Williams Syndrome

Genetics

-Contiguous gene deletion syndrome, ELN in the critical region, 1.5Mb, ~28 genes, 7q11.23

-AD, majority of cases de novo

Clinical findings/Dysmorphic features

-Cardiovascular disease (elastin arteriopathy, peripheral pulmonary stenosis, supravalvar aortic stenosis, hypertension)

-Connective tissue abnormalities (hoarse voice, hernia, rectal prolapse, joint limitation or laxity)

-ID (usually mild) and unique personality characteristics

-Growth abnormalities; endocrine abnormalities (hypercalcemia, hypercalciuria, hypothyroidism, early puberty)

-Facial features: broad brow, bitemporal narrowness, periorbital fullness, lacy iris pattern, strabismus, short nose, full nasal tip, malar hypoplasia, long philtrum, full lips, wide mouth, small jaw, and prominent earlobes

Etiology

-Prevalence of 1:7500

Pathogenesis

-ELN deletion causes the CV and CT problems, LIMK1 has been linked to the visuospatial construction cognitive deficit

Genetic testing/diagnosis

-Detection of recurrent 7q11.23 contiguous gene deletion of the Williams-Beuren syndrome critical region (WBSCR) --> encompasses ELN

-Can be detected using FISH or In/Del or microarray (~99%)

Others

-Overfriendliness, empathy, generalized anxiety, specific phobias, attention deficit disorder