

What is Down Syndrome?

People with Down Syndrome answer your questions about Down Syndrome (a video in their own words)

https://www.youtube.com/watch?v=o0VV3C_ydak&t=304s

<https://www.ds-int.org/Pages/Category/what-is-down-syndrome>

Myths and facts: <https://www.ds-int.org/myths-and-facts>

Facts about Down Syndrome

<http://www.edsa.eu/down-syndrome/>

Down syndrome is a common condition and the single most common cause of a learning disability. It occurs at the rate of about 1 in 600 – 1000 births. The life expectancy of a person with Down syndrome is now 60 to 65 years with some living even beyond this age.

Down syndrome has existed since [SEP]the beginning of mankind, but has been described in the scientific literature as a specific condition only since 1866.

Children with Down syndrome are born to parents of all social and educational levels, in all ethnic groups, to parents of all ages. The chance of having a baby with Down syndrome increases with maternal age, even more after 35 years, however most babies with Down syndrome are born to mothers under 35.

Chromosomes and genes

Down syndrome is a chromosomal disorder. A person with Down syndrome has an extra copy of chromosome 21 (all or part of the extra chromosome). This means the person will have 47 chromosomes instead of 46. The reason why this happens is still unknown. Chromosome 21 is the smallest of the human chromosomes. Down syndrome cannot be prevented from occurring, nor can it be cured. But with the current knowledge of genetics we know much more about the functioning of the chromosomes, especially about the number 21. This helps us to understand the typical features of the syndrome and may enable us to provide better medical care and intervention methods in the future.

What is the effect of the extra chromosome?

Medical status and cognitive development are affected. Some individuals with Down syndrome may have serious health problems, others have few or no additional health problems. They usually experience some degree of cognitive disability.

The development of children with Down syndrome varies widely and, like everyone, they continue to learn into adult life, if given the opportunity to do so. However it is important to note that, like everyone else, each child or adult must be considered as an individual and it is not possible to predict the future development of a baby with Down syndrome.

Down Syndrome – information in all european languages

In the internet there are a lot of answers on the question „What is Down Syndrome“. Correct and up-to-date information can be found in all languages on the websites of the DS Associations and EDSA members <http://www.edsa.eu/members/> One example from Down's Syndrome Scotland:

What is Down's syndrome?

Down's syndrome is a genetic condition caused by the presence of a full or partial third copy of chromosome 21 in the body's cells. It is sometimes shortened to “Ds” and in some other countries it's called Down syndrome. It is named after the British doctor who first recognised it in 1886, Dr John Langdon Down.

People who have DS are not all the same; every person who has the conditions is as individual as everyone else in the population and have more in common with their families than with another person who has the condition.

The word Syndrome means a set of signs and features that are found together. This is the same for any syndrome not just Down's syndrome.

Approximately one in every 1,000 babies is born with Down's syndrome and in Scotland it is estimated that there are 70 babies born with Down's syndrome each year with the total number of people living with Down's syndrome in Scotland estimated to be 4,500.

Down's syndrome is the most frequently recognised form of learning disability. A learning disability affects a person's ability to learn, it does not mean they cannot learn. Children with Down's syndrome follow the same

developmental path as for all children and individuals have the same range of feelings and emotions as we all do. Most importantly, everyone who has Down's syndrome is a unique individual, full of potential.

What causes Down's syndrome?

Down's syndrome usually occurs randomly at the time of conception and affects males and females alike. It is rarely hereditary and nothing either parents did before or during pregnancy can have caused it.

Inside the cells in our bodies we all have chromosomes which contain information that make each of us unique, such as our eye colour, hair colour and height.

Most people have 46 chromosomes inside every cell of their body which go together in twenty-three pairs – at conception babies receive 23 chromosomes from mum and 23 from dad which make up the 46 pairs.

People born with Down's Syndrome have an extra full or partial copy of chromosome 21 which means they have 47 chromosomes in the cells in their body – one extra chromosome that randomly happens. It is this extra chromosome 21 that causes the characteristics of the condition.

There are three types of Down's syndrome: Trisomy 21, Translocation and Mosaic Down's syndrome.

Trisomy 21 Down's syndrome

The most common type of Down's syndrome (occurring in about 94% of people with Ds) is Trisomy 21, so called as all the cells in the person's body have a full extra copy of chromosome 21, so there are 3 copies of chromosome 21 in every cell.

Translocation Down's syndrome

Around 4% of people with Down's syndrome have translocation Down's syndrome. This is when the extra chromosome 21 material translocates or moves itself to attach to another chromosome in the cell, this is typically chromosome 14 or 17.

Mosaic Down's syndrome

Mosaic Down's syndrome occurs when only some of the cells in the body have the extra chromosome and some cells don't. About 2% of people with Down's syndrome have this type. People with mosaic Down's syndrome are likely to have milder learning disabilities.

DS is a lifelong condition and there is no cure; however, there are many ways to ensure that each individual with Down's syndrome is given the right type and amount of support that they need to develop to their full potential. Today the average life expectancy for a person with Ds is about 60 years old with a small number of people living into their 70s and beyond.

With permission of DS Scotland

Estimation of the number of people with Down syndrome in Europe

"Estimation of the number of people with Down syndrome in Europe" an interesting and important paper, published in the *European Journal of Human Genetics*.^[1]_[SEP] The authors of this paper Gert de Graaf

(Netherlands), Frank Buckley (UK) and Brian Skotko (USA) worked on a **study of the European populations of people with Down Syndrome**, that also EDSA members assisted. Here is the scoop:

- From 1901 until 2015, we have stitched together registries across Europe to calculate a country-by-country estimate of the number of people with Down syndrome, the number of selective pregnancy termination because of Down syndrome, and impact of such terminations on birth prevalence and population prevalence.
- As of the most recent years, we estimate there were 8,031 annual live births of children with Down syndrome in Europe, which would have been around 17,331 births annually, absent selective terminations.
- The estimated reduction of live birth prevalence was, on average, 54%, varying between 0% in Malta and 83% in Spain.
- As of 2015, we estimate there are 417,000 people with Down syndrome living in Europe; without elective terminations, there would have been about 572,000 people with DS, which corresponds to a population reduction rate of 27%.
- Such statistics can be important barometers for prenatal testing trends and resource allocation within countries. Disability awareness initiatives and public policy initiatives can also be better grounded with these more precise estimates.

The paper is publicly available in the „European Journal for Human Genetics“ 2021 and can be found at <https://rdcu.be/b9w27> (along with the country-by-country statistics in

our 279-page supplement).  