

# Introduction to Down syndrome



## History of Down syndrome



Down syndrome is a genetic disorder that was named after John Langdon Down, the doctor who first recognised it as a distinct condition in 1866. It is a lifelong condition that develops when a baby is still in the womb. About one in every 550 babies in Ireland is born with Down syndrome. It affects all ethnic groups equally.



## What is Down syndrome?

In every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits (or, in other words, genes make you 'you') and are grouped along rod-like structures called chromosomes. In traditionally-developed individuals, each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21.



## Important points about Down syndrome

The most important thing to note is that Down syndrome is just a **part** of someone. It does not define them. People with Down syndrome are people first and foremost and deserve the respect and treatment that you would give any other person.

As such it is incorrect to say “Down syndrome person”. The correct term is “person with Down syndrome” due to the fact that – as discussed – the person’s Down syndrome is only a part of what makes them who they are.

Down syndrome is not a sickness or a disease, it is a genetic disorder or chromosomal anomaly that occurs at the time of conception.

# Types of Down syndrome

There are three types of Down syndrome: trisomy 21 (nondisjunction), translocation and mosaicism.

## TRISOMY 21

This is the most common cause of Down syndrome. Trisomy 21 results in an embryo with three copies of chromosome 21 instead of the usual two. 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 type of Down syndrome accounts for 95% of cases.

## TRANSLOCATION

In translocation - which accounts for about 4% of cases of Down syndrome - the total number of chromosomes in the cells remains 46; however, an additional full or partial copy of chromosome 21 attaches to another chromosome, usually chromosome 14. The presence of the extra full or partial chromosome 21 causes the characteristics of Down syndrome.

## MOSAICISM

Mosaicism (or Mosaic Down syndrome) is diagnosed when there is a mixture of two types of cells, some containing the usual 46 chromosomes and some containing 47. Those cells with 47 chromosomes contain an extra chromosome 21. Mosaicism is the least common form of Down syndrome and accounts for only about 1% of all cases of Down syndrome. Research has indicated that individuals with mosaic Down syndrome may have fewer characteristics of Down syndrome than those with other types of Down syndrome. However, broad generalizations are not possible due to the wide range of abilities people with Down syndrome possess.

Regardless of the type of Down syndrome a person may have, all people with Down syndrome have an extra, critical portion of chromosome 21 present in all or some of their cells. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome.

## Common attributes in people who have Down syndrome

People with Down syndrome often have similar characteristics. Not everyone will have all of them, but they often include:

- Eyes that slant upwards, small ears, flat back of head, small mouth, protruding tongue, flattened nose bridge, white spots on the iris known as Brushfield spots.
- Lower than average height
- Broad hands with a single crease across the palm
- Loose skin on the back of the neck
- Loose joints (babies in particular may seem 'floppy')
- Low muscle tone (hypotonia)
- Low birth weight
- Vertical skin folds (epicanthic folds) between the upper eyelids and inner corner of the eye
- Delayed mental and physical development. It might take babies and children with Down syndrome a longer time to reach developmental milestones such as learning to crawl, walk, speak or read

It is important to keep in mind that a child with Down syndrome may still be able to acquire most, if not all, of the mental and social skills that most other people develop. Their development just usually occurs at a slower pace.



## Support for new parents

Having a baby is an exciting but also a tiring time for new parents. When you are told that your newborn baby has Down syndrome it is quite natural to experience a wide range of differing emotions.

It is very common for parents of a new baby who has Down syndrome to experience a number of emotions at any given time, particularly during the first few weeks and months. Many parents feel scared, sad, angry, isolated, confused, overwhelmed whereas others find it easy to accept the special needs that their new baby has.

**There is no right or wrong way for a parent to react after discovering that their child has Down syndrome.**

The Down Syndrome Centre is dedicated to offering practical and emotional support to the families and carers of children with Down syndrome, along with providing early intervention services to babies and children with Down syndrome.

Please don't hesitate to pick up the phone and give us a call – even if you don't know what to say or what you are looking for.

## Our Support Mum



*“A diagnosis of Down syndrome does not define your child. Your little baby is still just a little baby. This baby is 50% you and 50% your partner. This baby doesn’t belong to an unknown and scary tribe of babies called “downs babies”. This is just your baby and it just happens to have Down syndrome. The rest is all uniquely your baby. Their personality, their likes and dislikes. All your own family traits – not traits of Down syndrome. Don’t buy into the old wives tales of “Oh they are all so loving” or “They all love music” etc etc. Your baby is your baby and has his or her own personality. They are an individual.*

*The more you get to know your baby, the less important the Down syndrome will become. It really is just a part of your child. It is a learning delay full stop. Don’t overthink it! Keep it simple in your head. Remember they will do all the same things that other babies do. They will eat, sleep and poo for the first few months. They will smile and laugh, rollover, crawl, walk – just in their own time. As with any baby I say enjoy it, it goes way too fast!” Triona Cussen*

Part of our services here in The Down Syndrome Centre includes our Support Mum, Triona Cussen whose youngest son, Joshua has Down syndrome. Triona is delighted to speak with new parents and offer her insights and support.



## Contact Us

To contact our team or to be put in touch with Triona contact our main reception on (01) 661 8000 or email [info@downsyndromecentre.ie](mailto:info@downsyndromecentre.ie)

We love visitors and will be delighted to put the kettle on and have a chat with you!

We also recommend that new parents check out our New Parent booklet at [www.downsyndromecentre.ie/useful-information/new-expectant-parents/](http://www.downsyndromecentre.ie/useful-information/new-expectant-parents/)

