



Williams Syndrome

WHAT IS WILLIAMS SYNDROME?

Williams syndrome is a rare disorder that affects development and different parts of the body including blood vessels, heart, and joints. It is characterized by intellectual disability and distinct facial features. About 1 in 7,500 to 18,000 people are affected by Williams syndrome.

WHAT CAUSES WILLIAMS SYNDROME?

Williams Syndrome is caused by the deletion of genetic material from a specific region of chromosome 7, which affects a gene responsible for producing a protein called elastin. Elastin is very important for the strength and flexibility of blood vessels.

HOW IS WILLIAMS SYNDROME DIAGNOSED?

There is no routine screening for Williams syndrome in newborns. Diagnosis is usually based on the presence of specific facial features and the other common medical problems seen with the condition. A special genetic test called fluorescein-in-situ hybridization can be used to confirm the diagnosis.

WHAT ARE THE FEATURES OF WILLIAMS SYNDROME?

There are many characteristic features associated with Williams syndrome:

- Facial features include: puffy eyes, a short nose, wide mouth, full lips, full cheeks, and a small chin (see Figure 1).
- Skeletal features include: short stature, sloping shoulders, long neck, and limited joint movement.
- Other development features include: strong verbal abilities, highly social personalities, and a love for music.



Fig. 1: Facial features of Williams Syndrome include puffiness around the eyes, short nose, wide mouth, full lips, full cheeks, and a small chin.

WHAT OTHER MEDICAL ISSUES ARE LINKED WITH WILLIAMS SYNDROME?

People with Williams Syndrome can have narrowing of large arteries in the body. Specifically, this condition is linked with a problem called Supravalvular Aortic Stenosis (SVAS) which involves narrowing of a blood vessel from the heart. This problem can lead to heart failure if not treated. Other issues in the body may include low birth weight, difficulties in weight gain, digestive and urinary tract problems and higher risks with anesthesia.

WHAT EYE PROBLEMS CAN OCCUR WITH WILLIAMS SYNDROME?

Children with Williams Syndrome may have eye conditions such as [strabismus](#) (specifically [esotropia](#) or eye crossing), [amblyopia](#) ([lazy eye](#)), and other specific eye movement problems. They may also have an abnormal development of the retina (the inner layer of the back of the eye).

WHAT DEVELOPMENTAL ISSUES CAN BE SEEN IN CHILDREN WITH WILLIAMS SYNDROME?

Children with Williams Syndrome often have mild to moderate developmental delays, which can lead to learning difficulties and lower IQ. They tend to be very interested in other people, have outgoing personalities, and may have other problems like attention deficit disorder, phobias, and anxiety.



WHAT RESEARCH IS BEING DONE FOR WILLIAMS SYNDROME?

Currently, there is no cure for Williams syndrome. However, the National Institute of Health and the National Institute of Neurological Disorders and Stroke are conducting research to better understand the causes and look for treatments for Williams syndrome.

WHERE CAN I FIND MORE INFORMATION ABOUT WILLIAMS SYNDROME?

- Further information can be found at williams-syndrome.org.

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