Williams Syndrome

Williams syndrome is a genetic condition caused by a microdeletion mutation on chromosome 7 which results in elfin facies, extreme friendliness with strangers, as well as concurrent medical problems. The long arm of chromosome 7 contains the elastin gene which is usually lost in the disease. Along with the elfin facies and friendliness with strangers, people with Williams syndrome also have mental retardation, cardiovascular problems, and hypercalcemia, resulting from increased sensitivity to vitamin D.



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Pathophysiology

Microdeletion Of Long Arm Of Chromosome 7
Long Arm pulling Chromosome (7) Slot-machine
Williams syndrome is caused by a microdeletion on the long arm of chromosome 7.

Elastin Gene
Elastic-pants
The elastin gene is on the long arm of chromosome 7, often where the microdeletion occurs.

Signs and Symptoms

Elfin Facies
Elf-costume
Elfin facies is a term commonly used to describe the appearance of people with Williams syndrome. Individuals with Williams syndrome tend to have widely spaced teeth, a long philtrum, and flattened nasal bridge.

Intellectual Disability
Tar Covered Book
Intellectual Disability (formely Mental retardation) is present in most cases of Williams syndrome.

Extreme Friendliness with Strangers
Talking to Strangers
Individuals with Williams syndrome typically display a hallmark trait of lack of social inhibition leading to extreme friendliness with strangers. Infants with Williams syndrome will often hug complete strangers and the level of friendliness observed is often inappropriate for the social setting.

Well Developed Verbal Skills
Talking
Despite physical and cognitive deficits, individuals with Williams syndrome exhibit well developed social and verbal abilities.

Cardiovascular Problems
Heart
Individuals with Williams syndrome experience many cardiac problems including heart murmurs or narrowing of major blood vessels.

Hypercalcemia
Hiker-calcified-cow
Hypercalcemia results from a hypersensitivity to vitamin D and can frequently be seen in patients with Williams. This can result in seizures and muscle rigidity.

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Increased Sensitivity To Vitamin D

Viking (D) Daisy

Patients with Williams syndrome can have increased sensitivity to vitamin D leading to hypercalcemia.

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