Your guide to newborn screening tests
This resource is available in Urdu, Chinese and Polish, and in an Easy Read format. NHS Health Scotland is happy to consider requests for other languages and formats. Please contact 0131 536 5500 or email nhs.healthscotland-alternativeformats@nhs.net
## Contents

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>About this booklet</td>
<td>2</td>
</tr>
<tr>
<td>Hearing screen test</td>
<td>7</td>
</tr>
<tr>
<td>Blood spot test</td>
<td>10</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>13</td>
</tr>
<tr>
<td>Congenital Hypothyroidism</td>
<td>14</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>16</td>
</tr>
<tr>
<td>Medium Chain Acyl-CoA Dehydrogenase</td>
<td>17</td>
</tr>
<tr>
<td>Deficiency (MCADD)</td>
<td>17</td>
</tr>
<tr>
<td>Sickle Cell Disorder</td>
<td>18</td>
</tr>
<tr>
<td>Blood spot test results</td>
<td>19</td>
</tr>
<tr>
<td>More information</td>
<td>22</td>
</tr>
</tbody>
</table>
About this booklet

The health professional taking care of you will explain what newborn screening tests are being offered. They will always ask for your permission before a test, and you can decide at any point that you do not want your baby to be tested. You can choose for your baby to have only some or all of the tests offered. This booklet provides more information on each of the tests, so that you can make an informed decision about having screening tests.

The newborn screening programme keeps a record of your baby’s personal screening results. Only authorised staff and appropriate healthcare professionals have access to this information. All NHS staff are bound by a strict code of confidentiality.

Throughout this booklet, we use the term ‘health professional taking care of you’. This is because there can be different specialists responsible for different screening tests.
During your pregnancy, you were offered a range of tests for you and your unborn baby. Although most babies are born healthy, a few may have rare problems that can only be picked up after a baby is born. This booklet is about the screening tests offered to all babies to ensure that, if they are one of the few that do have a problem, this is picked up and treatment is started as soon as possible. These tests are done in the first few weeks of life, and are known as ‘newborn screening tests’.

It is important that you read the information in this booklet carefully, and remember to keep it handy for any hospital visits or when you meet with your midwife or doctor. It will help prepare you for when you discuss the screening tests with them.

The following pages explain what conditions can be tested for and what the tests involve, so you can decide whether you want your baby to have them. It is important that you realise the reasons for screening, and understand the possible outcomes if you choose not to have the tests.

If you do not want your baby screened for any (or all) of the conditions please talk this through with your midwife or doctor.

For more information on newborn screening tests, please talk to the health professional taking care of you – they will be happy to help.
### Newborn screening tests: at a glance

<table>
<thead>
<tr>
<th>When</th>
<th>Screening test</th>
</tr>
</thead>
<tbody>
<tr>
<td>By 72 hours</td>
<td>Routine examination, <a href="#">page 6</a></td>
</tr>
<tr>
<td>Around day 5</td>
<td>Blood spot test, <a href="#">page 10</a>:</td>
</tr>
<tr>
<td></td>
<td>• Phenylketonuria (PKU), <a href="#">page 13</a></td>
</tr>
<tr>
<td></td>
<td>• Congenital Hypothyroidism (CHT), <a href="#">page 14</a></td>
</tr>
<tr>
<td></td>
<td>• Cystic Fibrosis (CF), <a href="#">page 16</a></td>
</tr>
<tr>
<td></td>
<td>• Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), <a href="#">page 17</a></td>
</tr>
<tr>
<td></td>
<td>• Sickle Cell Disorder (SCD), <a href="#">page 18</a></td>
</tr>
<tr>
<td>From birth to 4 weeks</td>
<td>Hearing screening test, <a href="#">page 7</a></td>
</tr>
<tr>
<td>By 8 weeks</td>
<td>Physical examination</td>
</tr>
<tr>
<td></td>
<td>(Details can be found in <em>A guide to the routine examination of the newborn. Ask your midwife for a copy.</em>)</td>
</tr>
</tbody>
</table>

Remember, you can discuss all screening tests with your midwife or health visitor.
Routine examination

All babies are examined carefully within the first three days of life so that any obvious physical abnormalities can be picked up as soon as possible. A booklet ‘Routine examination of the newborn’ has more information. You can get this from the health professional taking care of you.
Hearing screen test

The hearing screen test is a simple test that will be done within the first few weeks after your baby is born – it may even be done before you leave the maternity unit.

**Why is the test done?**

The test checks if your baby has a hearing loss. This is important for your baby’s development and it means that, if needed, you can get support and information at an early stage.

If your baby’s hearing is not tested, please ask the health professional taking care of you to make an appointment. One or two babies in every 1,000 are born with a hearing loss in one or both ears. Most of these babies are born into families with no experience or history of hearing loss.

**What happens during the test?**

The hearing screen test 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.
- Three small sensors will be placed on your baby’s head along with a small earpiece or headphones.

A computer will then measure how well your baby’s ears respond to clicking sounds.
Does the test hurt?
No. The test does not hurt and it is not uncomfortable. It will usually be done while your baby is asleep and it is very quick.

When will I get the results of the hearing screen test?
The results will usually be given to you at the time. If you have any concerns or questions about your baby’s results, please tell the health professional taking care of you and they will be able to help.

If the screening test shows a clear response from both of your baby’s ears
This means your baby is unlikely to have any hearing loss. After the test you may be given a checklist of the sounds that your baby should respond to as they grow older.

If you have any concerns about your baby’s hearing in the future, you can talk to your health visitor or doctor. They can arrange for another hearing screen test at any age.

If the screening test does not show a clear response from one or both of your baby’s ears
This is quite common and does not necessarily mean your baby has a hearing loss. You will be invited to have a second test.

Some other reasons why your baby might have to have a second test:
- Your baby may have been unsettled at the time of the test.
- There may have been background noise.
- Your baby may have had fluid or a temporary blockage in his or her ears after birth. This is very common and usually clears in time.
Remember

Even if you can see that your baby responds to sound, it is still very important that they have the hearing screen test. This is because those babies that do have a hearing loss will usually respond to some sounds. If your baby’s hearing is affected, it is important to find out as soon as possible so that you can be given the support and advice you need.

If the second test does not show a clear response from one or both of your baby’s ears, then further tests will be carried out. The health professional taking care of you will give you further information to explain this more.
Blood spot test

Some health problems can’t be seen by just examining the baby, but they can be picked up through a blood test.

All newborn babies in Scotland are eligible for a blood spot test. Your midwife will explain the test in detail, and ask for your permission to carry it out.

How is the blood spot test taken?

The newborn blood spot test is usually carried out around five days after the baby is born. The midwife pricks the baby’s heel to obtain a few small drops of blood. The blood is then put onto a printed card and sent to a laboratory for testing.

What will my baby’s blood be tested for?

The blood sample will be tested for several rare conditions, including:

- Phenylketonuria (PKU)
- Congenital Hypothyroidism (CHT)
- Cystic Fibrosis (CF)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
- Sickle Cell Disorder (SCD)
What is Phenylketonuria?
Phenylketonuria (PKU) is a rare, inherited disorder that affects around one in every 8,000 babies born in Scotland. It means they can’t digest phenylalanine. Phenylalanine is a natural part of the protein within our body and is found in most of our foods.

What happens if PKU is suspected?
If the blood test shows that your baby might have PKU, the health professional taking care of you will make arrangements for you to see a doctor who specialises in looking after children. The specialist will carry out further tests.

If these tests confirm that your baby has PKU, the condition can be treated by keeping to a special diet, to make sure that your baby will develop healthily.

What would happen if a baby is not screened for PKU?
It is your choice whether to have your baby’s blood tested. However, it is important to know that if your baby has PKU and this is not treated, phenylalanine would build up in your baby’s blood, and may cause severe damage to the brain which cannot be reversed.
What is Congenital Hypothyroidism?

Congenital Hypothyroidism (CHT) is a rare condition that affects approximately one in every 3,500 babies born in Scotland. ‘Congenital’ means that a baby is born with the condition. ‘Hypothyroidism’ means that the baby won’t produce enough of the hormone thyroxine, which is needed for healthy mental and physical development.

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

It is your choice whether to have your baby’s blood tested. If your baby has CHT and this is not treated, it would result in delayed growth and severe learning difficulties.

What happens if CHT is suspected?

If the blood test shows that your baby might have CHT, the health professional taking care of you will make arrangements for you to see a doctor who specialises in looking after children. The specialist will carry out some further tests.

If these tests confirm that your baby has CHT, the condition is easily corrected by giving your baby thyroxine as a medicine.
What is Cystic Fibrosis?
Cystic Fibrosis (CF) affects one in every 2,500 babies born in Scotland. It occurs when a baby inherits an altered form of the CF gene from each of its parents, which together cause CF. Both parents are healthy carriers of the altered gene and are unaffected by the condition themselves. One in 25 people in Scotland is a CF gene carrier. CF affects the lungs and the pancreas the most, causing chest infections and problems with digesting food.

How is the test for CF taken?
The baby’s blood is tested for a substance called ‘Immunoreactive Trypsinogen’ (IRT). If the levels are found to be high, an additional test for CF is carried out using DNA taken from the same blood spot sample.

If you have any concerns about your baby’s DNA being tested for this purpose, please discuss this with the health professional taking care of you.

If it is suspected that your baby has CF, you will usually be told at this stage. But if the test does not show a clear result, you might be asked for your baby to have another blood spot sample taken between 21 and 28 days of age.

It is important to understand that, on rare occasions, some cases of CF in babies may not be picked up by the screening tests.

What happens if CF is suspected?
The health professional taking care of you will make arrangements for you to see a CF specialist, who will confirm the diagnosis and ensure that your baby receives any necessary treatment. They will also see that you have all the information you need about the condition.

You may also wish to see a Genetic Counsellor, who can give more detailed information about what the results mean for you, your baby and your family.

What would happen if a baby is not screened for CF?
It is your choice whether to have your baby tested. If your baby has CF and this is not detected, there may be a delay
in diagnosis and your baby may develop problems that could be minimised or avoided with the correct early treatment.

**CF gene carriers**
The tests pick up a small number of babies who will be unaffected by the condition themselves, but may be healthy carriers of the gene like their parents. They will not need any special treatment but as carriers may pass the gene to the next generation.

If your baby is identified as a carrier, the health professional looking after you 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.

**What is Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)?**
About one in 10,000 babies born in Scotland has MCADD.

Babies with this inherited condition have problems breaking down certain fats, in order to make energy for the body. It can lead to serious illness, and in some cases even death, if the baby is not feeding regularly.

**What happens if MCADD is suspected?**
If the blood test shows that your baby may have MCADD, the health professional taking care of you will make arrangements for you to see a specialist doctor as soon as possible.

Further tests including DNA analysis will be carried out to confirm whether your baby has MCADD, and you will be asked to make sure that your child eats regularly and to pay special attention to feeding when your baby is unwell. This can prevent serious illness and allow babies with MCADD to develop healthily.

**What would happen if a baby is not screened for MCADD?**
It is your choice whether to have your baby’s blood tested. If your baby has MCADD and this is not detected, it can lead to serious illness or in some cases death (if your baby goes for a very long time without food, particularly if your baby develops an infection).
What is Sickle Cell Disorder?
Sickle Cell Disorder (SCD) is an inherited condition affecting one in 2,500 babies born in the UK. It is a condition that affects the quality of the cells that carry oxygen in the blood. The blood cells of someone with SCD change from a round to a ‘sickle’ shape, and get stuck in the small blood vessels. This can cause pain and damage to the baby’s body, serious infection, or in some cases death.

What happens if SCD is suspected?
If the blood test shows that your baby has SCD, the health professional taking care of you will make arrangements for you to see a haematologist (a doctor who specialises in treating blood disorders).

Your baby will be started on treatment, including antibiotics and immunisations, to prevent serious illness.
**What would happen if a baby is not screened for SCD?**

It is your choice whether to have your baby’s blood tested. If your baby has SCD and this is not detected, your baby would develop painful symptoms and damage that could be minimised and even avoided with the correct treatment.

**SCD gene carriers**

Screening identifies babies who are genetic carriers of SCD or other unusual red blood cell disorders. They are healthy and unaffected by the condition but, as carriers, may pass the gene on to the next generation.

---

**Blood spot test results**

**Why are repeat tests sometimes needed?**

Occasionally it is necessary to repeat the newborn blood spot test because:

- there was not enough blood on the card for testing
- the blood spot card was damaged
- one of the results was unclear and needs to be repeated.
What happens to the blood spot test results?
Most tests show there are no health problems, and the results are therefore not usually reported directly to the family. However, your community midwife or health visitor will be able to give you this information.

If your baby’s blood spot test shows any possible health problems, the health professional taking care of you will ask you to make an appointment to see them, and your GP will also be informed.

What happens to the blood spot cards after testing is complete?
The stored blood spots can also be used to test for some other disorders which are not part of the standard screening programme. This may be useful if your baby becomes ill and the doctor requests further tests, but this would always be discussed with the baby’s parents first.

Left over blood spots can also be used anonymously for other monitoring and laboratory purposes such as comparing different screening methods and developing new tests. Occasionally it is necessary to use identifiable specimens, in which case the parents’ permission would always be sought.

If you do not want the stored blood spot card to be used for research, please ask the midwife to write ‘no research’ in the comments box on the blood spot card.
Thank you for taking the time to read this booklet. The information can be a lot to take in. Please talk to the health professional taking care of you if you have any questions or concerns.

You may also find the following contacts useful:

**British Thyroid Foundation**  
A patient-led charitable organisation dedicated to helping those with thyroid disorders.  
Tel: 01423 709 707  
www.btf-thyroid.org

**Cystic Fibrosis Trust**  
The Trust works to improve the lives of people with CF, raise the profile of CF and fund research.  
Tel: 020 8464 7211  
www.cftrust.org.uk

**CLIMB (Children Living with inherited Metabolic Diseases) – the National Information Centre for Metabolic Diseases**  
A national organisation working on behalf of children, young people and families affected by metabolic disease.  
Tel: 0800 652 3181  
www.climb.org.uk

**Healthtalkonline**  
Offers a directory of personal experiences aimed at patients, their carers, family and friends, doctors, nurses and other health professionals.  
www.healthtalkonline.org
National Deaf Children’s Society Scotland
Provides information and support for deaf children and their families.
Tel: 0800 800 8880
or 0141 354 7850
Textphone: 0141 332 6133
www.ndcs.org.uk

National Society for Phenylketonuria (NSPKU)
Offers support for people with PKU, their families and carers.
Tel: 0208 364 3010
www.nspku.org

NHS inform
Tel: 0800 22 44 88
Textphone: 18001 0800 22 44 88
(the helpline also provides an interpreting service)
www.nhsinform.co.uk/screening

United Kingdom Newborn Screening Programme Centre
Has responsibility for developing, implementing and maintaining a high quality, uniform screening programme for all newborn babies and their parents.
http://newbornbloodspot.screening.nhs.uk/public
This publication is available online at www.healthscotland.com or telephone 0131 536 5500.

Traditional Chinese

您也可以登入 www.healthscotland.com 瀏覽本刊物，或撥打 0131 536 5500 查詢。

Polish

Ta publikacja jest dostępna online na stronie www.healthscotland.com lub pod numerem telefonu 0131 536 5500, gdzie można także zgłaszać wszelkie zapytania.

Urdu

www.healthscotland.com پر و استیاب ہے یاکسی سوالات کے لیے 0131 536 5500 پر نئیلے فون کریں۔

This resource is available in Urdu, Chinese and Polish, and in an Easy Read format. NHS Health Scotland is happy to consider requests for other languages and formats. Please contact 0131 536 5500 or email nhs.healthscotland-alternativeformats@nhs.net
Useful contact numbers

NHS 24  08454 24 24 24

GP

Health visitor/public health nurse

Nearest accident and emergency department

Local hospital

Other

This booklet has been designed to be kept with your Scottish Woman-Held Maternity Record, so that you can keep all your information together in one place.

www.nhsinform.co.uk/screening
www.readysteadybaby.org.uk
www.healthscotland.com/pregnancynewborn