

Introduction to Down Syndrome

Written and Reviewed by the My Child Without Limits Advisory Committee

What Is Down Syndrome?

Down syndrome is the most common cause of mild to moderate mental retardation and the mental and physical medical problems that often come with it. It was named after John Langdon Down, the first physician to identify the syndrome.

Who Does Down Syndrome Affect?

Down syndrome occurs in one out of 800 live births. It affects all races and economic groups.

What Causes Down Syndrome?

A fertilized egg normally has 23 pairs of chromosomes. However, most of the people who have Down syndrome have an extra (or third) copy of chromosome 21. Having this extra copy of chromosome 21 changes the body's and brain's normal development.

In most cases Down syndrome is caused by a random error in cell division that happens during formation of the mother's egg or father's sperm. Because of this error, when fertilization does take place, the embryo has an additional third chromosome 21 or "trisomy 21."

It is not believed that Down syndrome is a result of the parents' behavior or environmental reasons.

Down Syndrome Risk Factors

Researchers have found that it is more likely for an older mother to have reproductive cells with an extra copy of chromosome 21. So an older mother is more likely to have a baby with Down syndrome than a younger mother.

However, most of the babies who have Down syndrome (about 75%) are born to mothers who are 35 or younger. This is because older mothers tend to have fewer babies. (Only about nine percent of total births occur in mothers over age 35 - but about 25% of babies with Down syndrome are born to women in this age group.)

The likelihood that a woman under age 30 who becomes pregnant will have a baby with Down syndrome is less than one in 1,000, but the chance of having a baby with Down syndrome increases to 1 in 400 for women who become pregnant at age 35. The likelihood of Down syndrome continues to increase as a woman ages, so that by age 42, the chance is one in 60 that a pregnant woman will have a baby with Down syndrome, and by age 49, the chance is one in 12.

Because the chances of having a baby with Down syndrome increase with the age of the mother, many health care providers recommend that women over age 35 have pre-birth testing for the condition. Testing the baby before it is born to see if he or she is likely to have Down syndrome allows parents and families to prepare for the baby's special needs.

Parents who have already have a baby with Down syndrome or who have abnormalities in their own chromosome 21 are also at higher risk for having a baby with Down syndrome.

Down Syndrome Diagnosis

A newborn baby with Down syndrome often shows unique physical signs. The doctor usually recognizes these features immediately after the baby is born. These may include a flattened face, an upward slant to the eye, a short neck, abnormally shaped ears, white spots on the iris of the eye (called Brushfield spots), and a single, deep crease on the palm of the hand.

However, a child with Down syndrome may not have all of these features.

Once the baby is born, a blood test can confirm whether the baby has Down syndrome.

This test is called a chromosomal karyotype. To get the results from this test, cells from the baby's blood needs to "grow" for about two weeks. The cells are then examined under a microscope to see if there is extra material from chromosome 21.

Down Syndrome Signs and Symptoms

Most people with Down syndrome have mild or moderate mental retardation. Mental retardation is a disability that limits a person's intellectual abilities as well as the behaviors that people use to function in their everyday lives.

People with Down syndrome may also have delayed language development and slow motor development, which is the ability to use their muscles.

Some of the common physical signs of Down syndrome include:

- A flat face with an upward slant to the eye, a short neck, and abnormally shaped ears
- A deep crease in the palm of the hand
- White spots on the iris of the eye
- Poor muscle tone, loose ligaments
- Small hands and feet

Even though people with Down syndrome may have some physical and mental features in common, the symptoms of Down syndrome can range from mild to severe.

What Are People with Down's Syndrome Able to Do?

Children and adults with Down syndrome have a wide range of abilities. A person with Down syndrome may be very healthy or may have unusual and demanding medical and social problems at virtually every stage of life.

It's important to remember that every person with Down syndrome is a unique individual. Each child will develop at his or her own pace. It may take children with Down syndrome longer than other children to reach developmental milestones, but many of these milestones will eventually be met. Therefore, parents should not compare the progress of a child with Down syndrome to the progress of other siblings or even to other children with Down syndrome.

Down Syndrome Common Health Problems

A variety of health conditions are often seen in people who have Down syndrome, including:

- Congenital heart disease - (heart disease that is present at birth)
- Hearing problems
- Intestinal problems, such as blocked small bowel or esophagus
- Celiac disease (a digestive disease that damages the small intestine)
- Eye problems, such as cataracts (a cloudiness in the lens of the eye)
- Thyroid dysfunctions (problems with the gland that affects metabolism)
- Skeletal problems
- Dementia—similar to Alzheimer's

Newborns with Down Syndrome

Babies with Down syndrome often have poor muscle tone and a tongue that sticks out. Therefore, feeding babies with Down syndrome usually takes longer. Mothers breast-feeding infants with Down syndrome should seek advice from an expert on breast feeding to make sure the baby is getting enough nutrition.

Poor muscle tone can cause constipation in babies. Also, a malformation of the upper part of the spine located under the base of the skull, which is present in some individuals with Down syndrome, can cause compression of the spinal cord if it is not treated properly.

Medical care for infants with Down syndrome should include the same well-baby care that other infants receive. In addition, special attention should be paid to problems that are more common in children with Down syndrome.

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Children with Down Syndrome

A child with Down syndrome is often slow to turn over, sit, stand, and respond. This may be related to the child's poor muscle tone. Verbal skills may take longer to develop, compared to other children, and may not reach the level some parents would like. But children with Down syndrome usually do develop the communication skills they need. In fact, many children with Down syndrome can fit well into regular classes at school.

During the early years of life, children with Down syndrome are 10 to 15 times more likely than other children to develop leukemia, a disease that can be fatal. Children with Down syndrome tend to develop respiratory infections that keep coming back, middle ear infections, and tonsillitis (infected tonsils). Children with Down syndrome also get pneumonia more frequently than most other children.

Down Syndrome Treatment

Down syndrome cannot be cured. However, early treatment can help many people with Down syndrome to live productive lives well into adulthood.

Children with Down syndrome can often benefit from speech therapy, occupational therapy, and exercises to help improve their motor skills. They might also be helped by special education and attention at school.

Some of the medical problems common in people with Down syndrome, like cataracts, hearing problems, thyroid problems, and seizure disorders, can be also treated or corrected.

It has been suggested that children with Down syndrome might benefit from medical treatment that includes amino acid supplements and a drug known as Piracetam. Piracetam is a drug that some people believe may improve the ability of the brain to learn and understand. However, there have been no controlled clinical studies with Piracetam to date in the U.S. or elsewhere that show its safety and efficacy.

Down Syndrome Prognosis

The life expectancy for people with Down syndrome has increased substantially. In 1929, the average life span of a person with Down syndrome was nine years. Today, it is common for a person with Down syndrome to live to age 50 and beyond.

In addition to living longer, people with Down syndrome are now living fuller, richer lives than ever before as family members and contributors to their community. Many people with Down syndrome form meaningful relationships and eventually marry.

Now that people with Down syndrome are living longer, the needs of adults with Down syndrome are receiving greater attention. With assistance from family and caretakers, many adults with Down syndrome have developed the skills required to hold jobs and to live semi-independently.'

Teenagers with Down Syndrome

Like all teenagers, individuals with Down syndrome undergo hormonal changes during adolescence. Therefore, teenagers with Down syndrome should be educated about their sexual drives.

Scientists have medical evidence that males with Down syndrome generally have a reduced sperm count and are rarely able to father children. However, females with Down syndrome have regular menstrual periods and are capable of becoming pregnant and carrying a baby to term.

Health Problems in Adults with Down Syndrome

Premature aging is a characteristic of adults with Down syndrome. In addition, dementia, or memory loss and impaired judgment similar Alzheimer disease patients, may appear in adults with Down syndrome. This condition often occurs when the person is younger than forty years old. Family members and caretakers of an adult with Down syndrome must be prepared to get involved if the individual begins to lose the skills needed to live independently.

Resources

<p>Administration on Developmental Disabilities Administration for Children and Families U.S.Department of Health and Human Services Mail Stop: HHH 300F 370 L'Enfant Promenade S.W. Washington,DC 20447 (202) 690-6590</p>	<p>American Speech, Language and Hearing Association 10801 Rockville Pike Rockville,MD 20852 1-800-638-8255 or 1-888-321-ASHA http://www.asha.org/</p>
<p>Learning Disabilities Association of America 4156 Library Road Pittsburgh,PA 15234-1349 (412) 341-1515 or 1-888-300-6710 http://www.ldanatl.org/</p>	<p>March of Dimes 1275Mamaroneck Avenue White Plains, NY 10605 (914) 428-7100 1-888-MODIMES (1-888-663-4637) http://www.modimes.org/</p>
<p>National Down Syndrome Congress 1370 Center Drive, Suite 102 Atlanta, GA30338 1-800-232-6372 (770) 604-9500 http://www.ndscenter.org/</p>	<p>National Down Syndrome Society 666 Broadway New York, NY 10012 1-800-221-4602 (212) 460-9330 http://www.ndss.org/</p>
<p>National InformationCenter for Children and Youth with Disabilities P.O. Box 1492 Washington,DC 20013-1492 1-800-695-0285 (202) 884-8200 http://www.nichcy.org/</p>	<p>National Society of Genetic Counselors 233 Canterbury Drive Wallingford, PA19086-6617 (610) 872-7608 http://www.nsgc.org/</p>