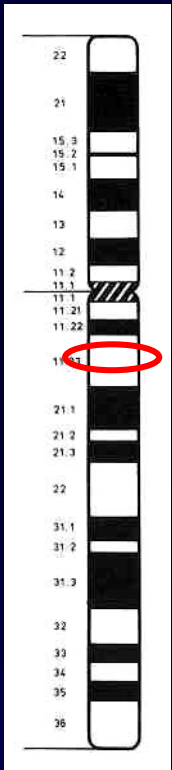


# Williams Syndrome



- **Incidence 1:20,000**
- **Supravalvular aortic stenosis**
- **Long philtrum, “elfin” face, stellate iris**
- **Hoarse voice, hyperextensibility**
- **Social seeking personality**
- **Mental deficiency, mild-moderate**
- **Infantile hypercalcemia**
- **7q11.23 microdeletion (requires FISH)**

# Williams Syndrome

Often present with supravalvular aortic stenosis or because of learning delay and loquacious Personality.





Stellate iris pattern is usually not evident in individuals with dark iris color.



# Abnormal FISH Test (UF Cytogenetics Laboratory)



Abnormal #7, missing the Williams Syndrome Critical Region (WSCR) probe.

# Williams Syndrome: Repeat Units at Breakpoints

Franke U: Hum Mol Genet, 1999

