Turner Syndrome

Background

Turner syndrome (TS) is a genetic condition characterized by short stature, delayed or absent puberty, infertility, heart defects and certain learning disabilities. TS is caused by a missing or partially missing X chromosome, only affecting females.

Signs and symptoms related to growth and bone development may be subtle and develop slowly. Some features may be identified before birth due to effects on the developing heart and kidneys, while others are noticeable at birth or during infancy. As the child grows, expected growth spurts may not occur.

TS can also affect the heart, thyroid, and kidneys. Associated conditions may include congenital heart defects, hypertension, and urinary tract infections. Individuals with TS are also at increased risk for metabolic syndrome, including central obesity, insulin resistance (pre-diabetes), high blood pressure, high cholesterol and type 2 diabetes.

An individual with TS often has a cognitive profile that includes normal intelligence and verbal capabilities but weaknesses in the areas of visual–spatial, executive, and social cognitive function. This includes variables in things like nonverbal communication, slowed response times and immaturity. A child may have trouble recognizing social cues and maintaining personal boundaries. Things like facial expressions are also easily misinterpreted. Issues of spatial awareness and abstract concepts make certain subjects particularly challenging. Spatial awareness can also affect spelling, punctuation, mapping, understanding time, changing point of view, and even drawing and handwriting.

While there is no cure for TS, treatments aim to manage the symptoms and complications. The primary treatment for nearly all girls with TS includes hormone therapies. Growth hormone therapy is usually given as a daily injection to increase height during early childhood to the teen years. Estrogen therapy is usually started around the age of 11 or 12 to begin puberty.

Top Takeaways for School

Turner syndrome (TS) is a chromosomal abnormality affecting females. The main symptom is short stature.

Delays in physical growth and sexual development may lead to issues with low self-esteem, anxiety, and depression.

Students with TS may have normal intelligence but often experience difficulties with visual-spatial and executive functioning. Girls with TS also have a higher rate of attention-deficit/hyperactivity disorder (ADHD).

Students with TS are prone to recurrent ear infections. Hearing loss and sensitivity to noise could require coordination and planning (e.g., alarms, announcements, assemblies).

Most students with TS respond well to an organized and structured plan. It is helpful for them to know the daily schedule, and some may find a sudden change to routine difficult.

Girls with TS are highly verbal but may struggle with understanding social relationships. They may be rigid and inflexible towards behaviors of others. Social cues and facial expressions are often misunderstood.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnoses: Impaired physical mobility, impaired swallowing, low situational self-esteem, ineffective breathing patterns
- Student-specific triggers, avoidance, or intervention
- Elimination interventions and equipment (consider catheterization brand/system, French size, frequency, and cleaning techniques; location of procedure; level of assistance)
- Use of specialized equipment, adaptive equipment, and orthotics
- Skin check, pressure relief techniques
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Can strategies be implemented to assist the student with executive function (e.g., plan, prompts, organizers, agendas)?
4. Does the student need additional adult support to access the academic curriculum in the least restrictive environment?
5. Would schedule flexibility support the student?
6. Does the student need support with gross or fine motor skills in the classroom?
7. Does the classroom environment support the student's needs and/or equipment? (e.g., desk/seating options, ability to reach objects, flash pass for bathroom or nurse)
8. Will staff receive education/training to implement the student-specific emergency plan?

Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinics
kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinics

Tuner Syndrome Foundation
turnersyndromefoundation.org/

Turner Syndrome Support Society: How to help your child survive and succeed at school: A Guide for Parents and Teachers
tss.org.uk/downloads/TSSS%20education%20booklet.pdf

For more information, please scan the QR code or visit: KennedyKrieger.org/SHNIC

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