

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.



PLAY PICMONIC

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 (formerly 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.

Increased Sensitivity To Vitamin D

Viking (D) Daisy

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