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

GTF2I: multifunctional transcription factor TFII-I
NCF1: neutrophil cytosolic factor 1 (oxidative burst)

7q11.23 Deletion Region

GTF2IP1
NCF1P1
Elastin
GTF2I
NCF1

1.6 - 2.0 Mb

300 Kb region of repeats

GTF2I: multifunctional transcription factor TFII-I
NCF1: neutrophil cytosolic factor 1 (oxidative burst)

Franke U: Hum Mol Genet, 1999