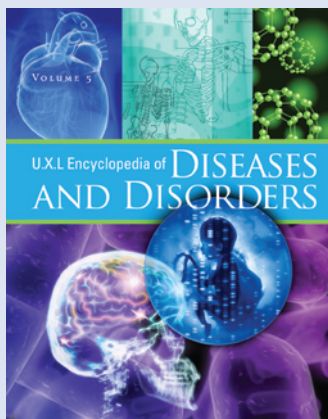



# U·X·L® Encyclopedia of Diseases and Disorders


## Explaining medical concepts without confusing jargon



### U·X·L Encyclopedia of Diseases and Disorders

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Down Syndrome

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). *Diabetes Overview*. Available online at <http://diabetes.niddk.nih.gov/dm/pubs/overview/index.htm> (updated September 2006; accessed on September 21, 2008).

National Library of Medicine (NLM). *Diabetes—Introduction*. Available online at <http://www.nlm.nih.gov/medlineplus/tutorials/diabetesintroduction/html/index.htm> (accessed on September 21, 2008). This is an online tutorial with voiceover; viewers have the option of a self-playing version, an interactive version with questions, or a text version.

TeenHealth. *Diabetes Basics*. Available online at [http://kidshealth.org/teen/diabetes\\_basics/index.html](http://kidshealth.org/teen/diabetes_basics/index.html) (updated September 2007; accessed on September 21, 2008).



### Down Syndrome

**Definition**  
Down syndrome, or trisomy 21, is a genetic disorder caused by the presence of an extra copy of chromosome 21 or by a portion of chromosome 21 translocated (attached) to another chromosome in one of the affected child's parents.

**Description**  
Children with Down syndrome have some degree of mental retardation (average IQ scores are 35–70) as well as characteristic facial features that include a head that is smaller than average, upward-slanting eyes, and a flattened nose. The hands are short and broad with short fingers, and they often have a single crease across the palm. Another characteristic feature of Down syndrome is hypotonia, which is the medical term for poor muscle tone. Children with Down syndrome often need extensive physical therapy in order to learn to walk and move normally. In addition, normal growth is slowed; most of these children never reach full adult height.  
Babies with Down syndrome are often born with severe heart defects or blockages of the esophagus and small intestine. These conditions may require surgery shortly after birth. These children are also at increased risk of childhood leukemia.

**Also Known As**  
Trisomy 21


**Cause**  
An extra copy of human chromosome 21

**Symptoms**  
Small head; distinctive facial features; slow physical, mental, and social development

**Duration**  
Lifelong

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Down Syndrome



Young boy with Down syndrome. SHUTTERSTOCK

Adolescents and adults with Down syndrome are more likely than other people to develop health problems that include frequent infections, cataracts, gastrointestinal reflux disease, hearing problems, sleep apnea, dislocated hips, and hypothyroidism.

**Demographics**  
Down syndrome occurs in about one in every 800 live births in the United States, or about 6,000 children per year. These babies, however, represent only about a quarter of those conceived with trisomy 21. The condition is linked to so many heart defects and other problems that affect survival before birth that about 75 percent of fetuses conceived with Down syndrome are miscarried.  
Down syndrome occurs with equal frequency in all races and ethnic groups, as far as is known. Boys are slightly more likely to be affected than girls.

**Causes and Symptoms**  
Down syndrome results from genetic errors during the formation of germ cells (eggs and sperm) or during cell division shortly after the egg is fertilized by the sperm. The most common form of Down syndrome, responsible for about 95 percent of cases, occurs when an egg or sperm

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▲ Easy-to-understand descriptions of diseases and disorders satisfy the curious reader and support student assignments.

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Down Syndrome

### The Children's Advocate

John Langdon Haydon Down (1828–1896) was the British doctor who pioneered the care and education of children with the syndrome that now bears his name. He published the first scientific description of the condition in 1866. Down had originally hoped to become a chemist and studied under Michael Faraday, one of the most gifted scientists of his time, but then decided to enter medical school in 1853. Down was regarded as exceptionally talented; his professors predicted a brilliant future for him in medicine and were stunned when he turned down a prestigious position at the London Hospital to become superintendent of an asylum for mentally retarded children in Surrey in 1858. At that time, the field of mental retardation was considered unworthy of serious interest or concern, and affected children were regarded as beyond help.

For the next ten years Down worked at the Earlwood Asylum, turning it into a model institution for the care of mentally ill as well as retarded children and adolescents. In 1868 he founded a school for the education of children with trisomy 21, which was then termed mongolism. Down believed that these children could indeed learn and contribute to society. He also advocated for higher education for women, arguing against the widespread belief that allowing females to study at the university level would make them more likely to have retarded children.

Two of Down's sons became doctors and continued his work at the school he founded. Although the genetic cause of trisomy 21 was not known in Down's day—he himself attributed it to tuberculosis in the children's parents—he was an important advocate for those affected by the syndrome. The cause of Down syndrome was finally identified in 1959 by Jérôme Lejeune (1926–1994), a French pediatrician and geneticist.

carrying two copies of chromosome 21 is involved in conception. The reason for the extra copy in the abnormal germ cell is a genetic error called nondisjunction. During the normal process of germ cell formation, the paired chromosomes in the cell divide so that each daughter cell has only one member of the pair. In nondisjunction, one daughter cell gets both members of the chromosome pair and the other cell has none. If a germ cell carrying two copies of chromosome 21 is fertilized by a normal germ cell from the other parent, the child will have three copies of chromosome 21. This genetic error is called a full trisomy 21.

Some children with Down syndrome have some body cells with the extra copy of chromosome 21 and some body cells without the extra copy. This condition is called mosaic trisomy 21. It is thought to result from random errors in cell division during the early stages of fetal development. Mosaic trisomy 21 accounts for about 2 percent of children with Down syndrome.

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▲ Sidebars — such as short bios of famous people — fascinate young readers.

