

Williams Syndrome

WHAT ARE THE CHARACTERISTICS OF WILLIAMS SYNDROME?

The characteristic facial features include puffiness around the eyes, short nose, wide mouth, full lips, full cheeks and a small chin. They can have a short stature, sloping shoulders, long neck and limited movement in their joints. They also have strong verbal abilities, highly social personalities, and a love for music.

WHAT CAUSES WILLIAMS SYNDROME?

Williams Syndrome is a rare disorder caused by a deletion of genetic material from a specific region of chromosome 7 that affects an elastin gene. Elastin is a protein that gives blood vessels strength and the ability to stretch.

HOW IS WILLIAMS SYNDROME DIAGNOSED?

Williams Syndrome is usually diagnosed with the characteristic facial features and the other common medical problems associated with it. A special genetic test called fluorescein-in-situ hybridization can be used to confirm the diagnosis.

WHAT MEDICAL ISSUES ARE ASSOCIATED WITH WILLIAMS SYNDROME?

A cardiovascular problem called Supravalvular Aortic Stenosis (SVAS) can be associated with Williams Syndrome. SVAS is the narrowing of the large blood vessel which carries blood from the heart to the rest of the body. This problem could cause shortness of breath, chest pain and ultimately heart failure if not treated. Decreased birth weight and failure to gain weight normally is also common. There can also be digestive and urinary tract difficulties such as constipation. They may also be at increased risk when given general anesthesia.

WHAT SORT OF EYE PROBLEMS COULD A CHILD HAVE WITH WILLIAMS SYNDROME?

Certain ocular findings such as [strabismus](#) and [amblyopia](#) can be seen in children with Williams Syndrome. The types of strabismus would include infantile [esotropia](#), dissociated vertical deviation, and oblique muscle dysfunction.

WHAT DEVELOPMENTAL ISSUES COULD BE SEEN IN A CHILD WITH WILLIAMS SYNDROME?

Mild to moderate developmental delays are seen in Williams Syndrome which could cause learning difficulties. Individuals with Williams Syndrome tend to have an extreme interest in other people with their outgoing personalities. Attention Deficit Disorder, problems with phobias, and anxiety are commonly associated.

WHAT RESEARCH IS BEING DONE FOR CHILDREN WITH WILLIAMS SYNDROME?

There is no cure for Williams Syndrome. Research is being done by the National Institute of Health and the National Institute of Neurological Disorders and Stroke to understand more about the genetic and neurobiological origins of Williams Syndrome.



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

WHERE CAN I GET MORE INFORMATION ABOUT WILLIAMS SYNDROME?

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

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