

Down's syndrome Guide

What is Down's syndrome

There is no such thing as a typical person with Down's syndrome. Like all people, they vary a lot in appearance, personality and ability. People with Down's syndrome have learning difficulties. Some have more serious difficulties than others. It is hard to tell in babies how much they will be affected as children, or when they are grown up. It is common today for adults with Down's syndrome to get jobs and live fairly independent lives. However, most people with Down's syndrome need long-term help and support.

Down's syndrome is named after John Langdon Down, the British physician who described the syndrome in 1866. The condition is characterised by a combination of major and minor differences in structure. Often Down syndrome is associated with some impairment of cognitive ability and physical growth, and a particular set of facial characteristics. Down syndrome in a fetus can be identified with amniocentesis during pregnancy, or in a baby at birth.

“Down's syndrome is a genetic condition caused by the presence of an extra chromosome in the body's cells. Down's syndrome is not a disease, and it is not a hereditary condition which occurs by chance at conception”

Down syndrome, or Down's syndrome is a chromosomal disorder caused by the presence of all or part of an extra 21st chromosome (Trisomy 21). People do not usually expect to have a baby with Down's syndrome. It does not usually run in families. Some people think that only older women can have a baby with Down's syndrome, but this is not true. Anyone can have a baby with Down's syndrome, but the risk does go up with age. All women, whatever their age, have a small risk of delivering a baby with Down's syndrome.

Down's syndrome is a chromosomal abnormality where there is an extra chromosome. Down's syndrome is one of the most common genetic abnormalities affecting around 1 in every 800 births worldwide. The risk of Down's syndrome increases with a women's maternal age.

Health and development for people with Down's syndrome

People with Down syndrome tend to have a lower than average cognitive ability, often ranging from mild to moderate developmental disabilities. A small number have severe to profound mental disability.

Down's syndrome and anomaly baby scan screening are a high priority issue with parents. Increasingly parents expect modern antenatal care to include the best baby scan screening and diagnosis for their unborn baby. Currently nuchal screening for Down's syndrome is not widely available at routine NHS antenatal visits so many women opt for an invasive test such as amniocentesis (amino) or chorionic villus sampling (CVS). Amino and CVS are invasive tests (procedure involving incision into the body) and increase the risk of miscarriage.

Physical features of people with Down syndrome

Many of the common physical features of Down syndrome may also appear in people with a standard set of chromosomes, including microgenia (an abnormally small chin), an unusually round face, macroglossia (protruding or oversized tongue), an almond shape to the eyes caused by an epicanthic fold of the eyelid, poor muscle tone, and a larger than normal space between the big and second toes. Health concerns for people with Down syndrome include a higher risk for congenital heart defects, gastroesophageal reflux disease, recurrent ear infections, obstructive sleep apnea, and thyroid dysfunctions.

Early childhood intervention, screening for common problems, medical treatment where indicated, a conducive family environment, and vocational training can improve the overall development of children with Down syndrome. Although some of the physical genetic limitations of Down syndrome cannot be overcome, education and proper care can improve quality of life.

Down syndrome can result from several different genetic mechanisms. This results in a wide variability in individual symptoms due to complex gene and environment interactions. Prior to birth, it is not possible to predict the symptoms that an individual with Down syndrome will develop. Some problems are present at birth, such as certain heart malformations. Others become apparent over time, such as epilepsy.

The most common manifestations of Down syndrome are the characteristic facial features, cognitive impairment, congenital heart disease (typically a ventricular septal defect), hearing deficits (maybe due to sensory-neural factors, or chronic serous otitis media, also known as Glue-ear), short stature, thyroid disorders, and Alzheimer's disease. Other less common serious illnesses include leukemia, immune deficiencies, and epilepsy.

Cognitive development

Cognitive development in children with Down syndrome is quite variable. It is not currently possible at birth to predict the capabilities of any individual reliably, nor are the number or appearance of physical features predictive of future ability. The identification of the best methods of teaching each particular child ideally begins soon after birth through early intervention programs.

Since children with Down syndrome have a wide range of abilities, success at school can vary greatly, which underlines the importance of evaluating children individually. The cognitive problems that are found among children with Down syndrome can also be found among typical children. Therefore, parents can use general programs that are offered through the schools or other means.

Education

In education, mainstreaming of children with Down syndrome is becoming less controversial in many countries. For example, there is a presumption of mainstream in many parts of the UK. Mainstreaming is the process whereby students of differing abilities are placed in classes with their chronological peers.

Children with Down syndrome may not age emotionally/socially and intellectually at the same rates as children without Down syndrome, so over time the intellectual and emotional gap between children with and without Down syndrome may widen. Complex thinking as required in sciences but also in history, the arts, and other subjects can often be beyond the abilities of some, or achieved much later than in other children.

Therefore, children with Down syndrome may benefit from mainstreaming provided that some adjustments are made to the curriculum.

Screening for Down's Syndrome

If you are looking for information on pre-natal testing, we recommend a Nuchal Translucency scan. A nuchal scan measures the amount of fluid at the back of the baby's neck. Together with a blood test a nuchal scan can estimate the risk of your baby having a chromosomal abnormality such as Down's syndrome. A nuchal scan is a non-invasive test (procedure which does not require incision into the body) that can only be performed between 11 weeks +4 days to 13 weeks +6 days of pregnancy. The blood test measures two concentrations BHCG (hormone) and PAPP-A (protein). Research suggests BHCG levels are higher and PAPP-A levels are lower in cases of Down's syndrome. A nuchal scan is not however a diagnostic test, it just gives a risk factor. Some private clinics in the UK

A number of Baby Premier clinics offer OSCAR (one stop clinic for assessment of fetal risk) for Down's syndrome screening. This means that the nuchal scan, blood test and result are done during one visit. The entire process usually takes around 2 – 2 1/2 hours. Other clinics provide a similar service but on a two-stop appointment system.

How are risk factors evaluated?

Down's syndrome risk factors are expressed as ratios. For example a ratio of 1:250 tells us 1 in every 250 pregnant women of the same age and gestation may have a baby born with Down's syndrome. Baby Premier's combined nuchal scan and blood test picks up approximately 90% of unborn babies with Down's syndrome compared to 30% when maternal age is just checked, 50% - 70% when only maternal age and blood test is checked, and 70% - 80% when maternal age and nuchal scan is checked.

NICE recommendations

The National Institute for Clinical Excellence (NICE) have recommended nuchal scanning combined with blood test during the first trimester of pregnancy to be the most effective non-invasive screening test. In the future NICE would like all women to be offered first trimester screening for Down's syndrome so that mums-to-be do not have to go through with unnecessary invasive tests. Unfortunately nuchal screening is not widely available at most NHS antenatal clinics and some clinics do not offer the combined blood test.

Advantages of nuchal screening:

- No risk to you and your baby
- No waiting lists
- Immediate results and advice
- Avoid unnecessary worry
- Peace of mind
- Time to make an informed choice

How can I get more information on Nuchal Testing for Down's syndrome?

Choosing whether to have a nuchal scan the tests is an important decision, for you and for your baby. If you are faced with this decision, you need to make sure you reach the right decision for you.

For more information on nuchal screening or any obstetric medical ultrasound scans please contact the Baby Premier advice and bookings clinic on **0845 345 7262** or email info@babypremier.co.uk

Baby Premier is the obstetric and gynaecological division of SMI (Specialist Medical Imaging Ltd), a medical ultrasound company established in 2004. Baby Premier is an accredited ultrasound service that offers a full range of obstetric and gynaecological ultrasound examinations to both self-funding and privately insured patients.