and her spirit is so deep," says Jordi.

Ellie has come a long way since her weeks in the NICU as a preemie who came 6-7 weeks early. She had already graduated from Early Supports and Services when she received a diagnosis of Turners syndrome at age 3. Up until that time, she had been diagnosed

with cerebral palsy that was caused by lack of oxygen from the cord being tightly wrapped around her neck at birth. But Jordi persisted and wanted a medical work up for Ellie's short stature. It was the testing from the endocrinologist that confirmed the cause of her small stature was Turners.

The following year, Ellie had surgery to correct the reflux going into her kidney which is a single, horseshoe shaped kidney. But as a result of the delay in getting diagnosed, her ureters and kidney were damaged by scar tissue and Ellie now has high blood pressure as a result. Ellie also has some fine and gross motor challenges, low muscle tone, some speech and other issues. Her academic challenges consist of math and spatial reasoning and social learning.

Jordi has wonderful hopes and dreams for her daughter. Of course, to be healthy, but also that Ellie will become fully who she really is and will make a difference in other people's lives. "She is a fighter and she has such grace. We want her to do whatever she wants to do and hope that her syndrome doesn't hold her back. Maybe she will be one of the rare women who can have a baby," says Jordi.

Jordi is feeling very good about Ellie's transition into a preschool program with children who do not all have disabilities. She is hopeful about the school year and all of the social opportunities Ellie will have. She is starting to make friends in the new program. It sounds like her strengths in the social area are already helping her connect and communicate! She does not want Ellie to have an aide with her as she doesn't want her to stick out socially. She is thinking of the future and the possibility of self-esteem issues, and doesn't want Ellie to feel she is different or feel insecure. It sounds like the phrase "Only as special as necessary" would apply to their approach to supports!

Jordi is a registered nurse and former child and family therapist. She is a substitute nurse in all 5 of her district's schools and has had lots of experience with children.

Her **advice to parents** is to:

- Be your child's best advocate
- Do all you possibly can to give them the very best life possible.
- Try to be involved in groups and learn from all the information sources available.

Her **advice to school nurses and teachers** is to:

- Know each child in the school who has a special condition.
- Be their advocates and educate yourself on their condition.
- Use your knowledge and skills to educate others on the team.
- Take an interest in this child.
- Know how vitally important it is to understand how this condition affects the learning process.
- It is a fairness issue to make sure every child gets what she/he needs.

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