Newborn blood spot screening for your baby

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In the first week after birth, you will be offered a blood spot screening test for your baby.

Why should babies be screened?

Newborn blood spot screening identifies babies who may have rare but serious conditions.

Most babies who are screened will not have any of these conditions but, for the small numbers who do, early treatment can improve their health and prevent severe disability or even death.

What are newborn babies screened for?

All babies in Northern Ireland are offered screening for six inherited metabolic disorders (IMDs), congenital hypothyroidism (CHT), cystic fibrosis (CF) and sickle cell disorders (SCDs).

Inherited metabolic disorders

It is important to let your health professional know as early as possible if you have a family history of any inherited metabolic disorder (IMD) (see page 5). Babies are screened for six IMDs. These are:

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

Approximately 1 in 5,000 babies born in Northern Ireland will have PKU and 1 in 10,000 will have MCADD. The other conditions are rarer, occurring in 1 in 100,000–300,000 babies.

Babies with these disorders cannot process certain substances in their food. Without treatment, babies with some of these conditions can become suddenly and seriously ill. The symptoms of the conditions are different; some may be life threatening or lead to severe developmental problems. They can all be treated by a carefully managed diet, which is different for each condition and may include additional medicines.

If babies are not screened, but are later found to have an IMD, it may be too late for the special diet to make a real difference.

Congenital hypothyroidism

About 1 in 2,000 babies born in Northern Ireland has congenital hypothyroidism (CHT). Babies with CHT do not have enough of the hormone thyroxine. Without this hormone, they do not grow properly and can develop serious, permanent, physical and mental disability.

Screening means babies with CHT can be treated early with thyroxine medicine, which will prevent serious disability and allow them to develop normally. If babies are not screened and are later found to have CHT, it may be too late to prevent them becoming seriously disabled.

Cystic fibrosis

About 1 in 2,500 babies born in Northern Ireland has cystic fibrosis (CF). This inherited condition can affect the digestion and lungs. Babies with CF may not gain weight well and may have frequent chest infections.

Screening means babies with CF can be treated early with a high-energy diet, medicines and physiotherapy. Although children with CF may still become very ill, early treatment is thought to help them live longer, healthier lives.

Screening for CF includes testing some babies for the most common gene alterations that cause the condition. This means screening may identify some babies who are likely to be genetic carriers of CF. These babies may need further testing to find out if they are a healthy carrier or have CF. Screening does not detect all carriers.

Sickle cell disorders

Less than 1 in 10,000 babies born in Northern Ireland has a sickle cell disorder (SCD). These inherited conditions affect the red blood cells, which carry oxygen around the body. Babies with a SCD have red blood cells that can change to a sickle shape and become stuck in the small blood vessels. This can cause pain and damage to the baby's body, serious infection, or even death.

Screening means babies with a SCD can receive early treatment, including immunisations and antibiotics, which, along with parent education, will help prevent serious illness and allow children to live healthier lives.

Screening may also identify babies who are genetic carriers of a SCD or another unusual red blood cell disorder. Carriers of sickle cell disorders are healthy and do not require treatment. However, they can experience some problems in situations where their bodies might not get enough oxygen, for example, if they are having an anaesthetic. Rarely, screening identifies other conditions, such as thalassaemia (a serious blood disease). These children also need to be referred for lifelong treatment and care.

What if there is a family history of IMD?

As early as possible before your baby is born, you should inform the health professional (obstetrician or midwife) looking after you if you or your partner has a family history of IMD. You may be offered referral to a genetic specialist, who will be able to answer any questions or discuss any concerns you may have.

You may be advised that your baby needs early screening. Details of the information given to you about early screening and anything special you will need to do after your baby is born will be recorded in your maternity hand-held record.

If early screening is recommended, the midwife/nurse will collect a small sample of blood from your baby's heel onto a blood spot card marked 'family history'. This will generally happen between 24–48 hours following birth and results will usually be available within 48 hours of the sample being taken.

Babies who are screened early because there is a family history of IMD will still need to have a routine blood spot screening test when they are five days old.

Babies with a family history of IMDs may require special feeding regimes following birth.

Where there is a family history of MCADD, it is important to ensure that your baby has a good milk intake. A term baby with a family history of MCADD should be fed every four hours from birth, and a pre-term baby every three hours. There are particular risks in the first 72 hours for breastfed babies due to the amount and content of breastmilk during this period. It is therefore recommended that breastfed babies receive top-ups of formula milk until a good supply of breastmilk is established.

What does the blood spot test involve?

When your baby is five days old, the midwife/nurse will prick your baby's heel using a special device to collect some drops of blood onto a card. The heel prick may be uncomfortable and your baby may cry.

How can I help?

- Make sure your baby is warm and comfortable.
- Be ready to feed and/or cuddle your baby.
- The information collected on your baby's blood spot card is important – make sure all the details are correct.

Are repeat blood samples ever needed?

Occasionally, the midwife or health visitor will contact you and ask to take a second blood sample from your baby's heel. This may be because there was not enough blood collected previously or the first result was unclear. The repeat results are usually normal.

Screening is recommended

Screening your baby for all these conditions is strongly recommended, because it could save your baby's life, however, it is not compulsory. **You can choose to have screening for SCD, CF or CHT individually but can only choose to have screening for all six IMDs or none at all.** If you do not want your baby screened for any or all of these conditions discuss it with your midwife. All your decisions will be recorded in your baby's personal child health record (PCHR or 'Red Book').

If you think your baby may not have been screened, speak to your midwife or GP.

Can my baby have the test later if I change my mind?

Yes. Babies can be screened up to 12 months of age for all the conditions except CF (only up to eight weeks of age). However, later screening may mean that it is too late for treatment to make a real difference. If you have any concerns about the tests, please discuss them with your health professional.

How will I hear about the results?

Most babies will have normal results, indicating that it is unlikely that they have any of these conditions. A health professional will usually inform parents of the screening results and record them in the personal child health record (PCHR or 'Red Book') before the baby is eight weeks old.

If you have not been given the results by the time your baby is eight weeks old, please speak to your health visitor.

A small number of babies will screen positive for one of the conditions. This does not mean they have the condition but that they are more likely to have it. They will be referred to a specialist for further tests. You will normally be contacted within three or four weeks of the initial test being carried out.

The purpose of screening is to identify babies more likely to have these conditions. Screening is not 100% accurate.

What happens to my baby's blood spots after screening?

After screening, newborn blood spots are stored for at least five years and they may be used in a number of ways:

- To check the result or for other tests recommended by your doctor.
- To improve the screening programme.
- For public health monitoring and research to help improve the health of babies and their families in the UK. This will not identify your baby and you will not be contacted.

The use of these blood spots is governed by a code of practice, available from your midwife (this code on storage and retention of blood spots is currently under review). Alternatively, you can visit: pha.site/PHEcodeofpractice

There is a small chance researchers may want to invite you or your child to take part in future research linked to the blood spot programme.
If you do not wish to receive invitations to take part in research, or you want further information, please talk to your midwife.

For local information:

All retained records relating to newborn blood spot screening meet the requirements of the Data Protection Act 2018. For further information on how the PHA uses and protects your information, find our privacy notice at pha.site/privacynotice

This leaflet is based on high-quality research evidence and the views of parents and health professionals. It has been adapted in Northern Ireland with the permission of the UK Newborn Screening Programme Centre and is used under the terms of the Open Government Licence v3.0

For translations of this leaflet, follow the QR code, ask your midwife or visit:
pha.site/newbornbloodspot

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Newborn Blood Spot Screening

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