



DOWN SYNDROME

FACT SHEET



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Down Syndrome

What is Down syndrome?

The word syndrome means a collection of characteristics. The name Down Syndrome comes from the doctor, John Langdon Down who first described the condition in 1866. It is a genetic condition caused by the presence of an extra chromosome. People with Down syndrome have 47 chromosomes in some or all of their cells instead of the usual 46.

Why does it happen?

Down syndrome has an estimated incidence of about 1 in every 800 live births. It affects both boys and girls equally and is associated with increased maternal age. It is caused by the presence of extra chromosomal material and can occur in three main ways.

1. **Trisomy 21.** This accounts for about 95% of cases. Here, the person has three copies of chromosome number 21 instead of the usual two. This occurs by chance.
2. **Translocations** account for 3 - 4 % of cases. A translocation is a genetic anomaly where a chromosome or part of one is joined to another one. Translocations that cause Down syndrome involve an **extra** copy of chromosome 21 (or part of one) attached to another chromosome. The extra material may have been inherited from either parent or may have arisen when the baby was conceived.
3. **Mosaicism** is seen in about 1 percent of people with Down syndrome. In this case, some cells have 47 chromosomes (with an extra 21) and others have the usual 46. This is likely to be the result of an error in cell division soon after conception.

What are the chances of it happening in another pregnancy?

For each pregnancy the risk of Down syndrome can be calculated based on the results of the serum screening tests and taking into account other factors such as the age of the mother. The risk of Down syndrome increases with the age of the mother.

If a couple have already had a pregnancy affected by Down syndrome, which was known to be caused by a translocation, carried by one of the parents, the risk to subsequent pregnancies will be 1 in 2 or 50:50.

How does Down syndrome affect a person?

In general, people who have the same syndrome share the same characteristics. However, it is important to realise that not all of the features of this condition will be seen in all people with Down syndrome. Characteristic features associated with Down Syndrome include:

- Characteristic facial appearance
- Learning disability
- Heart abnormalities in 40% of cases
- Hearing problems
- Over- or under-active thyroid
- Respiratory tract problems
- Floppiness or poor muscle tone
- Loose or bendy joints
- Diabetes
- Increased risk of developing Alzheimer's Disease or dementia
- Problems with the intestinal and digestive tract

Does a person with Down syndrome look different?

People with Down syndrome have a characteristic facial appearance that includes:

- Eyes that slant upwards and outwards

- A fold of skin that runs vertically between the two lids at the corner of the eye (the epicanthic fold).
- A low hairline.
- A flatter than usual face
- A flat nasal bridge
- A large tongue
- Small low set ears

People with Down syndrome are often shorter than the general population and often have broad hands with short fingers.

Can a person with Down's syndrome walk?

Yes. However, because the muscle tone is usually poor at birth and in childhood, children often take longer to achieve their physical milestones such as sitting up and walking.

Do people with Down syndrome need medical treatment?

Around 40% of children suffer with a congenital heart abnormality, which may require surgery. This surgery may be minor or major depending upon the condition. The most common problem is an atrial septal defect or hole in the heart. People with Down syndrome may also need to take medication to treat other aspects such as diabetes and thyroid problems.

What sorts of lives do people with Down syndrome lead?

Most people with Down syndrome lead happy healthy lives. In some cases a person with Down syndrome may be limited with regards to physical activity due to heart problems. The physical and mental ability of people with Down syndrome varies considerably and their rate of progress is slower. Physiotherapy, occupational therapy, speech therapy and educational programs can enable children to reach their maximum potential and more and more children with Down syndrome are attending mainstream schools. Most teenagers with Down syndrome will achieve a degree of independence. Most will be able to dress themselves, speak so that they can be understood and have normal desires and interests.

Can people with Down syndrome have children?

Infertility is not a problem associated with Down syndrome. However, most people with Down syndrome do not go on to have children due to their learning difficulties.

Do people with Down syndrome have a reduced life expectancy?

Many people with Down syndrome enjoy a healthy life, and a lifespan of 40-60 years can be expected.

Who can I contact for more information?

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