



Chr
7

Williams syndrome is caused by a **deletion on chromosome 7** (~30 genes), leading to **abnormal elastin production** resulting in **cardiovascular and connective tissue issues**. It is typically not inherited, but can be transmitted in an **autosomal dominant** manner, with a prevalence of 1 in 7,500 to 10,000.

AD

Autosomal
dominant

CLINICAL FEATURES

Face:

- “Elfin facies”
- Prominent earlobes
- Broad forehead with bitemporal narrowing
- Periorbital fullness
- Malar hypoplasia
- Long philtrum
- **Full lips**



CVS:

- **Supravalvular aortic stenosis**
 - With or without coarctation
- Pulmonary artery stenosis
- Renal artery stenosis



Neurologic:

- **Stellate iris**
- “Cocktail party personality”
- Sleep disturbance
- **Intellectual disability**
- **Developmental delay**



Gastrointestinal:

- **Hernias** (umbilical, inguinal)
- **Rectal prolapse**
- Failure to thrive
- **Constipation**

Endocrine:

- **HYPERcalcemia**
- Hypothyroidism



DIAGNOSIS

Confirmatory Genetic Testing

- **Chromosomal microarray**
- **Fluorescence in situ hybridization [FISH]**
- Multiplex ligation-dependent probe amplification [MLPA]
- **Deletion of the 7q11.23 region, including the elastin gene (ELN)**



SURVEILLANCE

Labs:

- At diagnosis, then annually*
- Serum calcium
 - Urine calcium
 - Thyroid function tests

Imaging:

- Echocardiography and ECG (*at diagnosis*)
- Renal bladder ultrasound (*annually*)

At diagnosis, then annually, or PRN if issues arise.

- **Audiology**
- **Ophthalmology**

MANAGEMENT

Management focuses on supportive care and early intervention.

- **Physical, occupational, and speech therapies** for gross motor, fine motor, and language development.
- Enrolling in **early intervention programs** or special education helps address developmental delays and learning difficulties.
- Monitor for **hypercalciuria** and involvement of nephrology as needed.
- **Referral to cardiology** if anomalies on echocardiography or ECG.
- Aggressive **management of constipation** given risk of rectal prolapse.



February 2025

Dr. Katharine V. Jensen (Pediatric Resident, University of Alberta) and Dr. Karen Forbes (Professor of Pediatrics, University of Alberta) for www.pedscases.com