



Williams Syndrome

What is Williams syndrome?

Williams syndrome is a developmental disorder that affects many parts of the body. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems. Williams syndrome affects an estimated 1 in 7,500 to 10,000 people.

What are the symptoms of Williams syndrome?

Common symptoms of the condition include:

- specific facial features like a wide mouth, small upturned nose, widely spaced teeth, and full lips
- colic or feeding problems
- attention deficit hyperactivity disorder (ADHD)
- learning disorders
- inward bend of pinky finger
- specific phobias
- short stature
- speech delays
- sunken chest
- varying degrees of intellectual disability
- low birth weight and muscle tone
- kidney abnormalities
- farsightedness

Personality traits common in people who have the condition include:

- an interest in music
- aversion to physical contact
- being overly friendly
- sensitivity to loud noises

How is Williams syndrome diagnosed?

The condition can typically be diagnosed by physical characteristics. The diagnosis can be confirmed by doing a test called fluorescent in situ hybridization (FISH). This is a lab test that labels DNA sequences with a chemical that lights up under ultraviolet light. This enables scientists to see if the gene is missing or not.

Other tests to help diagnose the condition can include a kidney ultrasound, blood pressure check, and an echocardiogram. Tests can also show high blood calcium levels, high blood pressure, slack joints, and unusual patterning in the iris of the eye.

Treatment options for Williams syndrome

There is no cure for Williams syndrome. Treatment involves easing the symptoms connected to the syndrome. Narrowed blood vessels can be treated if they cause symptoms. Physical therapy and speech therapy can also be beneficial.

Treatment is usually based on the individual's symptoms. There is no standard treatment protocol. Regular checkups are necessary to look at the cardiovascular system and track any possible problems.

People with Williams syndrome should avoid taking extra calcium and vitamin D. This is because blood levels of these substances are already high.

Long-term complications of Williams syndrome

Medical conditions can affect the lifespan of those with Williams syndrome. Calcium deposits can cause kidney problems, and narrowed blood vessels can cause heart failure. The majority of affected people have some degree of intellectual disability. Learning self-help skills and getting early intervention in school can help encourage independence. Some people with Williams syndrome will need to live with a caregiver or in a supervised home.

Resources

- **Health Line** - www.healthline.com/health/williams-syndrome#overview1
- **Williams Syndrome Association** - <https://williams-syndrome.org>
- **Williams syndrome - Genetics Home Reference** - <https://ghr.nlm.nih.gov/condition/williams-syndrome>