iHV Institute of Health Visiting
Excellence in Practice

Supporting babies and children with Down syndrome and their families

Babies and children with Down syndrome have a genetic variation. Whilst they share some common features, they are all unique individuals with their own abilities, needs, interests and aspirations. Down syndrome is not a disease and people who have Down syndrome are not ill.

This Good Practice Point was co-developed and informed by a panel of experts by experience. The iHV values and acknowledges the expertise and important contribution of all those involved.

Please read this Good Practice Point in conjunction with the Good Practice Point on Supporting Children with Down syndrome: growth, feeding and development - https://bit.ly/3rfL5po

What families with lived experience say:

"Down syndrome is 1/47th of our daughter, please treat it as such."

"I wish I could go back to that scared 24 year old version of me and tell her not to worry, that life would be just fine, she was going to learn a lot, and at times it would be tough, but in general just fine, and actually more joy and more laughter than she would otherwise have ever known."

"Being able to turn to my health visitor for information & support when we were coming to terms with our newborn son's diagnosis helped us to make sense of the new world we had suddenly been thrust into. To know that there is an up to date and accurate resource available to all health visitors, empowering them with good practice points on Down syndrome fills me with hope that other new parents will be well supported as they navigate their journey through parenthood."

People with Down syndrome* have a genetic difference. This is most frequently an extra copy of chromosome 21 in all the cells in their body (95% of occurrences)[1].

Prevalence: There are currently estimated to be around 47,000 people living in the UK with Down syndrome[2]. Data from the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) gives an estimated Down syndrome incidence rate for England of 26.5 per 10,000 total births or one in every 377 births[1]. The chance of having a baby with Down syndrome increases as parents get older, but anyone can have a baby with this genetic variation[3].

Prenatal screening and pregnancy care: There are different types of testing for Down syndrome in pregnancy. Screening tests include the combined test, the quadruple test, and non-invasive prenatal test (NIPT). Screening tests provide a probability of the baby having Down syndrome, but there can still be false positives, and false negatives. An easy read guide is available here[4]: https://bit.ly/4300ARK

Short videos on diagnostic tests and making decisions about them are available here: https://bit.ly/2OaywIr

When a parent receives a higher-chance result from a screening test, they will be offered the following:

- not to have any further testing,
- to have a second screening test (NIPT) which can give a more accurate screening result and help parents to decide whether to have a diagnostic test or not,
- to have a diagnostic test, such as amniocentesis or chorionic villus sampling (CVS) – these will provide certainty whether a baby has Down syndrome or not. In rare cases these tests can cause a miscarriage[4].

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The information in this GPP version was reviewed on 23/02/2024.

All parents are different and will respond to the diagnosis in an individual way. It is therefore extremely important for health visitors to take the lead from the parents. When parents are told that their baby has Down syndrome it can come as a shock, and there can be a process of grief and mourning for the baby that they were expecting to have. Equally, some parents may not experience this.

Instead of guessing how a parent might be feeling, ask them how they are feeling. Do not offer unsolicited advice, instead follow the parents' lead. Ask open questions like, *"Do you have any questions? Is there any information that would be helpful for you now?"*

Offer resources and information in response to their questions and concerns. Some families choose not to have testing. This is really important to acknowledge, respect their decision and discuss if they wish to.

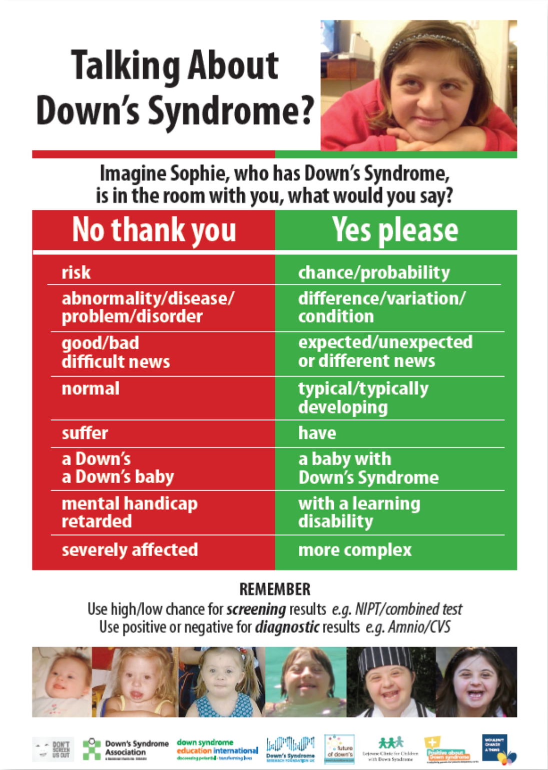
At some point during the screening/diagnosis pathway, expectant parents may find it helpful to talk to parents of children with Down syndrome.

Down's Syndrome Association guide to management of pregnancy[6]: https://bit.ly/3NnZEB3

Language

The language that is used matters as it can imply positive or negative attitudes simply by the words chosen. These attitudes then influence perceptions of whatever is being discussed. When people with Down syndrome and their families hear the syndrome being referred to as a 'risk' or 'abnormal' it upsets them, when they hear their child referred to as "a Down's boy" it sends the subtle message that people don't understand their world. It is so important that we show people through our words that we are all more than our diagnoses and do not portray people with Down syndrome as less than, or undesirable[5].

"Talking About Down's Syndrome?" is a free language resource which has been developed by a parent, Colette Lloyd[5]. This resource clearly highlights the terminology that is preferable, assisting everyone in non-offensive language.



Talking About Down's Syndrome?

Imagine Sophie, who has Down's Syndrome, is in the room with you, what would you say?

No thank you	Yes please
risk	chance/probability
abnormality/disease/ problem/disorder	difference/variation/ condition
good/bad difficult news	expected/unexpected or different news
normal	typical/typically developing
suffer	have
a Down's a Down's baby	a baby with Down's Syndrome
mental handicap retarded	with a learning disability
severely affected	more complex

REMEMBER

Use high/low chance for ***screening*** results *e.g. NIPT/combined test*
Use positive or negative for ***diagnostic*** results *e.g. Amnio/CVS*

Resources to support parents' transition to parenthood with "Different News"

- Looking Forward to Your Baby by the Down's Syndrome Association: https://bit.ly/3NnZEB3
- Dear Future Mom: an encouraging video made in response to a letter from a mother-to-be saying that she was scared of what the future held- https://bit.ly/2s2iMyl
- Down's Syndrome Research Foundation UK DSRF New Parents: https://bit.ly/2OyfXgj
- Looking Up Books: https://bit.ly/2rdBrXO
- Wide variety of stories from Positive About Down Syndrome's campaign #NobodyToldMe about parenting a child with Down's syndrome: http://bit.ly/34eMLBm

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Health

Some health issues are more common for people with Down syndrome, therefore there are more health and development checks for babies and children with this condition than for a typically developing baby[5]. These include heart, digestive, thyroid, hearing and vision checks[7]. Most health problems can be treated successfully with some, like acute myeloid leukaemia, having better outcomes than in the general population. People with Down syndrome also have lower incidence of solid organ tumours, hypertension and stroke. Life expectancy of people with Downs syndrome has increased dramatically over the past century (currently around 60 years)[1], mainly due to improvements in access to cardiac surgery and better treatment of respiratory infections.

Babies and children with Down syndrome will benefit from the same universal health visiting service and advice on ways to stay healthy as any other family (for example, healthy eating, vitamins and keeping active).

Good Practice Points for Health Visitors

The Down Syndrome Act 2022 became law in April 2022[2]. The act represents a significant opportunity to improve the life outcomes of people with Down syndrome, and to raise the understanding and awareness of the specific needs of people with Down syndrome.

Everyone with Down syndrome should receive early support to meet their needs, and families and carers should receive the help they need to enable them to guide their child's development.

iHV's expert panel has offered the following advice to support health visitors to meet the needs of babies and children with Down syndrome and their families:

- Offer hope, congratulate them on the birth of their baby.
- Concentrate on the short-term relevant issues, unless the family ask for long term. Handling things day by day is often all a family can manage at this stage.
- Recognise that every family is different and will deal with issues in different ways.
- Acknowledge concerns and respond to questions, including those that might appear unimportant or trivial – this does not mean that they are unimportant to the family asking them.
- Continuity of health visitor is important – families value "being known" and receiving personalised care.
- Don't forget to include all the usual universal health visiting support and advice – sometimes these can get overlooked (for example, safer sleep advice, accident prevention, oral health, immunisations, etc).
- Make sure you are familiar with the local offer for children with Special Educational Needs and Disabilities (SEND) and support parents to access local resources and groups that suit their individual needs and preferences.

Newborn examinations and checks

All babies with Down syndrome will receive the routine Newborn and Infant Physical Examination (NIPE) programme[8] which screens all newborn babies within 72 hours of birth, and then once again between 6 and 8 weeks for for the following conditions:

- congenital heart disease
- developmental dysplasia of the hip
- congenital cataracts
- cryptorchidism (undescended testes)

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Down syndrome - suggested schedule of health and development checks

The following are suggested ages for health and development checks. Check at any other time if there are parental or other concerns.

	Birth - 6 weeks	Special checks under 2 years	Preschool checks	School age
Thyroid blood test	**Newborn** routine heel prick-blood spot test	**From age 1 year** thyroid function should be discussed annually using results of either: • Annual fingerprick TSH test OR • Yearly thyroid blood tests, including thyroid antibodies		
Eye checks	**Newborn** routine check including congential cataract check	**Age 18-24 months:** Formal eye & vision examination including check for squint, and refraction for long or short sight	**Age 4 years**: Formal eye & vision examination including checks for squint. Refraction & assessment of near & distant vision & visual acuity	Repeat vision test **every year**, 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 **newborn** hearing screen	Full audiologist review by **10 months** including hearing test & impedance check	**Annual** audiological review as before	**2 year** audiological review or more frequently if recommended
Growth monitoring	Length, weight & head circumference should be checked frequently & plotted on Down syndrome growth chart		Height & weight should be checked & plotted on Down 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 & poor quality sleep. Low threshold for further testing using sleep studies			
Blood checks	**Newborn** blood test to check for abnormal blood film	If blood film is abnormal in first 6 weeks, follow up or repeat blood testing may be necessary under age 5		

Based on detailed recommendations for Medical Surveillance Essential for children with Down syndrome[7] which can be found at www.dsmig.org.uk

Further reading:
Emond A. Editor. Health for all Children. 5th ed. U.K.: Oxford University Press; 2019.

*In this Good Practice Point the words Down's syndrome and Down syndrome are used interchangeably depending on the use of terms within the research or organisation that is cited.

References

1. NHS England. NCARDRS Congenital Anomaly Official Statistics Report, 2020. 2.3 Prevalence of babies with Down's Syndrome, Edwards' syndrome and Patau's syndrome. 2022 [Accessed 23/02/24]. Available from: https://bit.ly/49GM0m9
2. Department of Health and Social Care. Down Syndrome Act 2022 guidance: call for evidence. 2022 [Accessed 23/02/24]. Available from: https://bit.ly/4bPqrRY
3. NHS (2023) What is Down's syndrome? Down's syndrome. https://bit.ly/49AdIRf
4. NHS. Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome. 2021 [Accessed 23/02/24]. Available from: https://bit.ly/4bL07IS
5. Lloyd C. Talking about Down's syndrome? Down's Syndrome Research Foundation UK. 2019 [Accessed 23/02/24]. Available from: https://bit.ly/3wlYQrB
6. Down's Syndrome Association. Management of Pregnancy, Information for Health Professionals. Management of Pregnancy when Down's syndrome has been diagnosed. 2019 [Accessed 23/02/24]. Available from: https://bit.ly/3wqnW8P
7. The Down Syndrome Medical Interest Group. Guidance for Essential Medical Surveillance. 2020 [Accessed 23/02/24]. Available from: https://bit.ly/49pp37g
8. GOV.UK. Guidance. Newborn and infant physical examination (NIPE) screening: programme overview. 2013 [Accessed 23/02/24]. Available from: https://bit.ly/3wsBNvg

This iHV Good Practice Point was last reviewed by the iHV team with minor updates in 2024.
With credit to the original GPP authors: "experts by experience": Colette Lloyd, Angie Emrys-Jones and Pippa Cusack; and health visiting advisor Karen Afford, first produced in 2019.