

Turner Syndrome (TS) *For Healthcare Providers*

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Turner syndrome will have all of these features.

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

- Short stature
- Premature ovarian failure
- Delayed sexual development
- Psychosocial effects of delayed ovarian development
 - Excessive shyness
 - Social anxiety
 - Delayed sexual development

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, non-verbal memory, and attention. Leads to problems with:
 - Math
 - Sense of direction
 - Manual dexterity
 - Social skills

Physical Features of TS

- **Lymphedema**
 - A cystic hygroma often develops in utero and may persist until birth. Resolution of the cystic hygroma may lead to a thick, short neck, “webbed” appearance, and/or low hairline.
 - Edema in an unusual pattern involving the dorsa of the hands and feet can be present at birth
 - Occasionally this persists into childhood
 - Can lead to abnormal nail beds with vertical nails

- **Heart abnormalities:**
 - Coarctation of the aorta
 - Bicuspid aorta valve
 - Hypertension
 - Dissection of aorta (rare complication)
- **Kidney abnormalities**
 - Often a horseshoe kidney
 - May not affect kidney function
- **Thyroid abnormalities**
 - Hypothyroidism
- **Ear infections and hearing loss**
 - The majority of TS women will also develop early sensorineural hearing loss
- **Eyes**
 - Strabismus
 - Red-green color blindness
- **Autoimmune disorders**
 - Girls with TS seem to have a higher than normal risk of other autoimmune disorders.
 - Celiac disease
 - Inflammatory bowel disease
 - Juvenile rheumatoid arthritis
 - Girls and women with TS are at increased risk for “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
- **Other possible physical features**
 - Crowded teeth due to a small, receding lower jaw and narrow, high-arched palate.
 - Short ring finger
 - Pectus excavatum
 - Broad chest
 - Scoliosis
 - Flat feet
 - Keloid scar formation

Recommended Routine Surveillance

- Cardiac surveillance
- Ophthalmologic evaluation
- Hearing evaluation
- Renal surveillance
- Yearly urine culture and annual BUN and creatine levels
- Monitor thyroid levels
- Routine dental care
- Regular speech/language evaluations if appropriate

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
 - Surveillance as needed
- Developmental evaluation
 - Speech therapy
 - Physical therapy
 - Occupational therapy
- Endocrinologist
 - Monitor hormone levels
 - Growth hormone therapy
 - Estrogen replacement therapy
- ENT
 - Hearing loss
- Geneticist / Genetic Counselor:
 - Diagnosis
 - Coordination of care
 - Genetic risk for family
 - Clinical trials
- Autoimmune
 - Monitor autoimmune disorders
- Nephrologist/Urologist

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- Monitor anomalies
- Bun/Creatine levels
- Psychologist
 - ADHD
 - Social skills deficits
 - Anxiety and depression

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient _____ has been diagnosed with Turner syndrome. Delayed speech, fine and gross motor delay, social skill deficits, and infertility characterize Turner syndrome. Medical complications with Turner syndrome include management of cardiac defects, autoimmune diseases, hypothyroidism, and renal anomalies. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

“[Turner Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



Resources

Turner Syndrome Treatment and Management – Endorsed by the AAP

<http://www.eje-online.org/content/177/3/G1.full>

Turner Syndrome Society

<http://turnersyndrome.org>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/turner-syndrome>