FACT SHEET

Williams Syndrome

What is Williams Syndrome?

Williams Syndrome is a rare, congenital (present at birth) disorder characterized by physical and developmental problems including an impulsive and outgoing (excessively social) personality, limited spatial skills and motor control, and intellectual disability (i.e., developmental delay, learning disabilities, and attention deficit disorders). Other features include characteristic "elfin-like" facial features, heart and blood vessel problems, hypercalcemia (elevated blood calcium levels), low birth weight, slow weight gain, feeding problems, irritability during infancy, dental and kidney abnormalities, hyperacusis (sensitive hearing), and musculoskeletal problems. Symptoms vary among individuals.

Although individuals with Williams Syndrome may show competence in areas such as language, music, and interpersonal relations, their IQ's are usually below average, and they are considered to have a mild to moderate intellectual disability. Scientists have learned that most individuals with Williams Syndrome have a deletion of genetic material on Chromosome 7. This probably causes the physical and developmental problems experienced by these individuals.

How is it manifested?

- characteristic facial appearance
- heart and blood vessel problems
- hypercalcemia (elevated blood calcium levels)
- low birth-weight / low weight gain
- feeding problems
- irritability (colic during infancy)
- dental abnormalities
- kidney abnormalities
- hernias
- hyperacusis (sensitive hearing)
- musculoskeletal problems
- overly friendly (excessively social) personality
- developmental delay, learning disabilities and attention deficit

Who is affected?

It is estimated to occur in 1 in 20,000 births.

How is it diagnosed or detected?

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 Hybridisation (FISH), a diagnostic test of the DNA, detects the elastin deletion on Chromosome 7 in 95% to 98% of individuals with Williams Syndrome.

Additional Resources:

WILLIAMS SYNDROME ASSOCIATION (WSA) – www.williams-syndrome.org
The WSA is the only group in the United States devoted exclusively to improving the lives of individuals with Williams syndrome and their families. The WSA supports research into all facets of the syndrome, and the development of the most up to date educational materials regarding Williams syndrome.

CANADIAN ASSOCIATION FOR WILLIAMS SYNDROME (CAWS) – www.caws-can.org

The Canadian Association of Williams Syndrome (CAWS) was founded by a group of parents in 1984. CAWS is a national federation that provide support to Williams Syndrome individuals and their families.

BOOKS:

Williams Syndrome: Approaches to Intervention
by Eleanor Semel, Sue R. Rosner

Journey from Cognition to Brain to Gene:
Perspectives from Williams Syndrome
by Ursula Bellugi, Marie I. St. George