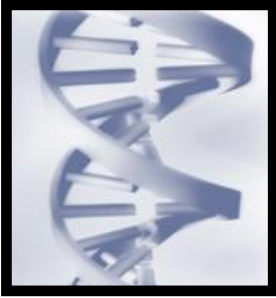


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Down's Syndrome And Its Characteristics

What Is Down's Syndrome?



Down's syndrome is a condition of childhood and adult life in which the individual shows delay of mental development. The child is slower to learn normal physical skills and mental tasks. Intellectual development is impaired into adult life. The condition is incurable and children born with Down's syndrome will have it all their life.

Down's syndrome results from abnormal cell division in the earliest days of the embryo's life (see later). The overall incidence is about 1 in every 700 live births, but there is marked variability in incidence depending upon maternal age. In younger mothers, the incidence is about 1 in 2000 live births; but for mothers over 40 years of age this rises to 1 in every 40 live births. As the number of older mothers having babies has risen rapidly in recent years so too has the need for ante-natal screening to detect the condition early in pregnancy. Some children with Down's syndrome are born to younger women. A couple who have had one child with Down's syndrome are at increased risk of having another.

Children with Down's syndrome are happy, contented children who show relatively little in the way of behavioural problems and who usually have a loving and intimate relationship with their parents. While a lower than average IQ is common, many have normal IQs and can be taught in normal schools. These days, parents who have a child affected by Down's syndrome have every reason to be optimistic about their future.

What Are Down's Syndrome Causes?

Down's syndrome results from abnormal cell division in the very earliest days of the human embryo. Very shortly after the egg has been fertilised by the sperm, it divides into two cells. Normally there will be 23 pairs of chromosomes (ie 46 chromosomes) in each divided cell. In Down's syndrome, one of these chromosomes (known as chromosome number 21) divides again, with the result that there are three and not two of this chromosome in each cell (ie a total of 47). Thereafter every cell of the body will show this same pattern. The presence of this extra chromosome 21 in each cell is associated with the development of Down's syndrome (which is also known as 'trisomy 21').

What Are The Characteristics Of Down's Syndrome?

The diagnosis is usually made at birth when the new-born baby is noticed to possess various characteristic features. Any one of these features can occur in normal people as an isolated peculiarity. When they occur together, however, Down's syndrome becomes more likely.

These peculiarities may include:

- a small round head with a short, broad neck
- a flat face and short nose with a flat bridge
- an upward and outward slant to the eyes which may have heavier skin folds around them than is usual
- small white spots on the iris
- a largish, furrowed tongue
- broad hands with short fifth fingers

Some children with Down's syndrome have heart problems, particularly atrial and ventricular septal defects (ie

holes in the heart). They tend to suffer from more catarrhal upper respiratory tract infections than other children, perhaps associated with the shape of their faces.

Will there be any tests or investigations?

The diagnosis of Down's syndrome may be confirmed by analysis of chromosomes using tissue culture. White cells taken from the blood are commonly used for this purpose though cells taken from the lining of the mouth can also be used.

The risk of having a baby with Down's syndrome rises with the age of its mother. It is common practice for mothers over the age of 40 to be offered screening tests early in their pregnancy through which the risk of carrying a baby with Down's syndrome can be assessed. A variety of screening tests are available including:

- estimation of nuchal skin fold thickness (the thickness of skin on the babies neck as shown on an ultrasound scan)
- blood tests to detect abnormal proteins
- amniocentesis (a sample of the fluid surrounding the baby in the womb is withdrawn using a needle through the mother's abdomen. This is then subjected to genetic tests)
- chorionic villous sampling (in which a small piece of the placenta (afterbirth) is sampled and subjected to genetic testing).

There is no consensus in the UK regarding which of these tests or which combination of tests should be offered. Policies vary according to availability in a particular locality.

What treatment might be necessary?

Though treatment may be needed for abnormalities that are part of the Down's syndrome – eg heart defects - there is no treatment for the syndrome itself. Nevertheless, children born with Down's syndrome are pleasant, amenable and loving. Their mental and physical development is likely to be slower than for a normal child so special schooling, speech therapy and play therapy will be useful. Parents can be taught techniques to assist learning at home. The aim is to raise the child's abilities to a level at which he/she can be taught in a normal school and thereby gain the social skills which will enable him to live in his own home and to achieve a high quality of life in normal society. Many children with Down's syndrome do not achieve this level of ability and find learning and living easier and happier in the protected environment of a special school and/or a sheltered community.

Tell Your Doctor

- if you are intending to get pregnant and ask for pre-conceptual advice
- if you or any member of your family has had a child with Down's syndrome
- if you have any concerns about your pregnancy

Ask Your Doctor

- where you might obtain pre-conceptual genetic advice locally
- the location of the nearest specialist ante-natal screening centre
- what is the normal routine of screening for Down's syndrome and other pregnancy problems in your locality

By Dr Peter Stott

Useful Contact

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