

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

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- 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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- Chronic 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

<http://www.ncbi.nlm.nih.gov/books/NBK1249/>

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

<https://ghr.nlm.nih.gov/condition/williams-syndrome>