Williams Syndrome

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Definition

What is Williams Syndrome? Williams Syndrome (WS) is a genetic disorder that is present at birth and affects males and females equally. It has been identified in individuals in countries all over the world. Williams Syndrome occurs in approximately l/20,000 births. Williams Syndrome occurs spontaneously, not as the result of an inherited characteristic from either parent. He syndrome causes medical and developmental problems. Many people with WS remain undiagnosed or are diagnosed at a relatively late age.

Introduction

Chromosomal studies of individuals with WS show a deletion of elastin on chromosome #7. Elastin is an important part of elastic fibers found in the connective tissue of many organs. The missing elastic explains some of the characteristics of WS, such as certain facial features, hoarse voice, bladder and bowel diverticula, heart problems, and joint problems. Other characters, such as high calcium levels, M.R. and unique personality traits, remain unexplained.

Facial features include a small, up-turned node, long philtrum (upper lip length), wide mouth, full lips, small chin, and puffiness around the eyes. Blue or green-eyed children might have a "starburst" or white, lacey pattern on their iris. Facial features are more prominent as the person ages.

Diagnosis

Genetic clinics around the country can order the FISH test for the elastin deletion. The test can be ordered by any primary care provider and be sent to a reliable cytogenetics lab. The order should read: FISH test for the deletion of elastin on chromosome #7 to confirm or rule out Williams Syndrome.

Prevention and Treatment

Heart and blood vessel problems: the majority of individuals with WS have some type of heart or blood vessel problem. Sometimes there is narrowing in the aorta or in the pulmonary arteries. At times, the narrowing is so severe that surgical correction is needed. There is an increased risk for blood vessel narrowing and for high blood pressure as people with WS age. Regular evaluation by a cardiologist and frequent blood pressure checks are necessary.

High calcium levels: Some young children with WS will have high blood calcium levels. The cause is unknown but this can cause extreme irritability or "colic" type symptoms. Too much calcium can also cause kidney problems. Sometimes nutrition and medical treatment is needed. Many times, the high calcium levels resolve during childhood, but lifelong problems in calcium levels or vitamin D metabolism may occur and should be monitored. All pediatric vitamin preparations contain vitamin D so it is recommended that individuals with WS DO NOT receive multi-vitamin supplementation.

Eye and Ear problems: Some individual with WS may have strabismus (cross eyes, lazy eye, or squint). This may be subtle. Some have farsightedness. An ophthalmologist (special eye doctor) should do regular checkups. Frequent ear infections are more common in people with WS. An ENT specialist can treat these people and make sure their hearing is okay. Children with WS can actually have very sensitive hearing. Loud noises can be painful or startling. This often improves with age.

Dental concerns: People with WS should have routine dental care. Malocclusion (problems with bite), absent teeth, and malformed teeth are common. Many individuals require orthodontia.

Digestive system: Gastroesphageal reflux disease (GERD) is common in people with WS. Tests to determine reflux need to be done, particularly is people are refusing to eat or are vomiting. Chronic constipation is common in people with WS. Dietary intervention, including fiber and fluids, can help tremendously.

Diverticulitis can occur, due to chronic constipation. People with a lot of recurring stomach pain should be checked for this.

Musculoskeletal problems: Babies and young children often have low muscle tone and joint laxity. The low tone is why infants sometimes have trouble feeding. Joint stiffness or contractures may develop and worsen as people age. Physical therapy is very helpful to improve muscle tone, strength, and range of motion.

Growth and development: Many children with WS have lower birth weights than their brothers or sisters. They typically gain weight slowly. Adult stature is usually smaller than average. Most people with WS have some degree of mental retardation. Young children may reach developmental milestones, such as walking or toilet training, at a later age than is considered typical. Individuals may be quite distractable and Attention Deficit/Hyperactivity Disorder is often diagnosed during the school years. Individuals with WS seem to be very good with language and auditory memory, but have difficulty with visual, spatial or fine motor skills. An interesting fact is that many individuals with WS have been found to have great music abilities.

PERSONALITY:

People with WS are wonderfully social. They have great, expressive language skills and are very polite. They are typically unafraid of strangers and children prefer contact with adults rather than peers. This puts individuals with WS at risk for exploitation. Safety issues surrounding their overly friendly personality need to be considered.

Emergency Situations – What can go wrong?

There have been unexplained, sudden deaths and anesthesia complications in individuals with WS receiving general anesthesia. Pediatric anesthesia monitoring is recommended for children. Cardiac status needs to be closely reviewed prior to surgery.

Conclusion

WS can affect many different body organs. No two people with WS have, however, the same problems. Since many of the health problems develop over time, ongoing medical monitoring and supervision is important. Most people with WS manage to live active, full lives. Most people are able to master self-helps skills and complete academic and/or vocational school. Many adults with WS are successfully employed in supervised or independent jobs.

Information for this article was obtained from the American Academy of Pediatrics, committee on Genetics: Health Care Supervisor for Children with Williams Syndrome and from the above web-sites.

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