

Down syndrome

What is Down syndrome?

Down syndrome is a common genetic variation that usually causes delay in physical, intellectual and language development. A child with Down syndrome is at higher than normal risk for many medical conditions, most notably congenital heart disease (often corrected by surgery), hearing and vision deficits, leukemia and Alzheimer's disease. There is wide variation in mental abilities, behavior and physical development in individuals with Down syndrome. Each individual has his/her own unique personality, capabilities and talents. Individuals with Down syndrome benefit from loving homes, early intervention, inclusive education, appropriate medical care and positive public attitudes. In adulthood, many people with Down syndrome have jobs, live independently and enjoy recreational opportunities in their communities.

What is the cause?

Down syndrome is caused by an error in cell division, which results in an extra 21st chromosome. A child with Down syndrome has 47 chromosomes instead of the usual 46. A sperm or egg with an abnormal number (two chromosomes of 21) merges with a normal mate (who has 1 chromosome 21). The exact causes of the chromosomal rearrangement and primary prevention of Down syndrome are currently unknown.

What are some signs?

There are many clinical signs of Down syndrome, but it is rare to find all or most in one person. Signs may include poor muscle tone (hypotonia), slanting eyes with folds of skin at the inner corners (called epicanthal folds), hyperflexibility (excessive ability to extend the joints), short broad hands with a single crease across the palm on one or both hands, small head size, flat nasal bridge, small mouth and ears, broad feet with short toes, short neck, small oral cavity and/or short, high-pitched cries in infancy. Besides having a distinct physical appearance, children with Down syndrome frequently have specific health-related problems. A lowered resistance to infection makes these children more prone to respiratory problems. Some also may have a condition known as Atlantoaxial Instability, a misalignment of the top two vertebrae of the neck which makes these individuals more prone to injury if they participate in certain activities which overextend or flex the neck. Parents need to have their child examined by a physician to determine this condition, proper diagnosis can help prevent serious injury.

Online Resources

Siskin Children's Institute • www.siskin.org

Down Syndrome Association of Middle Tennessee • www.dsamt.org • 615-386-9002

Kids Quest • www.cdc.gov/ncbddd/kids/default.htm

National Association for Down Syndrome • www.nads.org • 630-325-9112

National Down Syndrome Congress • www.ndscenter.org • 1-800-232-6372

National Down Syndrome Society • www.ndss.org • 1-800-221-4602

Tennessee Disability Pathfinder • www.familypathfinder.org