

📖 Topic Page: [Down syndrome](#)

Definition: **Down's syndrome** from *Philip's Encyclopedia*

Human condition caused by a chromosomal abnormality. It gives rise to varying degrees of mental retardation, decreased life expectancy, and perhaps physical problems, such as heart and respiratory disorders. The syndrome was first described by a British physician, J.L.H. Down. It is caused by the presence of an extra copy of chromosome 21, and detected by counting chromosomes in the cells of the fetus during pre-natal testing. There is evidence that the risk of having a Down's child increases with maternal age. Originally called 'Mongolism' by Down, this term is now obsolete.

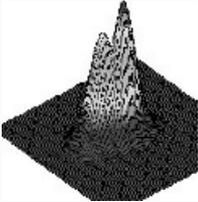


Image from:

[Population distributions for \$\alpha\$ -fetoprotein \(AFP\)... in *Encyclopedia of the Human Genome*](#)

Summary Article: **Down Syndrome**

From *Encyclopedia of Global Health*

Down syndrome, also called Down's syndrome or trisomy 21, is a congenital disorder caused by the presence of all or part of an extra 21st chromosome. This gives people with Down syndrome 47 chromosomes, rather than 46. It acquired the name after the British doctor John Langdon Haydon Down (1828–1896) who first described it in 1866. The outward physical signs of the disorder, usually identified at birth, are a range of major and minor differences in body structure, including an impairment of cognitive ability and also physical growth.

The incidence of Down syndrome is estimated at 1 for every 800 to 1,000 births, and it was first recognized by John L.H. Down as a different form of mental retardation in 1866, and four years later he published his report “Observations” on an Ethnic Classification of Idiots which was published in the Clinical Lecture Reports from London Hospital. Down, born in Torpoint, Cornwall, was from a well-connected family—his great-great grandfather on his father's side was the Roman Catholic Bishop of Derry, and the daughters of his sister married into the Darwin and Keynes families. Apprenticed to his father, a village apothecary, Down later went to work as a surgeon in London, and then to the Royal London Hospital and the Royal College of Surgeons, and became a Fellow of the Royal College of Physicians. He then worked at the Earlswood Asylum for Idiots in Surrey, and conducted much of his work at the Normansfield Asylum, in Teddington, Middlesex. His work used many terms such as mongolism which have long since stopped being used. There were various programs in the United States and also Nazi Germany involving identifying people suffering from Down syndrome, and then embarking on forcible sterilization, even though the cause remained unknown at the time, but was believed to be genetic.

In 1961, some 19 prominent geneticists wrote an open letter to *The Lancet* in which they argued that the “mongoloid” description should be dispensed with, and the journal supported the new term *Down's Syndrome*. Four years later, the delegate to the World Health Organization from Mongolia also objected to the term, forcing it to be dropped as an official term.

The vast majority of the people who suffer from Down syndrome have a third chromosome which is associated with the chromosome 21 pair. This is why the disorder is often known as trisomy 21. There are also some 4 percent of sufferers who have an abnormal condition known as translocation. This is because in their bodies, the extra chromosome in the 21 pair has broken off and attached itself to

another chromosome. In spite of much research into the cause of Down syndrome, the reason for the chromosomal abnormalities is still unknown although it has been shown that there is a higher incidence of Down syndrome in the offspring of women who give birth over the age of 35. Although statistically the number of Down syndrome births in children remains one in 800–1,000, the level of incidence in women who give birth over the age of 40 are one in 40. This has led to a range of tests which can be used to diagnose Down syndrome prenatally by checking for the presence of the abnormal chromosome in samples of the fetal cells which can be collected from the amniotic fluid.

As well as an extra chromosome, there are many common physical features associated with Down syndrome. These involve having shorter limbs and neck, poor muscle tone, and a single transverse palmar crease. Those affected with Down syndrome also usually have an enlarged protruding tongue and lips, a sloping underchin, and eyes which have an almond shape, sometimes with an inner epicanthal fold, and low-set ears. There are also several problems such as a lower-than-average cognitive ability, at its most extreme involving moderate mental retardation, with some having severe mental retardation, as well as heart and kidney malformations. Some 40 percent of people with Down syndrome also tend to suffer from congenital heart disease.

A number of notable people who suffered from Down syndrome include the German actor Bobby Brederlow, the New York actor Chris Burke, the Belgian actor Pascal Duquenne, the London actor Max Lewis, the Scottish film actress Paula Sage, the artist Judith Scott, and the Argentine singer Miguel Tomasin who sung with the avant-rock band Reynols. In addition, Stephanie Ginnsz, an actor, was the first with Down syndrome to take the lead part in a motion picture. Anne de Gaulle (1928–1948), daughter of the French politician Charles de Gaulle, suffered from Down syndrome; and there has been retrospective speculation that Charles Waring Darwin, son of the naturalist Charles Darwin, was also a sufferer from Down syndrome, evidence for this included the fact that Darwin himself conducted a correspondence with John Langdon Haydon Down.

Modern healthcare has meant that many people with Down syndrome now live into adulthood, although those with major heart defects that cannot be subjected to corrective surgery die as children. Overall in the United States and western Europe, people with Down syndrome have a life expectancy of about 55, lower than normal adults, mainly because the condition means that they age prematurely. Many have managed to be self-supporting to a large degree, being able to work in the home, or a sheltered workshop. With increasing tolerance in the wider society, and the greater awareness of Down syndrome, there is a much more general acceptance of the disorder and people with the disorder which was not the case even 30 years ago when many Western countries had asylums where sufferers would spend most of their lives. The work undertaken by the National Down Syndrome Society in the United States, which holds Down Syndrome Month each October, has been very effective in raising the profile of the disorder. The Down Syndrome Awareness Day is held on October 20 each year in South Africa.

SEE ALSO:

Degenerative Nerve Diseases; Healthcare, U.S. and Canada.

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