



## The Learning Curve

*“Enhancing the Knowledge of DDDS Professionals”*

### Facts About Down Syndrome ...

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#### Did you know that...

- **More than 400,000 people in the United States have Down Syndrome.**

#### Down Syndrome (DS) Facts

- **Down Syndrome represents approximately 5,000 births per year in the United States.**
- **One in every 733 live births is a child with Down Syndrome.**
- **Life expectancy for individuals with DS has increased dramatically in recent years, from 25 in 1983 to 56.**
- **Most people with DS have IQs that fall in the mild to moderate range of intellectual disability.**

(Article condensed from National Down Syndrome Society website).

For centuries, people with Down syndrome have been alluded to in art, literature, and science. It wasn't until the late 19th century, however, that John Langdon Down, an English physician, published an accurate description of a person with Down syndrome. It was this scholarly work, published in 1866, which earned Down the recognition as the "father" of the syndrome. Although others had previously recognized the characteristics of the syndrome, it was Down who described the condition as a distinct and separate entity.

Throughout the 20th century, advances in medicine and science enabled researchers to investigate the characteristics of people with Down syndrome. In 1959, the French physician, Jerome Lejeune, identified Down syndrome as a chromosomal anomaly when he observed 47 chromosomes present in each cell of individuals with Down syndrome instead of the usual 46. It was later determined that an extra partial or complete 21st chromosome results in the characteristics associated with Down syndrome.

Down syndrome occurs in one out of every 733 live births, and more than 400,000 people in the U.S. have this genetic condition. One of the most frequently occurring chromosomal abnormalities, Down syndrome affects people of all ages, races and economic levels. Today, individuals with Down syndrome are active participants in the educational, vocational, social and recreational aspects of our communities. In fact, there are more opportunities than ever before for individuals with Down



syndrome to develop their abilities, discover their talents and realize their dreams. For example, more teens and adults with Down syndrome each year are graduating from high school, going to college, finding employment and living independently.

The opportunities currently available to individuals with Down syndrome have never been greater. However, it is only through the collective efforts of parents, professionals, and concerned citizens that acceptance is becoming even more widespread. It is the mission of the National Down Syndrome Society to ensure that all people with Down syndrome are provided the opportunity to achieve their full potential in all aspects of their lives.



#### What Causes Down Syndrome

The human body is made of cells. All cells contain a center, called a nucleus, in which genes are stored. Genes, which carry the codes responsible for all our inherited characteristics, are grouped along rod-like structures called chromosomes.

Normally, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when some or all of a person's cells have an extra full or partial copy of chromosome 21.

The most common form of Down syndrome is known as **Trisomy 21**. Individuals with Trisomy 21 have 47 chromosomes instead of the usual 46 in each of their cells. The condition results from an error in cell division called nondisjunction. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This error in cell division is responsible for 95 percent of all cases of Down syndrome.

Down syndrome also encompasses two other genetic conditions: **mosaicism** and **translocation**. Mosaicism occurs when nondisjunction of chromosome 21 takes place in one of the initial cell divisions after fertilization causing a person to have 46 chromosomes in some of their cells and 47 in others. The least common form of Down syndrome, mosaicism accounts for only 1 to 2 percent of all cases. Translocation, which accounts for 3 to 4 percent of cases of Down syndrome, occurs when part of chromosome



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666 Broadway  
New York, NY 10012  
Phone: (800) 221-4602/  
(212) 460-9330

<http://www.ndss.org/>



Down Syndrome Association of  
Delaware  
PO Box 747  
Middletown, DE 19709  
Phone: (302) 995-1004

<http://www.dsadelaware.org/>



21 breaks off during cell division and attaches to another chromosome, usually chromosome 14. While the total number of chromosomes in the cells remains 46, the presence of an extra part of chromosome 21 causes the characteristics of Down syndrome.

The cause of the extra full or partial chromosome is still unknown. What we do know that it is not caused by environmental factors or anything the mother does before or during her pregnancy. Maternal age is the only factor that has been linked to an increased chance of having a baby with Down syndrome resulting from nondisjunction. A 35-year-old woman has a one in 350 chance of conceiving a child with Down syndrome. By age 45, the incidence has increased to one in 30. However, because younger women have higher fertility rates, 80 percent of babies with Down syndrome are born to women under the age of 35. Once a woman has given birth to a baby with Down syndrome, the chance of having a second child with Down syndrome is about 1 in 100, although age may also be a factor.

Maternal age, however, is not linked to the chance of having a baby with translocation. Most cases are sporadic, chance events, but in about one third of translocation cases, one parent is a carrier of a translocated chromosome. For this reason, the chance of translocation in a second pregnancy is higher than that seen in nondisjunction.

#### Tips for Proper Use of language for DS

Down vs. Down's. The preferred spelling is "Down syndrome," rather than "Down's syndrome." While Down syndrome is listed in many dictionaries with both spellings, the preferred spelling in the United States is "Down syndrome," because an "apostrophe s" connotes ownership or possession.

People with Down syndrome should always be referred to as people first. Instead of "a Down syndrome child," it should be "a child with Down syndrome." Also avoid "Down's child" and describing the condition as "Down's," as in, "He has Down's."

Down syndrome is a condition or a syndrome, not a disease.

People "have" Down syndrome, they do not "suffer from" it and are not "afflicted by" it.

It is clinically acceptable to say "mental retardation," but you may want to use the more socially acceptable "cognitive disability" or "cognitive impairment."



#### Down Syndrome Myths and Truths

**Myth: DS is a rare genetic disorder.**

**Truth:** DS is the most commonly occurring genetic condition. One in every 733 live births is a child with DS, representing approximately 5,000 births per year in the United States.

**Myth: Most children with DS are born to older parents.**

**Truth:** Most children with DS are born to women younger than 35-years-old simply because younger women have more children. However, the incidence of births of children with DS increase with the age of the mother.

**Myth: People with DS are severely "retarded."**

**Truth:** Most individuals with DS have IQs that fall in the mild to moderate range of intellectual disability (formerly known as "retardation"). Children with DS fully participate in public and private educational programs. Educators and researchers are still discovering the full educational potential of individuals with DS.

**Myth: Adults with DS are unemployable.**

**Truth:** Businesses are seeking young adults with DS for a variety of positions. They are being employed in small- and medium-sized offices: by banks, corporations, nursing homes, hotels and restaurants. They work in the music and entertainment industry, in clerical positions, child-care, the sports field and in the computer industry. Individuals with DS bring to their jobs enthusiasm, reliability and dedication.

**Myth: Individuals with DS are always happy.**

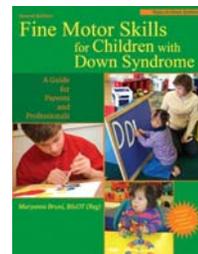
**Truth:** Individuals with DS have feelings just like everyone else in the population. They experience a full range of emotions. They respond to positive expressions of friendship and they are hurt and upset by inconsiderate behavior.

**Myth: Adults with DS are unable to form close interpersonal relationships leading to marriage.**

**Truth:** Individuals with DS date, socialize, form ongoing relationships and marry.

**Myth: DS can never be cured.**

**Truth:** Research on DS is making great strides in identifying the genes on chromosome 21 that cause the characteristics of DS. Scientists now feel strongly that it will be possible to improve, correct or prevent many of the problems associated with DS in the future.



#### Resources

The Department of Human Genetics at the Medical College of Virginia/Virginia Commonwealth University has a very nice booklet on this topic that is available free of charge.

#### *Additional Resources for Down Syndrome*

#### INFORMATION AND ASSISTANCE

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

American Speech, Language and Hearing Association  
<http://www.asha.org/>

Learning Disabilities Association of America  
<http://www.ldanatl.org/>

March of Dimes  
<http://www.modimes.org/>

National Down Syndrome Congress  
<http://www.ndscenter.org/>

National Down Syndrome Society  
<http://www.ndss.org/>

National Information Center for Children and Youth with Disabilities  
<http://www.nichcy.org/>

Mid-Atlantic Regional Human Genetics Network  
<http://www.pitt.edu/~marhgn/>

National Society of Genetic Counselors  
<http://www.nsgc.org/>

The ARC  
<http://www.thearc.org>