



## Newborn blood spot screening for your baby

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, the benefits of screening are enormous. 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 phenylketonuria, congenital hypothyroidism, cystic fibrosis, sickle cell disorders and MCADD (medium chain acyl CoA dehydrogenase deficiency).

### **Phenylketonuria**

About 1 in 6,000 babies born in Northern Ireland has phenylketonuria (PKU). Babies with this inherited condition are unable to process a substance in their food called phenylalanine. If untreated, they will develop serious, irreversible, mental disability.

Screening means babies with PKU can be treated early through a special diet, which will prevent severe disability and allow them to lead a normal life.

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

### **Congenital hypothyroidism**

About 1 in 3,000 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.

If babies are not screened for CF and they do have the condition, they can be tested later, but parents may have an anxious time before CF is recognised.



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.

## **Sickle cell disorders**

About one in 5,000 babies born in Northern Ireland has a sickle cell disorder (SCD). These inherited conditions affect the red blood cells. Babies with an 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 an 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 an SCD or another unusual red blood cell disorder. Carriers of sickle cell disorders are healthy and do not require treatment. Rarely, screening identifies other conditions, such as thalassaemia, which may affect red blood cells.

## **MCADD**

About 1 in 10,000 babies born in Northern Ireland has MCADD. Babies with this inherited condition have problems breaking down fats to make energy for the body. This can lead to serious illness or even death.

Screening means most babies with MCADD can be recognised early, allowing special attention to be given to their diet, including making sure they feed regularly. This care can prevent serious illness and allow babies with MCADD to develop normally.

Screening babies for MCADD is important so that those with the condition can be identified before they become suddenly and seriously ill.



## **What if there is a family history of MCADD?**

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 MCADD. You will 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 'MCADD family history'. This will 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 MCADD will still need to have a routine blood spot screening test when they are five days old.

Where there is a family history of MCADD, it is important to ensure that your baby has a good milk intake. A term baby 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 breast milk during this period. It is therefore recommended that breastfed babies receive top-ups of formula milk until a good supply of breast milk is established.

## **Will blood spot screening in Northern Ireland show up anything else?**

Blood spot screening may also identify a number of rare metabolic diseases such as homocystinuria.

## How will the midwife/nurse take the blood spots?

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 you help?

- Make sure your baby is warm and comfortable.
- Be ready to feed and/or cuddle your baby.

## 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, but it is not compulsory. 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 notes and in your baby's personal child health record ('Red Book').

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

## How will you hear about the results?

Most babies will have normal results, indicating that they are not thought to 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 ('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.

If your baby is thought to have one of these conditions, he or she will need further tests to confirm the results. 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 your 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. Alternatively, you can visit:

[www.gov.uk/government/publications/newborn-blood-spot-screening-code-of-practice-for-the-retention-and-storage-of-residual-spots](http://www.gov.uk/government/publications/newborn-blood-spot-screening-code-of-practice-for-the-retention-and-storage-of-residual-spots)

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 further information about family history of MCADD, visit:

[www.nrls.npsa.nhs.uk/alerts/?entryid45=132858](http://www.nrls.npsa.nhs.uk/alerts/?entryid45=132858)

For translations of this leaflet, ask your midwife or visit:

[www.publichealth.hscni.net](http://www.publichealth.hscni.net)

## For local information:

All retained records relating to newborn blood spot screening meet the requirements of the 1998 Data Protection Act.

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.

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