Williams Syndrome

WHAT CAUSES WILLIAMS SYNDROME?

Williams Syndrome is 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. It is a rare disorder and is also called Elfin Facies Syndrome.

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 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.

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.

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 dysfunction.

WHAT DEVELOPMENTAL ISSUES WOULD AFFECT A CHILD WITH WILLIAMS SYNDROME?

Mild to moderate developmental delays are seen in Williams Syndrome which would 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.

Updated 3/2015