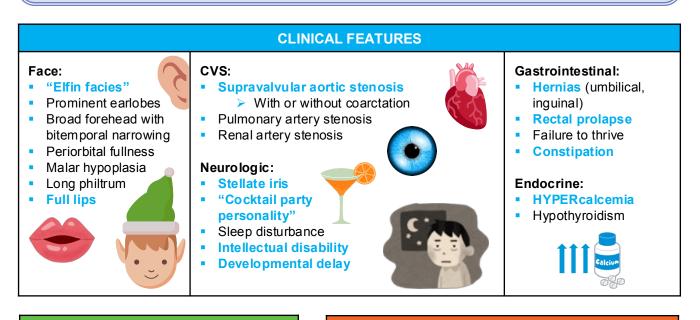


WILLIAM SYNDROME





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



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)

SURVEILLANCELabs:Imaging:At diagnosis, then annually• Echocardiography and
ECG (at diagnosis)• Urine calcium• 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.



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