Williams Syndrome (WS)

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 WS will have all of these features.*

- **Heart disease**
  - Pulmonary stenosis
- **Unique facial appearance**
  - Prominent lips with an open mouth
  - A long philtrum
  - Low nasal bridge with an upturned tip
  - Epicanthal folds
  - Partial absence of teeth or defective tooth enamel possible
  - Stellate/lacy iris pattern
- **Connective tissue problems**
  - Joint laxity and limitation
  - Soft, lax skin
  - Hypotonia
  - Inguinal/umbilical hernia
  - Bowel/bladder diverticula
  - Rectal prolapse
- **Hoarse or low pitched voice**
- **Progressive sensorineural hearing loss**
- **Vision/Eyes**
  - Farsighted
  - Strabismus
- **Intellectual disability (usually mild)**
  - Specific cognitive profile: strengths in verbal short term memory and language
- **Unique personality characteristics**
  - Overfriendly
  - Empathetic
  - Generalized anxiety
  - Attention deficit disorder
- **Growth abnormalities**
- Prenatal growth deficiency
- Failure to thrive in infancy
- Poor weight gain and linear growth in first four years
- Adults usually below 3rd % in height

- Endocrine abnormalities
  - Hypercalcemia
  - Hypercalciuria

**Recommended Routine Surveillance**

- Plotting growth on Williams syndrome growth charts
- Routine Blood work:
  - Serum ionized calcium and Thyroid studies
- Ophthalmologic and auditory evaluation
- Routine dental care
- Developmental evaluations

**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 decompensating.
- 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 for mitral valve prolapse, aortic insufficiency, hypertension, long QT interval, arterial stenosis
  - Cardiac defects may require surgery
- **Dentist:**
  - Microdontia
  - Enamel hypoplasia
  - Malocclusion
- **Developmental specialist:**
  - Speech therapy
  - Physical therapy
  - Occupational therapy
- **ENT:**

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  [www.gemssforschools.org](http://www.gemssforschools.org)
• Chromic otitis media
• Progressive sensorineural hearing loss
• Hypersensitivity to sounds

• Endocrinologist:
  • Hypercalcemia/hypercalciuria
  • Hypothyroidism
  • Early puberty
  • Increased in diabetes especially in adults

• Gastroenterologist:
  • Significant feeding problems may occur
  • Gastrointestinal reflux
  • Abdominal pain

• Geneticist / Genetic Counselor:
  • Diagnosis
  • Coordination of care
  • Genetic risk for family
  • Clinical trials

• Nephrologist:
  • Management of nephrocalcinosis, persistent hypercalcemia, and/or hypercalcemia

• Ophthalmology surveillance:
  • Strabismus
  • Hyperopia
  • Cataracts (adults)

• Orthopedist:
  • Joint limitation or laxity
  • Hypotonia
  • Scoliosis

• Psychological and psychiatric evaluation:
  • Attention deficit disorder
  • Anxiety
  • Phobias

• Urologist:
  • Urinary tract abnormalities

Sample Forms

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

  My patient______________________ has been diagnosed with Williams syndrome.  
  Cardiovascular disease, characteristic facial features and behaviors, connective tissue abnormalities, intellectual disability, growth abnormalities, and endocrine abnormalities

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characterize Williams syndrome. Medical complications with Williams syndrome include management of calcium levels, feeding difficulties, heart defects, hypertension and diabetes. Because of these, ______ needs the following accommodations.

Seven Important Aspects of School Life

“Williams 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

GeneReviews: Williams Syndrome

Williams Syndrome Association
http://www.williams-syndrome.org

American Academy of Pediatrics (AAP) – Health Supervision Guidelines
http://pediatrics.aappublications.org/content/pediatrics/127/1/195.full.pdf

Genetics Home Reference