



**Down's Syndrome
Association**

A Registered Charity No. 1061474

A Guide for Healthcare Professionals



Antenatal, Neonatal and Postnatal Care

A Down's Syndrome Association publication

Our resources and Information Team are here to help

Please see our website for up-to-date information: www.downs-syndrome.org.uk

If you would like to talk about any of the issues raised in this resource, then please get in touch with our helpline or by emailing us on info@downs-syndrome.org.uk.

Helpline Monday - Friday 10am-4pm | Telephone: 0333 1212 300

Who this booklet is for

This booklet has been written for healthcare professionals involved in antenatal, neonatal and postnatal care. It gives an overview of what Down's syndrome is, how it can be identified and some examples of people living with Down's syndrome today. Taking notice of their lived-experience of having Down's syndrome is imperative if health professionals are to provide new and prospective parents with up to date, balanced and accurate information about Down's syndrome. The main focus of this booklet is to provide advice and guidance on telling parents that their unborn or newly born baby has Down's syndrome. Therefore, any clinical information provided is brief in nature.

Tell It Right® Start It Right

The key objective of our Tell It Right® Start it Right campaign is to ensure that health professionals involved in antenatal, neonatal and postnatal care have up to date, accurate and balanced information about living with Down's syndrome today, in order to support expectant parents through screening choices and new parents after birth.

Our Royal College of Midwives accredited study days ensure that all the relevant professionals who attend gain accurate, balanced and up to date information about Down's syndrome and living with Down's syndrome.



To find out more visit www.downs-syndrome.org.uk/TellItRight/

Further DSA resources you may find useful:

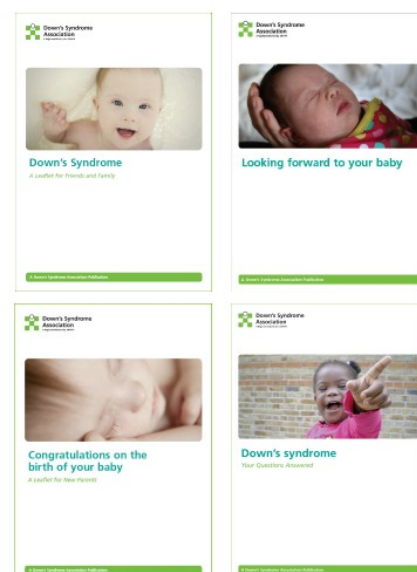
Tell It Right® Information Pack:

Download key resources for parents who have just had a diagnosis, pre- or post- natally.

Go to www.downs-syndrome.org.uk/TellItRight/

Closed Facebook groups for professionals:

- DSA Tell It Right® Group
- DSA Health Professional Network



Since the Nuffield Council on Bioethics Review in 2017, which recommended an evaluated introduction of NIPT in NHS settings, the DSA has been working with Public Health England in a review of the information materials that support antenatal screening and have been involved in the development and delivery of face to face training courses and an e-learning resource for health practitioners involved in delivering antenatal care. A similar programme of work has been undertaken with Antenatal Screening Wales. In Wales, NIPT has been offered, as a supplementary test, for women who have a higher-chance combined test result, since April 2018. Introduction in England is expected in the near future.

About Down's syndrome

What is Down's syndrome and when was it discovered?

Down's syndrome is the most commonly occurring chromosome condition.

Although Down's syndrome was not described medically until the 19th century, it is believed that people with Down's syndrome have always been part of society. However, it was not until 1866 that the English doctor, John Langdon Down, published a description of the condition which subsequently took his name.

In 1959 Professor Jérôme Lejeune proved that Down's syndrome is a chromosomal irregularity. Instead of 46 chromosomes usually present in each cell, Lejeune noted 47 in the cells of people with Down's syndrome; with a third copy of chromosome 21 (hence the name 'Trisomy 21'). It was later determined that this additional chromosomal material results in the physical characteristics associated with the condition and the different course in development.

There are three types of Down's syndrome:

- **Trisomy 21:** in which all the cells have an extra chromosome 21.
About 94% of people with Down's syndrome will have this type.
- **Translocation:** in which extra chromosome 21 material is attached to another chromosome.
Around 4% of people with Down's syndrome have this type.
- **Mosaic:** in which only some of the cells have an extra chromosome 21.
About 2% of people with Down's syndrome have this type.

All women have a chance of having a baby with Down's syndrome. Nothing a woman or her partner has done or not done can make any difference to this chance.

All people with Down's syndrome have a learning disability. This means that development and learning new things may take longer. There is now a greater understanding of how children with Down's syndrome learn. Support in schools can be provided and individuals with Down's syndrome can and do learn new skills throughout their lives.

Opportunities for inclusion within society are greater now than ever before. Individuals vary greatly, however, and have different needs. These needs cannot be predicted before the baby is born.

Some health conditions are more common in people with Down's syndrome. These include heart conditions, as well as problems with vision and hearing.

Many health conditions can be treated, though unfortunately around 5% of babies will not live past their first birthday. For babies without serious health problems survival is similar to that of other children.

Most people with Down's syndrome will live into their 60s or longer. Most people who have Down's syndrome lead healthy and fulfilled lives.

Parents say that their children have a positive influence on family life. With support, many more adults are now able to get jobs, have relationships and live in homes of their choosing.



What is the incidence and prevalence of Down's syndrome?

- For every 1,000 babies born in England and Wales, one will have Down's syndrome.¹
- Down's syndrome typically occurs in around 1 in every 750 pregnancies, resulting in around 1 in every 1,000 live births being affected.
- It is estimated that there were approximately 37,090 people with Down's syndrome living in England and Wales in 2011.²

¹ Morris JK, Springett A. (2014). The National Down Syndrome Cytogenetic Register for England and Wales 2013. Annual Report. Queen Mary University of London, Barts and The London School of Medicine and Dentistry.

² Wu, J and Morris, KJ (2011) The population prevalence of Down's syndrome in England and Wales in 2011 Eur J Hum Genet. 2013 Sep; 21(9): 1033-1034.

Antenatal testing and diagnosis

How is Down's syndrome diagnosed antenatally?

Screening tests will indicate the individual chance of the baby having Down's syndrome, but a definite diagnosis is reliant on an invasive procedure.

Screening tests

The combined test

Combined testing is offered in the first trimester of pregnancy, between 10 weeks, 0 days and 14 weeks, 1 day. The combined test involves a blood test which measures 2 proteins associated with pregnancy: beta-human chorionic gonadotropin (beta-hCG) and pregnancy-associated plasma protein-A (PAPP-A). In addition, an ultrasound scan will be used to measure CRL (crown rump length) and nuchal translucency (NT).

If women choose to be screened for Down's syndrome, the dating scan and NT scan can be carried out at the same time (between 11 weeks, 2 days and 14 weeks, 1 day). The sonographer will measure the thickness of the NT at the back of the baby's neck. Information from the blood test combined with maternal age, the CRL and NT measurement will be used to work out an individual chance* factor of the baby having Down's syndrome.

Nuchal translucency (NT) over 3.5mm

The NT is the small space at the back of the baby's neck that is measured if the sonographer thinks that it looks large, or if the woman has requested screening for Down's, syndrome Edwards syndrome and Patau's syndrome.

Most babies where the NT measures over 3.5mm will have no other problems, however it is more likely that these babies could have certain issues that the health professional will discuss with the woman. One of these is that the baby is slightly more likely than most to have a baby with a chromosomal condition.

A woman may be offered a chorionic villus sampling (CVS) or amniocentesis in order to assess whether the baby is affected.

The quadruple test

If it has not been possible to offer women the combined screening in early pregnancy, then they will be given the opportunity to have the quadruple blood test. This test measures 4 proteins associated with pregnancy (all types of hCG, unconjugated oestriol, alphafetoprotein and inhibin A) and is performed between 14 weeks, 2 days and 20 weeks, 0 days. This information combined with maternal age will be used to work out the chance* of the baby having Down's syndrome.

Non-invasive antenatal test (NIPT)

Where NIPT has been introduced in NHS settings, the NIPT screening test is offered to women if they have had a higher chance* result from combined or quad screening (a result greater than 1 in 150).

NIPT is a further screening test that is more accurate than the combined or quad test. It will not give a definite result. It is a blood test taken in the usual way, as blood will have some of the baby's DNA in it. The blood is sent to the laboratory and tested to look for Down's syndrome, Edwards' syndrome or Patau's syndrome. The result will tell whether there is a low or high chance* that the baby has one of the syndromes. This test will not give a result for any other conditions including the sex of the baby. It takes about 10 days to get the result.

Less than 0.1% (that is less than 1 in 1000) of women with a low chance* result will have a baby with one of the conditions.

If you get a high chance* result for Down's syndrome, Edwards syndrome or Patau's syndrome; or the test cannot give a result, a woman will be offered a diagnostic test. Around 2% of tests will not give a result, often because there is not enough of the baby's cells in the blood. It is obviously a woman's decision as to whether she wants to choose a diagnostic test.

Some women will be unable to have the NIPT:

- if this pregnancy was ever twins
- some medical conditions (including if having had a tumour, transplant or recent blood transfusion).

If a woman with a higher-chanced* combined test result cannot be offered NIPT screening, she would be offered a diagnostic test.

*** We use the term 'chance factor' instead of refer to the 'risk factor' following feedback from parents regarding the negative connotation attached to the term 'risk'.**

Diagnostic tests

If the chance result from a screening test is calculated between 1:2 and 1:150, the mother will be offered either Chorionic Villus Sampling or Amniocentesis – these tests can provide a definite diagnosis. It is entirely a woman's personal decision as to whether she wishes to progress to a diagnostic test and she maybe doing this in order to prepare herself for the birth of her baby. It should never be assumed that a diagnostic test will lead to a decision to terminate the pregnancy.

What is chorionic villus sampling (CVS)?

CVS is a procedure during which a doctor removes a small amount of tissue from the woman's placenta (afterbirth) during pregnancy. The cells in this tissue are tested in the laboratory to look at the baby's chromosomes. A woman can usually have CVS from her 11th week of pregnancy to her 14th week.

CVS can cause a miscarriage in more than 1% but less than 2% of pregnancies (that means 1 to 2 in every 100 women could lose their baby). A miscarriage is most likely to happen up to three weeks after the CVS. No one knows why this happens or who it will happen to. It can happen whether or not the baby has a chromosome condition. CVS is

done early in pregnancy which is when miscarriages are slightly more common in all pregnant women.

Because CVS is a specialised procedure, a woman may not be able to have it done in the maternity unit she usually goes to. Instead, she may be offered an appointment at a different maternity unit.

What is an amniocentesis?

An amniocentesis is a procedure to remove a small amount (about 15 to 20 millilitres) of amniotic fluid from around the baby in the womb. The cells from the baby that are floating in this fluid can be tested in the laboratory to look at the chromosomes. An amniocentesis is usually done after 15 weeks.

An amniocentesis can cause a miscarriage in 1% of pregnancies (that means one in every 100 women will lose their baby). A miscarriage is most likely to happen up to three weeks after the amniocentesis. No one knows why this happens or who it will happen to. It can happen whether or not the baby has a chromosome condition.

Two types of test are used to look at the baby's chromosomes:

- A 'polymerase chain reaction' (or PCR)
The results are available after three working days.
- A 'full karyotype'
The results are usually ready within two to three weeks.

A full karyotype test is usually only requested if there is a history of chromosomal abnormalities or if a scan has indicated an increased chance of having a chromosomal or genetic disorder³.

³ Screening tests for you and your baby. NHS leaflet – download for free from: www.screening.nhs.uk

Offering antenatal testing for Down's syndrome

Clinical guidelines

All pregnant women in England, Wales and Scotland are offered antenatal screening. It is important that this option is presented and discussed in a non-directive manner at first contact, as this will prepare women and their partners for the choices they may need to make at a later date.

All women should be offered screening for Down's syndrome. Women should know that it is their choice to embark on screening for Down's syndrome. (NICE 62 - 1.7.2.1)

The NICE clinical guideline 62⁴ states that information about Down's syndrome and screening for Down's syndrome should be provided to women at first contact, prior to proceeding with any screening tests.

Information about screening for Down's syndrome should be given to pregnant women at the first contact with a healthcare professional. This will provide the opportunity for further discussion before embarking on screening.

Specific information should include:

- ***The screening pathway for both screen-positive and screen-negative results***
 - ***The decisions that need to be made at each point along the pathway and their consequences***
 - ***The fact that screening does not provide a definitive diagnosis and a full explanation of the risk score obtained following testing***
 - ***Information about Chorionic Villus Sampling and Amniocentesis***
 - ***Balanced and accurate information about Down's syndrome***
- (NICE 62 - 1.7.2.5)**

The guidelines also state that this should be done in a space that allows for discussion, preceding the booking appointment.

Information about antenatal screening should be provided in a setting where discussion can take place; this may be in a group or on a one-to-one basis. This should be done before the booking appointment. (NICE 62 - 1.1.1.9)

⁴ NICE Clinical Guidelines 62 – accessible from: www.nice.org.uk/guidance/CG62

We recognise that midwives are required to provide pregnant women with a large amount of information during a very short space of time at the first contact appointment. However, in order for women to make an informed choice about antenatal testing, time *must* be given to providing balanced and accurate information about both screening and Down's syndrome. Please be aware that The DSA welcomes contact from pregnant women directly or any health professional supporting them. You can contact our helpline on 0333 12 12 300.

Pregnant women should be informed about the purpose of any test before it is performed. The healthcare professional should ensure the woman has understood this information and has sufficient time to make an informed decision. The right of a woman to accept or decline a test should be made clear.

(NICE 62 - 1.1.1.8)

Information about antenatal screening should include balanced and accurate information about the condition being screened for.

(NICE 62 - 1.1.1.10)

Up to date information about Down's syndrome for healthcare professionals is provided later on in this booklet. There is also information available in the Screening Tests for You and Your Baby (5) leaflet, which should be given to all women at the booking appointment. This leaflet has recently been updated.

Parents have told us that they expect choices they make at any stage during the screening process to be respected by health professionals, which is supported by the NICE guidelines.

Tell it Right survey

The Down's Syndrome Association (DSA) conducted two surveys (in 2009 and repeated in 2014) to identify the antenatal and neonatal experiences of parents of a child with Down's syndrome.

Both surveys revealed that women did not recall being provided with enough information about Down's syndrome during their pregnancy.

I feel more should be discussed about Down's syndrome during early pregnancy with the midwife so women can make an informed choice. I decided not to go for screening because I would not go for an abortion and I know and believe that babies with Down's syndrome can grow into adulthood and lead a full life. However, not every pregnant woman realises that in early pregnancy.
(Tell it Right Survey, 2009)

Initial data from the 2014 survey identified that 67% of respondents reported not being provided with any information about Down's syndrome by their healthcare services prior to screening.

The DSA would like to be as supportive as possible both to healthcare professionals working in maternity services and to pregnant women and their families by finding a way to ensure that information about Down's syndrome is shared effectively.

Upon receiving a high chance result, guidelines state that women should be offered further support in the form of counselling by a trained practitioner.

If a pregnant woman receives a screen-positive result for Down's syndrome, she should have rapid access to appropriate counselling by trained staff. (NICE 62 - 1.7.2.6)

The 2009(7) survey indicated that some mothers felt that there was a need for more support once a diagnosis had been made.

There is all this time etc. put into screening and making it work, but when you have to make a decision based on these results there is just no one there to help you and offer support and advice. Overall, a very lonely and tough experience to go through. (Tell it Right Survey, 2009)

The fact that screening is offered, gives out a message about Down's syndrome, whether we are aware of it or not.

I did not feel at any point like any professional was pushing me either way but I do feel it's such a mixed message.

On one hand you are being told your baby could have Down's syndrome but it's okay, but then you are also being told in the same sentence that you can terminate your child.

My husband did not know much about Down's syndrome and was terrified. He said if we were being offered a termination it must mean that our child would really suffer.

(Tell it Right Survey, 2014)

It is important that parents are given the opportunity to discuss their results (of either screening or diagnostic tests) with an appropriate practitioner who has balanced, up to date and accurate knowledge about Down's syndrome, in order that they can make an informed decision about their pregnancy. A referral to our Information Service at this time might also be helpful 0333 12 12 300 or info@downs-syndrome.org.uk

What parents would like to know about the antenatal testing process

- Parents want to know what each part of the screening options can tell them and what choices they might have to make when presented with new information.
- Parents would like a discussion with their healthcare professionals about how they will receive their test results (e.g. in person, over the telephone, by letter), before choosing screening.
- Throughout the screening pathway parents want balanced, accurate and up to date information about Down's syndrome.
- Parents would like time to absorb the information that they are given, so that they can make an informed decision that is right for them.

Screening tests and results can be confusing. It is important that parents are helped to understand what screening tests can tell them. Sometimes parents call the DSA believing that high chance means that they are definitely going to have a baby with Down's syndrome. Others call the DSA believing that low chance means that they definitely will not have a baby with Down's syndrome.

Giving an antenatal diagnosis

The following advice refers to giving an antenatal diagnosis as a result of diagnostic testing (CVS or amniocentesis). How parents will receive their results should have already been discussed and agreed during the test appointment. If a woman's results come back as positive, we suggest the following tips which have been devised based on clinical guidelines and the feedback given to us by parents.

Our recommendations for giving an antenatal diagnosis:

- Disclosure made by an appropriate staff member (as specified by local guidelines)
- Tell parents as soon as possible and in person, or offer a face-to-face appointment as soon as possible if it has been agreed that a woman will receive the results by phone
- Explain what the results of the diagnostic test mean
- Person-first terminology (a baby with Down's syndrome). Provide balanced, accurate and impartial information about Down's syndrome
- Provide hard copies of written information (not photocopies). The DSA's booklet "*Looking Forward to Your Baby*" could be helpful here. Encourage parents to contact the DSA; have contact details available
- Provide information about the options that are available and discuss these
- Allow time for questions, provide further information and support where appropriate; offer counselling
- Respect and support any decision that the parents make

Postnatal diagnosis



How is Down's syndrome diagnosed postnatally?

The diagnosis of Down's syndrome is usually picked up soon after the birth of the baby because of the way he or she looks. There are a number of physical characteristics associated with the condition which may lead a parent or medical professional to suspect that the baby has Down's syndrome. All babies with Down's syndrome will have a collection of some of these features, which will vary at an individual level; these features are also found in members of the general population.

A chromosome test will need to be done on the baby before a positive diagnosis can be made; the doctor will take a blood sample from the baby and send it for chromosome analysis.

We recognise that in the period shortly after a baby has been born, where you suspect that the baby might have Down's syndrome, there is a period of uncertainty and you may feel unconfident in what you can or should say to parents. Overwhelmingly, parents tell us that what they value during this period is honesty. A parent should understand that, until the results of a test, you cannot say whether or not the baby has Down's syndrome, but it is best to be honest about what you suspect and be open to answering questions from parents during this period. The giving of information at this stage is not detrimental and parents say they would rather this than feel that they are in a period of limbo, where professionals are reluctant to speak openly or answer questions.

Giving a postnatal diagnosis

Some of the guidance already laid out for giving a pre-natal diagnosis is just as relevant to giving a postnatal diagnosis. However, there are obviously some differences in the way in which the diagnosis is given because the baby has arrived.

These are our top tips for giving a postnatal diagnosis:

- Congratulate parents on birth of their new baby
- Be aware of your body language and non-verbal cues
- News should be shared by a consultant Paediatrician with the mother's midwife present (no more than two professionals present)
- Tell parents as soon as possible and ensure the baby is present, interact with baby if at all possible and call the baby by their name (if this has been chosen)
- Ensure partner or family member/friend is there to support mum in a suitable environment (consider privacy)
- Person-first terminology (the baby has Down's syndrome)
- Provide balanced, accurate and impartial information about Down's syndrome, but do not overload with detail. Answer questions, but do not list all the possible conditions a baby with Down's syndrome might have. Should additional discussions be needed about associated health conditions, there will be time to introduce these later on
- Provide hard copies of written information (not photocopies)
- Encourage parents to contact the DSA and their local support group; have contact details available
- Allow time for questions and arrange a follow up within 24 hours
- Encourage parents to enjoy bonding with their new baby and not to think too far ahead, they have a new addition to their family to get to know and this should be their main focus in the days that lie ahead.

If a healthcare practitioner believes that a baby may have Down's syndrome because some of the physical characteristics listed above have been noted, concerns should be raised with a Paediatrician and a course of action discussed before an examination of the new baby takes place. As the healthcare practitioner is likely to have already built up a relationship with the parent, it may be more appropriate for them to explain why the Paediatrician wants to examine the baby. It is best to ensure that this happens as quickly as possible.

Some parents tell the DSA that they already suspected that their baby had Down's syndrome very soon after birth and many parents state a preference for openness and honesty from healthcare professionals; this will need to be judged on an individual basis. Healthcare professionals should be particularly aware of the message that their body language and non-verbal cues might be giving out to parents (e.g. glances between colleagues).

Gather Information

It is important that healthcare professionals have read up to date information about living with Down's syndrome before talking to parents about their baby's condition. This is where the DSA can help; take a look at our website www.downs-syndrome.org.uk or call our helpline 0333 12 12 300. We can send out hard copies of our New Parent information. Parents generally prefer hard copies to photocopies or copies that have been downloaded and printed. Some parents may feel that being given a photocopy or printed copy is second best; that somehow a value judgment is being made about the worth of their new baby. It is useful to have the DSA's contact details to hand and information about local Down's syndrome support groups, made up of local parents of children with Down's syndrome; we can provide details of the closest group. Some groups have a specific new parent contact, although not all new parents will be ready to talk to other parents at this stage. If the baby has a heart condition, the Down's Heart Group is a good source of information and advice. It can also be helpful to have gathered information about other local support services.

Environment matters

It is important to ensure the room where the discussion occurs is suitable; allowing privacy without people coming and going and no phone calls or other interruptions.

Telling the parents

It is really important to tell both parents together whilst the baby is present. If the mother is single or her partner cannot be present, it is good to try and ensure support for the mother through a family member or friend. If English is not the parent's first language, it will be helpful to have an interpreter present.

Receiving unexpected news may mean that new parents need a little more time to process what they are being told. Plenty of time should be allowed for the parents to absorb the information and to ask questions, it is really important not to rush through disclosure. Some parents may have a good or basic understanding of the condition already, perhaps because they already know someone with Down's syndrome, whereas others may not.

If the parents ask a question that the healthcare practitioner does not know the answer to, it is important they are honest and reassure the parents that they will find out. It is best to take the parents' lead in each situation and check along the way that they have understood what they have been told. Healthcare professionals should use their judgment, but it can really help some parents if they interact with the baby and call him or her by their name. Before leaving, a follow up chat within 24 hours should be arranged.

People will react in different ways to the news that their baby has Down's syndrome. Many will experience a complex mixture of love, shock, sadness, fear, anxiety and protectiveness. There is no right way for a new parent to react, it will vary from person to person. Try to take your cue from the new mum / couple. Be ready to support and reassure through the next couple of days when emotions may change quite quickly.

Our daughter was diagnosed shortly after birth. The main Paediatrician on duty over the next few days, although not a specialist in Down's syndrome, was able to give us information and reassurance, and definitely took the time to do so. (New Parent)

What support do parents need in the early days?

The new mum may not feel she needs it, but she should be offered the choice of moving to a single room. It can be helpful to offer the partner somewhere private to make phone calls and the option to stay overnight. If possible, all relevant staff should be encouraged to regularly visit mum and interact with the baby. It is important to talk to the parents about what will happen next to reduce any anxieties that they may have. It can be helpful to parents if you inform their GP, Community Midwife or Health Visitor, as appropriate. It may be useful to signpost materials that are available about the condition, or refer them to the DSA for information and/or resources.

Feeding

It is important to check how well the baby is feeding and give support to breastfeed if needed. As with any new mother, particularly if it is their first child, feeding can sometimes be a source of worry. Almost all mothers, who want to, can breastfeed or provide breast milk for their baby. Sometimes breastfeeding is established easily with no more problems than with any other baby and sometimes breastfeeding can be harder and needs more time, patience and perseverance. Some babies will become better at feeding as they grow older and will be able to be fully breast fed. Some mothers choose not to breastfeed or find that because of their circumstances, breastfeeding is not right for them.

A few babies have major medical problems which affect feeding. Babies with gastrointestinal tract (GI tract) disorders who need an operation will not be allowed to feed at first and will get nutrients intravenously. Babies with severe heart conditions may be unable to feed because they are tired or breathless; mothers of these babies can express breast milk by hand or pump to build up their milk supply. Their milk can be given to their babies by naso-gastric tube when the babies are well enough. With patience and following surgery for any medical disorders, these babies can often fully breastfeed eventually. The DSA's New Parents' Guide contains further information and advice regarding breastfeeding.

Health Checks

It is important that babies with Down's syndrome get checked for any associated health conditions that may or may not be present. The Down's Syndrome Medical Interest Group (DSMIG) has some useful guidance. <https://www.dsmig.org.uk/wp-content/uploads/2018/09/Neonatal-Guidelines-amended-7-1-18-NB-edit-26.9.18-new-date.pdf>

Leaving hospital

Ensure that parents have all the contact information that they need for the future. If they do not already have a copy, tell the parents about the Personal Child Health Record (PCHR) insert for babies born with Down syndrome. Babies with Down's syndrome can take longer to regain their birth weight; the weight and growth charts in the PCHR insert will help to capture a more accurate record of how well the new baby is progressing. The PCHR insert also contains the Down Syndrome Medical Interest Group (DSMIG) schedule of minimum basic health checks (see page 16 table 1) for parents and professionals to refer to.

An example of good practice:

When Sam was born he didn't feed very well and was jaundiced and I got the feeling that something wasn't right, a sense of they all knew but weren't saying anything. After 3 days the consultant popped his head round the door and asked us to come to another room, he said 'bring Sam with you'. My midwife was in his office and they very gently told us that Sam has Down's syndrome, the consultant was cuddling Sam at that point. They had some leaflets for us to read, had been in touch with the specialist at the Child Development Centre and arranged for her to come and see us and they'd phoned the DSA about us meeting up with other parents. (New Parent)

Like any other baby, some babies with Down's syndrome may have additional health problems which will require a period in the Neonatal Intensive Care Unit (NICU).

Common questions that parents ask



Do people with Down's syndrome have medical problems?

Certain medical problems are more common in people with Down's syndrome. However, with advances and increased access to medical care most of these problems are treatable.

None of these problems are unique to people with Down's syndrome, as they also appear in the rest of the population. It is also important to remember that some people with Down's syndrome do not experience any health problems.

Medical problems which are more common in people with Down's syndrome:

- Almost half of all babies born with Down's syndrome will have heart problems, some of which may require heart surgery
- Hearing problems
- Sight problems
- Thyroid disorder
- A poorer immune system
- Respiratory problems, coughs and colds
- Obstructed gastrointestinal tract

This table contains the recommended routine health checks for babies with Down's syndrome following birth.

	Birth – 6 weeks	Special checks under 2 years	Preschool checks	School age
Thyroid blood tests	Newborn: routine heel prick – blood spot test	From age 1 year thyroid function should be discussed annually using the results of either: <ul style="list-style-type: none"> • Annual fingerpick TSH test or • 2 yearly thyroid blood tests, including thyroid antibodies 		
Eye checks	New-born: routine check including congenital cataract check	Age 18-24 months: Formal eye and vision examination including check for squint and refraction for long or short sight	Age 4 years: Formal eye and vision examination including check for squint, refraction and assessment of near and distant vision and visual acuity	Repeat vision test every 2 years or more frequently if recommended by optometrist or ophthalmologist
	Visual behaviour to be monitored at every review particularly in the first year			
Hearing checks	Universal new-born hearing screen	Full audiological review by 10 months including test and impedance check	Annual audiological review as before	2 yearly audiological review or more frequently if recommended
Growth monitoring	Length, weight and head circumference should be checked frequently and plotted on Down's syndrome growth charts		Height and weight should be checked and plotted on Down's syndrome growth charts at least annually while growing. (BMI checked if concern regarding overweight)	
Heart checks	By age 6 weeks formal heart assessment including Echocardiogram	At all ages low threshold for reviewing heart status if signs or symptoms develop		From adolescence onwards as part of routine health checks listen to heart for signs of acquired heart disease
Breathing checks	Enquire at every review for uneven breathing during sleep and poor quality sleep. Low threshold for further testing using sleep studies.			
Blood checks	New-born blood test to check abnormal blood film*	If blood film is abnormal in first 6 weeks, follow up or repeat blood testing may be necessary until age 5		

* A new test has been developed at the University of Oxford to detect early signs of a potentially fatal condition that can also develop into full blown leukaemia in children with Down's syndrome. Early intervention greatly increases chances of survival for children who develop symptoms and new guidelines for doctors recommend that all children with Down's syndrome receive a blood test within three days of birth to identify if they are at risk.

The new British Society for Haematology (BSH) guidelines, published in the British Journal of Haematology, are a result of years of research.

The new guidelines recommend that a full blood count, which measures levels of different types of blood cell in the blood, is taken within three days of birth in children with Down's syndrome. If blood tests reveal that there are high levels of abnormal cells in the bloodstream and there are physical symptoms, babies should be given a test, developed by the Oxford group, to screen for mutations to a gene known as 'GATA1'. GATA1 gene mutations are present in both TL-DS and AML in children with Down's syndrome.

Children with TL-DS who do not experience life-threatening symptoms should be monitored for a number of years to check for any signs of progression into leukaemia, so that treatment can start immediately if it does.

How does Down's syndrome affect development?

All people with Down's syndrome will have some degree of learning disability. This will affect a child's ability to learn compared with other children of their age; it does not mean they cannot learn. The range of ability is wide and varies amongst individuals. Most children with Down's syndrome will learn to walk, talk and meet other developmental milestones, but often later than their peers. Early intervention programmes, which are now widespread for children with learning disabilities, can help in all areas of child development. These programmes tend to include speech and physical therapy as well as home teaching programmes for the child and family. Children and adults with Down's syndrome can and do continue to learn throughout their lives just like the rest of the population. The Down's Syndrome Association has lots of resources and information we can share with new families which should help them.

Just as with all children, there is a great deal of individual variation in the ages at which different skills develop. The PCHR insert for babies born with Down syndrome contains an outline of the usual progress of children with Down's syndrome for some milestones, which is also accessible via the DSMIG website. A few children will have additional health problems which may slow their development, however all will continue to develop at their own pace. Most children will learn to communicate and learn to read and write although some children will have more complex needs, such as a dual diagnosis with another condition – e.g. Autistic Spectrum Condition (ASC).

Children with Down's syndrome have a specific learning profile and there are really effective teaching methods and strategies which build on their learning strengths. Nowadays, around 80% of children attend mainstream primary school with increasing numbers continuing their education within a mainstream secondary setting.

Further information about health and development can be found in the Early Support Booklet (12) for parents with children who have Down's syndrome.

Should I be doing anything special or different now?

In the early days it's important that parents just get to know and love their new baby. First and foremost, they have a baby with the same needs as any other baby. Early intervention services (e.g. Speech & Language Therapy, Physiotherapy, Portage) are important in supporting the development of children with Down's syndrome so referrals for the family will need to be made either by the Health Visitor, Paediatrician or GP. However, parents need to be reassured that their baby will not miss out if services are not involved immediately; most are in place by about 10 months.

What is life like now for people with Down's syndrome?

In the past it was believed that there were many things that people with Down's syndrome could not do when, in fact, they had never been given the opportunity to try. Today these opportunities have never been greater with many people with Down's syndrome leading rich and varied lives.

Young people and adults with Down's syndrome

Many young people with Down's syndrome and their families have greater aspirations about the lifestyles they want to lead including living as independently as possible, having a job they like, enjoying relationships and marriage, having a wide circle of friends and being able to access all the same social and leisure facilities as other people.

Advances and increased access to medical care have also meant that people with Down's syndrome are living much longer. People with Down's syndrome are now living into their 60s, with a growing number living even longer.

People with Down's syndrome can have a good quality of life and research has shown that the vast majority say they enjoy their lives.

A study evaluated surveys from 2,044 parents or guardians.

- 99 % of parent/guardians said they loved their child with Down syndrome
- 79 % felt their outlook on life was more positive because of their child

A second study evaluated responses to similar questions from 822 brothers and sisters age 9 and older.

- 94 % expressed feelings of pride about their sibling
- 88 % said they felt they were better people because of their sibling with Down syndrome

A third study evaluated survey responses from 284 people with Down syndrome. Their average age was 23. The responses were that:

- 99 % said they were happy with their lives
- 97 % liked who they are
- 96 % liked how they look
- 86 % indicated they could make friends easily

Reference: Skotko BG, Levine SP, Goldstein R. 2011. Self-perceptions from people with Down syndrome. *Am J Med Genet Part A* 155:2360–2369

Who we are

The Down's Syndrome Association (DSA) is the only charity in England, Wales and Northern Ireland focusing solely on all aspects of living successfully with Down's syndrome. Our aim is to help people with Down's syndrome live full and rewarding lives.

Established in 1970 the DSA has evolved from a parent member organisation into a leading national charity with a network of affiliated support groups across the country

Information and training

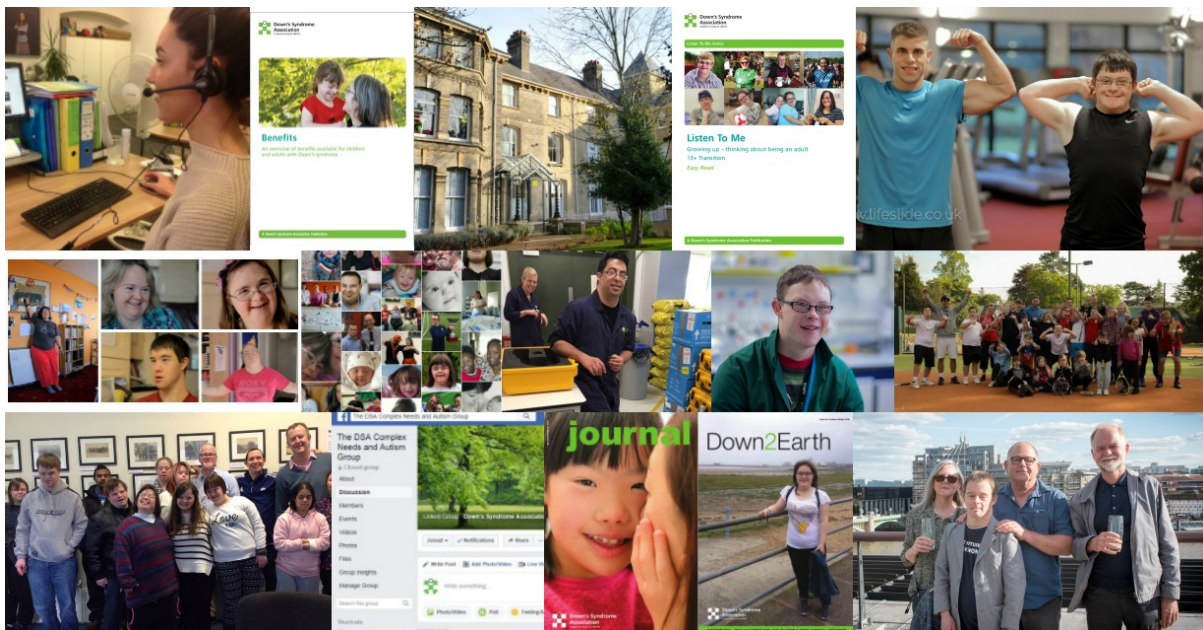
- Increase awareness and understanding of Down's syndrome through our Helpline, training, publications and resources, our website and social media.
- Provide support and advice to individuals with Down's syndrome, their friends, family and carers and the professionals who work with them.
- We offer training that will benefit people with Down's syndrome throughout their lives: Tell it Right® Start It Right for the time of diagnosis; education and development training to promote learning opportunities; and training for carers who support people who are ageing.

DSActive

Working with professional clubs, DSActive helps children and adults with Down's syndrome get active and healthy in a supportive and inclusive environment.

WorkFit

WorkFit is our tailored service dedicated to helping people with Down's syndrome to find appropriate and meaningful work opportunities. We provide bespoke training and ongoing support for employers. We find the right employment opportunities, for the right people.



Reviewed and updated 2018

The Down's Syndrome Association provides information and support on all aspects of living with Down's syndrome.

We also work to champion the rights of people with Down's syndrome, by campaigning for change and challenging discrimination.

A wide range of Down's Syndrome Association publications can be downloaded free of charge from our website.

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