

WHO CAN HELP THE FAMILIES?

The Williams Syndrome Association (WSA) is devoted exclusively to improving the lives of individuals with Williams syndrome. The Association strives to locate individuals with the syndrome and their families and disseminate timely and accurate medical and educational information. It provides members with support through yearly regional conferences and social gatherings, quarterly newsletters and biennial conventions. The WSA actively supports research into educational, behavioral, social and medical aspects of the syndrome.

HOW CAN I JOIN?

There are a variety of membership options in the WSA. Simply complete the form below and return it to the National office, or call us at (248) 244-2229 to receive membership information and a packet of materials on Williams syndrome.

Name _____
Address _____
City _____ State _____ Zip _____
Phone _____
Email _____

Individual with WS/Relationship:
_____/_____

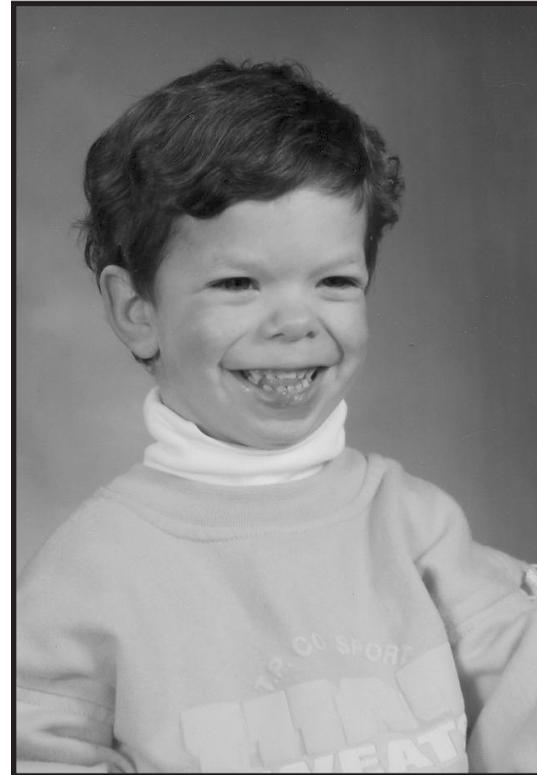
Date of Birth _____



williamssyndrome
ASSOCIATION

NATIONAL OFFICE
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Troy, MI 48084
800.806.1871

FACTS ABOUT



WILLIAMS SYNDROME

Williams syndrome is a genetic condition which occurs in 1 in 10,000 births. It causes medical and developmental problems.

WHAT IS WILLIAMS SYNDROME?

Williams syndrome is a genetic disorder that was first recognized as a distinct entity in 1961. It is present at birth, and affects males and females equally. It can occur in all ethnic groups and has been identified in countries throughout the world.

WHAT ARE THE COMMON FEATURES OF WILLIAMS SYNDROME?

Characteristic facial appearance - Most young children with Williams syndrome are described as having similar facial features. These features, which tend to be recognized only by a trained geneticist or birth defects specialist, include a small upturned nose, long philtrum (upper lip length), wide mouth, full lips, small chin, and puffiness around the eyes. Blue and green-eyed children with Williams syndrome can have a prominent "starburst" or white lacy pattern on their iris. Facial features become more apparent with age.



Heart and blood vessel problems - The majority of individuals with Williams syndrome have some type of heart or blood vessel problem. Typically, there is narrowing in the aorta (producing supravalvular aortic stenosis [SVAS]), or narrowing in the pulmonary arteries. There is a broad range in the degree of narrowing, ranging from trivial to severe (requiring surgical correction of the defect). Since there is an increased risk for development of blood vessel narrowing or high blood pressure over time, periodic monitoring of cardiac status is necessary.

Hypercalcemia (elevated blood calcium levels) - Some young children with Williams syndrome have elevations in their blood calcium level. The true frequency and cause of this problem is unknown. When hypercalcemia is present, it can cause extreme irritability or "colic-like" symptoms. Occasionally, dietary or medical treatment is needed. In most cases, the problem resolves on its own during childhood, but a lifelong abnormality in calcium or Vitamin D metabolism may exist so blood levels should be periodically monitored.

Low birth weight/Slow weight gain

- Most children with Williams syndrome have a slightly lower birth-weight than their brothers and sisters. Slow weight gain, especially during the first several years of life, is also a common problem and many children are diagnosed as "failure to thrive". Adult stature is often smaller than average.

Feeding problems - Many infants and young children have feeding problems. These problems have been linked to low muscle tone, severe gag reflex, poor suck/swallow, tactile defensiveness etc. Feeding difficulties tend to resolve as the children get older, although GE Reflux (heartburn) appears to be common and can occur at any age.

Irritability (colic) during infancy - Many infants with Williams syndrome have an extended period of colic or irritability. This typically lasts from 4 to 10 months of age, then resolves. It is sometimes attributed to hypercalcemia, but in most cases the cause is not identified. Abnormal sleep patterns with delayed acquisition of sleeping through the night may be associated with the colic.

Dental abnormalities - Slightly small, widely spaced teeth are common in children with Williams syndrome. They may also have a variety of abnormalities of occlusion (bite), tooth shape or appearance. Most of these dental changes are readily amendable to orthodontic correction.

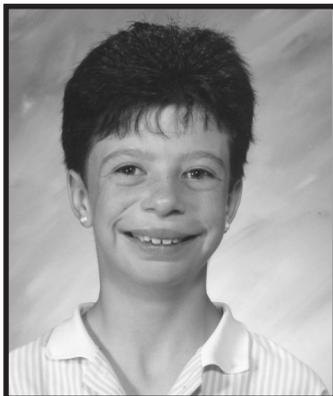
Kidney abnormalities - There is a slightly increased frequency of problems with kidney structure and/or function.

Hernias - Inguinal (groin) and umbilical hernias are more common in Williams syndrome than in the general population.

Hyperacusis (sensitive hearing) - Children with Williams syndrome often have more sensitive hearing than other children. Certain frequencies or noise levels can be painful and/or startling to the individual. This condition often improves with age.

Musculoskeletal problems - Young children with Williams syndrome often have low muscle tone and joint laxity. As the children get older, joint stiffness (contractures) and spinal curvature may develop. Physical therapy is very helpful in improving muscle tone, strength and joint range of motion.

Overly friendly (excessively social) personality - Individuals with Williams syndrome have a very endearing personality. They have a unique strength in their expressive language skills, and are extremely polite. They are typically unafraid of strangers and show a greater interest in contact with adults than with their peers.



Developmental delay, learning disabilities and attention deficit - Most people with Williams syndrome have some degree of intellectual handicap. Young children with Williams syndrome often experience developmental delays; milestones such as walking, talking and toilet training are often achieved somewhat later than is considered normal. Distractibility (short attention span) is a common problem in mid-childhood, which appears to get better as the children get older.

Older children and adults with Williams syndrome often demonstrate intellectual "strengths and weaknesses." There are some intellectual areas (such as speech, long term memory, and social skills) in which performance is relatively strong, while other intellectual areas (such as fine motor and spatial relations) are significantly deficient.



WHAT IS THE CAUSE OF WILLIAMS SYNDROME?

Williams syndrome is not caused by anything the parents did or did not do either before or during pregnancy. Individuals with Williams syndrome are missing ~20 genes on chromosome #7 including the gene that makes the protein elastin (a protein which provides strength and elasticity to vessel walls). It is likely that the elastin gene deletion accounts for many of the physical features of Williams syndrome. Some medical and developmen-

tal problems are probably caused by deletion of additional genetic material neighboring the elastin gene on chromosome #7. In most families the child with Williams syndrome is the only one to have the condition in his or her entire extended family. However, the individual with Williams syndrome has a 50% chance of passing the disorder on to each of his or her children.



HOW IS WILLIAMS SYNDROME DIAGNOSED?

Many individuals with Williams syndrome remain undiagnosed or are diagnosed at a relatively late age. This is of concern since individuals with Williams syndrome can have significant and possibly progressive medical problems. When the characteristics of Williams syndrome are recognized, referral to a clinical geneticist for further diagnostic evaluation is appropriate. The clinical diagnosis can be confirmed by a blood test. The technique known as fluorescent in situ hybridization (FISH), a diagnostic test of the DNA detects the elastin deletion on chromosome #7 in 99% of individuals with Williams syndrome. Recurrence risk is usually low, but Genetic counseling is advisable.

ARE MEDICAL PROBLEMS FREQUENT IN WILLIAMS SYNDROME?

Williams syndrome can affect many different body organs. However, it is important to remember that no two individuals with Williams syndrome have exactly the same problems. Since some of the medical problems can develop over time, it is

important that individuals with Williams syndrome receive ongoing medical monitoring and supervision. Despite the possibility of medical problems, most children and adults with Williams syndrome are healthy and lead active, full lives.

WHAT IS THE OUTLOOK FOR ADULTS WITH WILLIAMS SYNDROME?

The vast majority of adults with Williams syndrome master self-help skills and complete academic and/or vocational school. They are employed in a variety of settings (ranging from supervised to independent jobs). Many adults with Williams syndrome live with their parents; others live in supervised apartments and some are able to live on their own.



WHO SHOULD CARE FOR INDIVIDUALS WITH WILLIAMS SYNDROME?

Given the complex nature of many of the problems found in individuals with Williams syndrome, many health and educational professionals should be involved in their care. Regular monitoring for potential medical problems is necessary and should be done by a physician familiar with the broad array of problems that can be seen in Williams syndrome.

Due to the intellectual "strengths and weaknesses," the expertise of developmental psychologists, speech and language pathologists, physical and occupational therapists, etc. who are familiar with Williams syndrome is also recommended. Multi-disciplinary Williams syndrome teams, with professionals available in all of these areas, can be an effective adjunct to local resources. When a Williams syndrome clinic is not close by, it is necessary for the family to seek out professionals in their communities to provide this crucial input.