Your baby!

Tests offered

what you need to know

Newborn Screening **Tests**

NHS SCOTLAND

Public Health Scotland

This booklet explains what conditions can be tested for and what the tests involve.

The newborn screening tests offered to all babies in the first few days and weeks of their life aim to ensure conditions are identified and treatment is started as soon as possible.

All screening and diagnostic tests in this booklet are provided free in Scotland by the NHS.

It's your choice whether you accept all or just some of the newborn screening tests we offer for your baby. You can also choose not to have your baby screened at all. But it's important that you understand the possible outcomes if you choose not to have the tests.

You should have a more detailed talk with your health professional, who will explain each test, the benefits and any drawbacks.

But it's important that you understand the possible outcomes if you choose not to have the tests.

We talk about 'your health professional' throughout this booklet – this can be a health visitor, paediatrician, audiologist, or any other specialist responsible for the screening tests.

Remember, you should discuss all of the newborn screening tests with your midwife or health visitor.

At a glance

Hearing screen

At a glance ...

- The hearing screen is a simple test done within the first few weeks of life, maybe even before you leave the maternity unit.
- If your baby's hearing has not been checked within four weeks of getting home from hospital, please ask your health visitor or GP to make an appointment.
- Hearing problems can develop at any time.
- If you have any worries about your baby's hearing in the future, talk to your health visitor or GP. They can arrange for a hearing assessment at any age.

Why is the hearing screen done?

Most babies who have a hearing loss are born into families that have not experienced hearing loss before. Finding out early is important for your baby's development and means that support and information can be offered from an early age.

Even if you can see that your baby is responding to sounds, it's still very important to have the hearing screen. Babies with hearing loss can still respond to some sounds.

What happens during the hearing screen?

The hearing screen is carried out by a trained health professional and can be done in one of two ways:

a small, soft earpiece will be placed in the outer part of your baby's ear

or

three small sensors will be placed on your baby's head and neck, and a small, soft earpiece or headphone will be put in or over your baby's ear.

Your baby may have both tests. A computer will then measure how well your baby's ears respond to clicking sounds.

Does the screen hurt?

No, it does not hurt and is not uncomfortable. It's very quick and often takes place while your baby is asleep.

What does the screen look for?

The screen looks for **a clear response from both ears**.

If the hearing screen shows a clear response from both ears:

- your baby is unlikely to have a hearing loss
- you will be given a list of the sounds your baby should respond to as he or she grows older.

If the hearing screen does not show a clear response from one or both ears:

- your baby may have a hearing loss.

Other reasons your baby may not show a clear response are:

- your baby may have had fluid or a temporary blockage in the ears after birth
- your baby may have been unsettled during the screen
- there may have been background noise.

An appointment will be made for you to see a hearing specialist at the audiology department for further assessment.

When will I get the results?

You'll usually get the results as soon as the screen is finished. If you have any worries or questions, speak to your health professional.

What will happen if my baby does not have the hearing screen?

If your baby has hearing loss and it's not detected early, this can affect their development. If it's detected early, they can get the early support, advice and treatment they need.

Facts

Two to three babies in every 100 screened do not show a clear response on hearing screening.

One to two babies in every 1,000 have a hearing loss in one or both ears.

It's very important to attend the audiology appointment if ...

the hearing screen does not show a clear response from one or both of your baby's ears. This is to confirm if he or she has a hearing loss.

Blood spot test

At a glance ...

- Some serious conditions cannot be seen by just examining the baby, but can be picked up through a blood test.
- All newborn babies in Scotland will be offered a blood spot test. Your midwife will explain the test in detail, and ask for your permission to carry it out.
- This one test enables nine conditions to be screened for – all from just a few drops of blood taken from your baby's heel.

How is the blood spot test done?

It's usually carried out around five days after the baby is born. The midwife pricks the baby's heel to get a few drops of blood. The blood is then put onto a card and sent for testing.

We may need to repeat the test if:

- there was not enough blood on the card for testing
- the card was damaged
- one of the results was unclear.

What is it tested for?

The blood spot test screens for nine inherited conditions.

If you, your baby's father or a family member already has one of these conditions (or a family history of it), please tell your health professional straight away.

Inherited conditions are usually passed from parents to children through **altered genes**.

What are ...?

Genes

Genes are the factors that determine your characteristics, from the colour of your hair to your blood group. For most of our characteristics, we inherit one gene from our mother and one from our father.

Babies only have the conditions if they inherit **two** altered genes – one from their mother and one from their father. Babies who inherit just one altered gene are known as **carriers**.

Carriers

Carriers do not have the inherited condition, but may pass the altered gene on to the next generation. If your baby is identified as a carrier, your health professional will give you more information and offer you an appointment with a genetic counsellor to discuss what this means for you, your baby and your family.

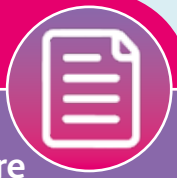
The blood sample will be tested for the following serious but treatable conditions.

Sickle cell disease

Sickle cell disease affects about one in 2,500 babies born in the UK. It's passed from parents to children through altered haemoglobin genes. Haemoglobin is the part of the blood that carries oxygen around the body.

The blood cells change to a 'sickle' (or hook) shape and get stuck in the small blood vessels. This can cause pain and damage to the baby or serious infection. In some cases, it causes death.

Facts



The most likely form of sickle cell disease is sickle cell anaemia, but there are other forms. All forms are screened for by the test.

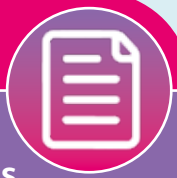
What if the test shows sickle cell disease?

Your health professional will arrange for you to see a haematologist (a doctor who specialises in blood conditions). Your baby will be started on treatment, including antibiotics, to prevent serious illness.

What would happen if my baby is not screened for sickle cell disease?

If your baby has sickle cell disease that is not found, he or she would develop pain and could suffer serious infection and damage to the body. This could be lessened or even avoided with the right treatment.

Facts



The blood spot test looks for sickle cell disease, but other blood problems can be found. If this happens, your health professional will arrange for you to see a haematologist, who will organise more tests for your baby.

CF (cystic fibrosis)

CF affects many parts of the body. The lungs and the pancreas are most affected, leading to regular chest infections and problems digesting food and absorbing the nutrients your baby needs. One in every 2,500 babies born in Scotland has the condition. Early diagnosis and treatment can lead to better health for children with this condition.

CF occurs when a baby inherits an altered gene from each parent. Both parents carry one altered gene, so are unaffected – a person needs to have both genes to have the condition. One in 25 people in Scotland is a CF carrier.

If the blood spot test for CF is not clear, you might be asked to have another sample taken between 21 and 28 days of age.

What if CF is suspected?

An additional test using DNA from the same sample will be carried out.

Your health professional will arrange for you to see a CF specialist, who will confirm the diagnosis and make sure your baby gets the treatment needed. The CF specialist will also give you information, advice and support.

Rarely, cases of CF in babies are not picked up by the screening test.

Some babies are unaffected by the condition but, like their parents, may be healthy carriers of the altered gene. These babies will not need any treatment, but may pass the gene to the next generation.

If you're worried about your baby's DNA being used for testing, please discuss it with your health professional.

What would happen if my baby is not screened for CF?

If your baby has CF that is not detected, there may be a delay in diagnosis. The baby might develop problems that could have been reduced or even avoided with early treatment.

Your health professional will give you more information and offer you an appointment with a genetic counsellor.

CHT (congenital hypothyroidism)

Babies with CHT do not produce enough of the hormone thyroxine, which is needed for healthy mental and physical development.

CHT is rare, affecting about one in every 3,500 babies born in Scotland.

What if the test shows my baby might have CHT?

Your health professional will arrange for you to see a specialist doctor for children, who will do some more tests.

Facts

CHT is easily corrected by giving your baby thyroxine as a medicine.

What would happen if my baby is not screened for CHT?

A baby with CHT who is not treated would grow at a slower rate than other babies, have severe learning difficulties and may die.

Inherited metabolic disorders

Inherited metabolic disorders mean that certain substances in food cannot be broken down in the baby's body.

The blood sample will test for whether your baby has any of the following six inherited metabolic disorders:

- PKU (phenylketonuria)
- MCADD (medium-chain acyl-CoA dehydrogenase deficiency)
- maple syrup urine disease
- IVA (isovaleric acidaemia)
- GA1 (glutaric aciduria type 1)
- HCU (homocystinuria).

Babies with MCADD deficiencies may have problems breaking down fat to make energy under certain circumstances.

Babies born with five of the six disorders find it harder to break down **certain amino acids**.

Amino acids

Amino acids are fundamental parts of all foods except pure fat and sugar.

When we eat foods containing protein, including when babies drink milk, we break the protein down into individual amino acids. Most of these are used to make new body proteins. Any leftover amino acids have to be broken down more and used for energy or removed as waste.

For babies with inherited metabolic disorders, certain leftover amino acids can build up in their blood and other organs, such as the brain, and cause serious problems.

What are the effects?

These disorders all carry a very high risk of delayed development and irreversible learning disabilities, which can be severe if they're not detected early. Some of these disorders can also cause life-threatening illness, even coma in certain circumstances, and they can be associated with other medical problems. Without treatment, babies can become suddenly and seriously ill.

What happens if the test shows my baby might have an inherited metabolic disorder?

Remember you can speak to your health professional at any time for information and support.

A member of the specialist inherited metabolic disorders team for children (a doctor, nurse or dietitian) will contact you by telephone and arrange to meet you and your baby that day. The team will arrange further tests and start any necessary treatment right away. They will be able to explain everything to you and answer all of your questions.

Facts

If they are detected early, effective treatment is available to prevent the ill effects of all six inherited metabolic disorders. This may include a carefully managed diet and/or medicines.

Treatment will need to continue for life, but it will allow your child to remain healthy and lead a full and active life.

What would happen if my baby is not screened for inherited metabolic disorders?

If the problem is not spotted, your baby will almost certainly develop irreversible learning disabilities, and by the time you

realise this it will be too late. For some of the disorders, your child can also become suddenly or seriously ill or may even die.

Due to technicalities of the test, it's only possible to test for all six disorders at once, it is not possible to test for some of the inherited metabolic disorders and not the others.

How do I get the results?

Most blood spot tests show no health problems. If that's the case, the results are sent to your health visitor, who will discuss them with you at the next routine check. If a possible health problem is found, we'll contact you directly.

What if I decide not to have the blood spot screening?

If you decide not to have your baby tested for one, some or all of the conditions, you'll be asked to sign a form confirming that the reasons for testing have been explained to you, and that you understand the possible effects of your baby not being screened. Please note it's only possible to test for all six inherited metabolic disorders at once.

Newborn physical examination

At a glance ...

- In addition to the screening tests, all babies are examined carefully within the first three days of life so that any obvious physical problems can be picked up as soon as possible.
- Usually, nothing is found that you need to worry about. If the health professional finds a problem, your baby will have further assessments and tests.
- The health professional carrying out the examination will give you the results straight away. If a referral for further assessment is needed, this will be discussed with you at the time of the examination.

What happens to my baby's information and blood samples?

We keep a record of your baby's personal screening information, including test results. Your baby's personal health information will be kept private, which means it is only shared with other staff involved in your baby's care. We regularly review what we do to make sure we offer the best service possible.

After being looked at, any of your baby's leftover blood samples will be stored in the laboratory for at least 12 months. The stored blood spots can be used to test for some conditions that are not part of the standard screening programme. This may be useful if your baby becomes ill and the doctor wants further tests, but **it would always be discussed with you first**.

Your baby's leftover blood samples may also be used for research, education and training. If this happens we will remove your baby's personal details. If we ever need to use samples that are not anonymous, **we will always ask for your consent before your baby's samples are used in this way**.

You have rights in relation to the access to, and use of, your baby's personal health information. For more information contact the NHS inform helpline free on **0800 22 44 88** (textphone **18001 0800 22 44 88**) or visit **www.nhsinform.scot/confidentiality** and **www.nhsinform.scot/data-protection**

If you do not want your baby's card to be stored after the 12-month testing period, or to be used for research purposes, please ask your midwife to record your preferences in the comments box on the request form.

More information

Please talk to your health professional if you have any questions or worries. You might also find the following contacts useful.

Metabolic Support UK

The leading patient organisation for inherited metabolic disorders, which supports thousands of patients worldwide, and works on behalf of affected children, young people and families.

Phone: 0845 241 2173

www.metabolicsupportuk.org

National Deaf Children's Society Scotland

Provides information and support for deaf children and their families.

Phone: 0808 800 8880 or 0141 354 7850

Textphone: 0141 332 6133

www.ndcs.org.uk/about-us/where-we-work/scotland/contact-us-scotland/

Cystic Fibrosis Trust

Works to improve the lives of people with CF, raise the profile of CF and fund research.

Phone: 0300 373 1000

www.cysticfibrosis.org.uk

National Society for Phenylketonuria (NSPKU)

Offers support for people with PKU, their families and carers.

Phone: 0303 040 1090

www.nspku.org

Healthtalk

Offers a directory of personal experiences aimed at patients, carers, family and friends, doctors, nurses and other health professionals.

www.healthtalk.org

British Thyroid Foundation

A patient-led charitable organisation dedicated to helping those with thyroid disorders.

http://btf-thyroid.org

NHS inform

The helpline is open every day, please check the website for the opening hours. It also provides an interpreting service.

Phone: 0800 22 44 88

Textphone: 18001 0800 22 44 88

www.nhsinform.scot/screening

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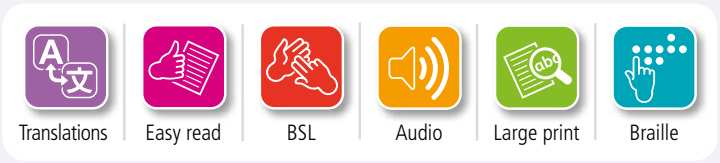

Other formats available at:

www.nhsinform.scot/newbornscreening

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