

2007
Edition



A parents' guide to newborn blood spot screening

A parents' guide to newborn blood spot screening in Scotland

Happily, most babies are perfectly healthy when they are born.

A small number however are born each year with problems. This is why all babies are examined carefully after birth so that any problem identified in this way can be assessed and treated as soon as possible. Some problems of body chemistry will not show up on the head to toe check but can be detected through a blood test.

How is the blood test performed?

The Dried Blood Spot Specimen or 'heel prick test' (formerly known as the Guthrie test) is usually carried out around the fifth day of life. The midwife takes several drops of blood, from the baby's heel, to fill a number of small circles on a special card. This is then sent to the laboratory for testing.

What conditions are being tested for?

The blood spot specimen is used to screen for three rare conditions:

- Phenylketonuria (PKU)
- Congenital Hypothyroidism (CHT)
- Cystic Fibrosis (CF)

What is Phenylketonuria (PKU)?

PKU is a rare disorder that affects around 1 in every 8,000 babies born in Scotland. Babies with this condition cannot digest a substance called phenylalanine which is present in everyday foods that are rich in protein, for example milk, meat, fish, cheese and eggs.

What happens if PKU is detected by the screening test?

Your baby will be seen by a paediatrician as soon as possible. Further tests will be required before a final diagnosis of PKU is made. If PKU is detected your baby must be given a special diet. This will help your baby to develop normally.

What would happen if a baby with PKU is not detected?

Should consent be withheld, and a baby with PKU not screened, the condition would remain undetected. Phenylalanine would accumulate in the baby's blood and result in severe damage to the baby's brain which cannot be reversed.



What is Congenital Hypothyroidism (CHT)?

This rare condition affects approximately 1 in every 3,500 babies born in Scotland. 'Congenital' means that a baby is born with the condition. Hypothyroidism means that the baby does not produce enough of the hormone called thyroxine because the baby's thyroid gland is missing or not working properly. This hormone is vital for normal mental and physical development.

What happens if Hypothyroidism is detected by the screening test?

Your baby will be seen by a paediatrician as soon as possible. Further tests will be required before a final diagnosis of congenital hypothyroidism is made. The condition is easily corrected by giving thyroxine by mouth. This will help your baby to develop normally.

What would happen if a baby with congenital Hypothyroidism is not detected?

A lack of thyroxine can result in slower than normal growth and severe learning difficulties.



What is Cystic Fibrosis (CF)?

Cystic fibrosis is a serious inherited condition that affects 1 in every 2,500 babies born in Scotland. The organs that are the most severely affected are the pancreas and the lungs, causing poor digestion and chest infections.

Early treatment may help affected children to maintain good nutrition and minimise chest infections, leading to improved quality of life. The majority of affected babies will be picked up by the screening test.

How does the screening test for cystic fibrosis work?

In screening for cystic fibrosis the blood spot specimen is tested in two stages. The first stage tests for a substance called Immuno Reactive Trypsinogen (IRT), which is present in increased amounts in the blood of babies with cystic fibrosis during the first few weeks of life. Several factors other than cystic fibrosis can affect the test result and therefore not all babies with high IRT will have cystic fibrosis.

In the second stage of the test, samples with high levels of IRT undergo genetic testing for cystic fibrosis by DNA analysis.

The majority of babies with cystic fibrosis will be diagnosed at this stage but in some cases it is necessary to test IRT in a second blood spot specimen on the 27th day.

If you are concerned about DNA analysis, it is not essential for this to be done. A repeat IRT test together with a sweat test may be used instead.

As the testing process for cystic fibrosis is more complicated than for the other conditions, it may take longer to provide a result for this part of the screening test. It should also be noted that on rare occasions the IRT test will fail to identify a baby with cystic fibrosis.

What does being a carrier mean?

A small number of carriers will also be detected through DNA testing. Also it should be noted that on rare occasions the IRT test will fail to identify a baby with cystic fibrosis. Being a carrier has no effect on health. 1 in every 25 people in the Scottish population is a carrier of cystic fibrosis and one or both parents of a child diagnosed as being a carrier are also carriers. However, knowledge of a child's carrier status is of value when they grow up and plan to have their own children. An appointment with a genetic counsellor will be offered, so that parents can have the opportunity to discuss the information more fully should they wish to do so.

What happens if cystic fibrosis is suspected?

A health care professional will contact you and arrange for you to bring your baby to the hospital. Further tests may be required before a final diagnosis is offered. You will also be offered an appointment with a genetic counsellor.

Why are repeat specimens sometimes required?

Occasionally it is necessary to collect a second heel prick blood specimen, for example:

- there was not enough blood to allow proper testing
- the blood spot card was damaged or did not reach the laboratory
- one of the results was unclear and requires to be repeated. Unclear means that the test result is not completely within normal ranges. There are several reasons why tests give an unclear level and often the repeat specimen is completely normal. If a result remains unclear then arrangements will be made to have your baby seen by a paediatrician.

What happens to the test results?

All results are sent to the Department of Community Child Health in the area where the baby is resident, within three weeks of the test being taken. Here the results are matched with the birth record to ensure that every baby has been tested and has a result recorded. Arrangements will be made to have a specimen taken from any baby where a result is not available.

Normal results are not currently reported directly to each family, but your community midwife or health visitor will be able to give you this information.

Any abnormal result, which requires your baby to be seen by a paediatrician, will be notified by telephone directly to the paediatrician concerned or to a health care professional who will arrange a hospital appointment. Your GP will also be contacted.



What happens to the blood spot cards after testing is complete?

The blood spot cards are stored in the laboratory so that, if necessary, one or more of the above screening tests can be repeated to check a particular result. If you do not wish your baby's card to be stored after the 12-month testing period, please inform your midwife.

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 further testing, please ask the midwife to tick the 'No Research' box on the blood spot card.

Which babies should have the blood spot test?

All babies born in Scotland are eligible. The midwife will explain the test fully and ask you to give consent on behalf of your baby.

It is important that you realise that a delayed diagnosis of any of the conditions below may lead to permanent damage to your baby. If you refuse testing, you may be asked to sign a form confirming that the reasons for testing have been explained to you and that you understand the possible consequences for your baby.

For general health care information you can contact NHS helpline on Freephone 0800 22 44 88 or if it is an emergency NHS 24: 08454 24 24 24.



ENGLISH

More information is available in other languages and formats at:

<http://www.healthscotland.com/publications>

If you do not have web access or need help to do this, contact your local NHS Board or phone the free NHS Helpline on **0800 22 44 88** (Textphone 18001 0800 22 44 88).

বাংলা

<http://www.healthscotland.com/publications> তে অন্যান্য ভাষা এবং ফর্ম্যাট সাইটে পাওয়া যায়।

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中文

想得到更多关于该信息的不同语言或者格式的版本,请访问以下网址:

<http://www.healthscotland.com/publications>

如果你不能浏览互联网或者需要别人帮助你浏览互联网,请联系你当地的NHS(全民健康服务),免费NHS帮助热线是**0800 22 44 88**(文本电话: 18001 0800 22 44 88)。

FRANÇAIS

Pour obtenir de plus amples informations en d'autres langues et formats veuillez cliquer sur le lien suivant: <http://www.healthscotland.com/publications>

Si vous n'avez pas accès à Internet ou avez besoin d'aide pour le faire, contactez votre agence locale NHS Board ou contactez le numéro gratuit du NHS Helpline au **0800 22 44 88** (Textophone 18001 0800 22 44 88).

LIETUVIŠKAI

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Jei neturite prieigimo prie interneto, arba jeigu jums reikalinga pagalba, susisiekite su savo vietine Sveikatos Apsaugos Taryba (NHS Board), arba skambinkite nemokamai NHS pagalbos telefonui: **0800 22 44 88** (Tekstofonas: 18001 0800 22 44 88).

हिन्दी HINDI

अन्य भाषाओं और फॉर्मेट्स में अधिक जानकारी निम्नलिखित पते पर उपलब्ध है
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POLSKI POLISH

Więcej informacji w innych językach i formatach znajdują Państwo na stronie internetowej <http://www.healthscotland.com/publications>

Jeżeli nie mają Państwo dostępu do Internetu lub potrzebują pomocy w znalezieniu informacji prosimy kontaktować się z lokalną siedzibą Narodowego Funduszu Zdrowia (NHS Board) lub telefonować pod bezpłatną infolinię NHS pod numer 0800 22 44 88 (numer telefonu testowego 18001 0800 22 44 88).

Русский RUSSIAN

Дополнительная информация доступна на других языках и в другом формате на: <http://www.healthscotland.com/publications>

Если вы не можете открыть веб-страницу или вам нужна помощь, обратитесь в местное Бюро НСЗ (NHS Board) или бесплатно позвоните в Службу Помощи НСЗ (NHS) по телефону 0800 22 44 88 (Текстофон 18001 0800 22 44 88).

ESPAÑOL SPANISH

Mayores informes se encuentran disponibles en otros lenguajes e idiomas en <http://www.healthscotland.com/publications>

Si no cuenta con acceso a internet o requiere ayuda para la consulta, contacte su NHS Board local marcando al conmutador, o marque gratis a la línea NHS Helpline al 0800 22 44 88 (Para textos telefónicos al 18001 0800 22 44 88).

URDU اردو

مزید تفصیلات کے لیے پرنٹنگ ڈیٹوں اور سکرینوں میں مزید معلومات دستیاب ہیں
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Additional information on the conditions being screened for can be obtained from:

National Society for Phenylketonuria (NSPKU)

Tel: 020 8364 3010

email: info@nspku.org

www.nspku.org

British Thyroid Foundation

Tel/Fax: 01423 709 707 (answerphone only)

www.btf-thyroid.org

Cystic Fibrosis Trust

Tel: 0845 859 1000 (Helpline)

www.cftrust.org.uk

Details of organisations and websites are included for your information. Inclusion in, or exclusion from this information leaflet does not imply the endorsement or otherwise of an organisation by the Scottish Government Health Department.

Your local contact details:

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