

Down Syndrome

Definition:

Down syndrome is a congenital disorder, caused by the presence of an extra 21st chromosome, in which the affected person has mild to moderate intellectual disability, short stature, and a flattened facial profile.

Description:

Down syndrome is a genetic condition that causes delays in physical and intellectual development. It occurs in approximately one in every 800 live births. Individuals with Down syndrome have an extra copy of the 21st chromosome that affect some or all cells. It is the most frequently occurring chromosomal disorder. Down syndrome is not related to race, nationality, religion or socioeconomic status.

Characteristics:

Common Physical Characteristics:

Down syndrome symptoms vary from person to person and can range from mild to severe. However, children with Down syndrome have a widely recognized appearance.

The head may be smaller than normal and abnormally shaped. For example, the head may be round with a flat area on the back. The inner corner of the eyes may be rounded instead of pointed.

Common physical signs include:

- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Separated joints between the bones of the skull (sutures)
- Single crease in the palm of the hand
- Small ears
- Small mouth
- Upward slanting eyes
- Wide, short hands with short fingers
- White spots on the colored part of the eye (Brushfield spots)

Physical development is often slower than normal. Children with Down syndrome may never reach an average adult height.

Children may also have delayed mental and social development. Common problems may include:

- Impulsive behavior
- Poor judgment
- Short attention span
- Slow learning

As children with Down syndrome grow and become aware of their limitations, they may also feel frustration and anger.

Many different medical conditions are seen in people with Down syndrome, including:

- Birth defects involving the heart, such as an [atrial septal defect](#) or [ventricular septal defect](#)
- Dementia may be seen
- Eye problems, such as cataracts (most children with Down syndrome need glasses)
- Early and massive vomiting, which may be a sign of a gastrointestinal blockage, such as [esophageal atresia](#) and [duodenal atresia](#)
- Hearing problems, probably caused by regular ear infections
- Hip problems and risk of dislocation
- Long-term (chronic) constipation problems
- [Sleep apnea](#) (because the mouth, throat, and airway are narrowed in children with Down syndrome)
- Teeth that appear later than normal and in a location that may cause problems with chewing
- Underactive thyroid ([hypothyroidism](#))

Children with Down syndrome have a higher incidence of infection, respiratory, vision and hearing problems as well as thyroid and other medical conditions. However, with appropriate medical care most children and adults with Down syndrome can lead healthy lives. The average life expectancy of individuals with Down syndrome is 55 years, with many living into their sixties and seventies.

Learning and Development:

It is important to remember that while children and adults with Down syndrome experience developmental delays, they also have many talents and gifts and should be given the opportunity and encouragement to develop them.

Most children with Down syndrome have mild to moderate cognitive impairments, but it is important to note that they are more like other children than they are different. Early Intervention services should be provided shortly after birth. These services should include physical, speech and developmental therapies. Most children attend their neighborhood schools, some in regular classes and others in special education classes. Some children have more significant needs and require a more specialized program.

Some high school graduates with Down syndrome participate in post-secondary education. Many adults with Down syndrome are capable of working in the community, but some require a more structured environment.

Implications for speech and language:

Although individuals with Down Syndrome tend to be good communicators from a young age and usually have strengths in non-verbal communication skills such as gesture and sign, speech and language skills are an area of particular difficulty. Spoken language skills are usually delayed relative to the children's non-verbal ability and this suggests a profile of specific language impairment. This should be a focus for targeted and intensive intervention throughout infancy and childhood, as it is in other language impaired children who show a gap between verbal and non-verbal abilities.

Causes:

Down syndrome is usually caused by an error in cell division called non-disjunction. It is not known why this occurs. However, it is known that the error occurs at conception and is not related to anything the mother did during pregnancy. Statistically the incidence of Down syndrome increases with advancing maternal age. However, 80% of children with Down syndrome are born to women under 35 years of age.

Diagnosing this disorder:

Speech-Language pathologists cannot diagnose this condition. Down syndrome is usually identified at birth or shortly thereafter. Initially the diagnosis is based on physical characteristics that are commonly seen in babies with Down syndrome. These include low muscle tone, a single crease across the palm of the hand, a slightly flattened facial profile and an upward slant to the eyes. The diagnosis must be confirmed by a chromosome study known as a karyotype.

Two types of procedures are available to pregnant women: screening tests and diagnostic tests. The screening tests estimate the risk of the baby having Down syndrome. Diagnostic tests tell whether or not the baby actually has Down syndrome.

Screening Tests

- At this time the most commonly used screening test is “The Triple Screen.” This is a combination of three tests that measure quantities of various substances in the blood. These tests are usually done between 15 and 20 weeks of gestation.
- Sonograms (ultrasounds) are usually performed in conjunction with other screenings. These can show some physical traits that are helpful in calculating the risk of Down syndrome.
- Screening tests do not accurately confirm the diagnosis of Down syndrome. In fact, false positives and false negatives frequently occur.

Diagnostic Tests

Three diagnostic tests are currently available:

- Amniocentesis which is performed between 12 and 20 weeks gestation
- Chorionic Villus Sampling (CVS) which is conducted between 8 and 12 weeks gestation
- Percutaneous Umbilical Blood Sampling (PUBS) which is performed after 20 weeks gestation

Resources:

Books for Parents:

- A Parent’s Guide to Down Syndrome, Toward a Brighter Future, by Siegfried M. Pueschel, M.D., Ph.D., M.P.H.
- Common Threads, Celebrating Life with Down Syndrome, by Cynthia S. Kidder and Brian Skotko
- Angel Behind the Rocking Chair, by Pam Vredevelt
- Choosing Naia, by Mitchell Zuckoff
- Expecting Adam: A True Story about Birth, Rebirth and Everyday Magic, by Martha Beck
- Angel Unaware, by Dale Evans
- Gifts of Love (25 minute video)
- A Promising Future Together – A guide for new parents of children with Down Syndrome, NDSS (video)
- Down Syndrome – The First 18 Months, by Will Schermerhorn (video)
- Precious Lives Painful Choices – a Prenatal Decision-making Guide, by Sherokee Ilse
- Count Us In, by Jason Kingsley and Mitchell Levitz.
- The Memory Keeper’s Daughter, by Kim Edwards
- Your Baby Has Down Syndrome (video); Down Syndrome Center, Children’s Hospital of Pittsburgh. Phone: 412-692-7963, Fax: 412-692-7428, Email: cannons@chplink.chp.edu
- Bebes con Síndrome de Down – una guía para padres, por Karen Stray-Gunderson (en español)

Support Groups:

1. DSACT- Down Syndrome Association of Central Texas:

DSACT offers year-round programs for children with Down syndrome and their families. Programs include social and recreational classes for children such as sign language, swimming, cooking, yoga and martial arts. Young adults may attend a specialized class at UT. Social groups for children ages 0-5, 6-12 and teenagers meet monthly.

The Learning Program offers a research-based curriculum improving early literacy and math skills for students with Down syndrome ages 2-11. Parents attend class simultaneously learning best practices to teach their child at home. For more information on the Learning Program contact our office.

Emotional support workshops are held throughout the year offering parents a safe place to discuss their experiences and feelings. Professionals provide strategies and methods to achieve and maintain emotional health. For more information about emotional support workshops contact our office.

2. Comite Latino

Parent group which meets every two months at someone's home. Provides food, social time, will look for information parents are in need of, provide access to resources, usually present on a topic related to Down Syndrome. Contacts are: Deborah Trejo 586-2552 or Rebecca Tobias 447-6974

Comite Latino

El Comite Latino de DSACT se dedica a servir a la comunidad hispanohablante en el area. Se espera incluir a toda la comunidad hispanohablante con hijos o familiares con el SD para que tenga el mismo acceso a informacion, servicios y recursos. Tenemos pláticas sobre temas importantes y nos ayudaremos uno al otro. Comuniquese con Rebecca Tobias (512) 447-6974 o Deborah Trejo (512) 586-2552 o quintosol@sbcglobal.net

Websites:

In English/Spanish:

- Down Syndrome Association of Central Texas (DSACT En Español) <http://www.dsact.com/DSACTspanish.html>
- National Down Syndrome Congress (Congreso Nacional de Síndrome de Down) <http://www.ndsccenter.org/>
- National Down Syndrome Society (Sociedad Nacional de Síndrome de Down) <http://www.ndss.org>
- National Association for Down Syndrome (Asociación Nacional para el Síndrome de Down) http://www.nads.org/pages_new/new_parents/support_spanish.html
- Light at the End of the Tunnel (Luz al Final del Túnel) <http://www.dsaoc.org/espanol/tunnelSpn.pdf>
- Welcome to Holland, by Emily Perl Kingsley (Bienvenido a Holanda, por Emily Perl Kingsley) <http://www.ensenemasamemas.org/SpecialNeedsDetails.asp?id=32#55>

In English:

- "A Future and A Hope" by Carlene Mattson <http://www.dsaoc.org/./programs/heart.asp>
- Band of Angels inspirational materials <http://www.bandofangelsfoundation.com>
- Mother Stories from Chicago, Illinois http://www.gigisplayhouse.com/mother_stories.asp
- Gifts: How Children with Down Syndrome have Enriched our Lives <http://giftsds.segullah.org/>
- Clinical research related to cognitive treatments <http://dsresearch.stanford.edu>

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Down Syndrome Facts. Down Syndrome Association of Central Texas. Retrieved from: <http://dsact.com/about/down-syndrome-facts/>