

## **Condition:** Williams syndrome (Williams-Bueren syndrome)

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### **Inheritance:**

Chromosomal; can be transmitted as dominant trait.

### **Genetic etiology:**

Microdeletion of chromosome 7q11.2 leading to deletion of elastin gene. Usually not visible cytogenetically, but detectable by FISH or comparative genomic hybridization.

### **Frequency:**

Approximately 1:7,500.

### **Clinical features:**

Infants are often born post-term with low birth weight. Many have hypercalcemia in the neonatal period. Supravalvar aortic stenosis is common. Characteristic facial features include stellate iris and long, smooth philtrum. There is developmental delay with a distinctive, outgoing personality. Failure to thrive is common in infancy, and growth delay persists. Hypertension may occur, sometimes due to renal artery stenosis.

### **Management:**

Surgical management of congenital anomalies; supportive care and management of cognitive problems.

### **Genetic counseling:**

Affected individuals can transmit the chromosomal deletion as dominant trait; couples with a sporadically affected child have low risk of recurrence. Some unaffected parents carry a chromosome 7 inversion that may predispose to the deletion, although testing for this inversion is not offered standardly. Prenatal diagnosis is possible.