

For further information ask your midwife,
visit www.newbornscreening-bloodspot.org.uk
For general health advice and information you
can call NHS Direct on 0845 4647 or visit
www.nhsdirect.nhs.uk



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All records kept 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 produced by the UK Newborn Screening Programme Centre, which is funded by the Department of Health for the whole of the UK.

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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 I have my baby screened?

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

Most babies screened will not have any of the 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 are screened for phenylketonuria and congenital hypothyroidism. In some areas babies are also screened for cystic fibrosis, sickle cell disorders and some other conditions. If you want to know which conditions are screened for in your area, please ask your midwife.

Phenylketonuria

About 1 in 10,000 babies born in the UK 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 that babies with the condition 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 4,000 babies born in the UK 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 that babies with CHT can be treated early with thyroxine tablets, 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.



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

After screening, newborn blood spots are stored for at least five years and 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 research to help improve the health of babies and their families in the UK. This research 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, or via www.newbornscreening-bloodspot.org.uk

In the future there is a small chance researchers may want to invite you or your child to take part in research linked to the blood spot programme. If you do not wish to receive invitations to take part in research please let your midwife know.

How will I 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 let parents know the screening result and record it in the baby's personal child health record by the time the child is **6-8 weeks old**.
- Some babies are found to be carriers. Their parents will usually be told by the time the child is **6-8 weeks old**.
- If a baby is thought to have phenylketonuria (PKU), parents will be contacted before the baby is **3 weeks old** and given an appointment to see a specialist.
- If a baby is thought to have congenital hypothyroidism (CHT), parents will be contacted before the baby is **3 weeks old** and given an appointment to see a specialist.
- If a baby is thought to have cystic fibrosis (CF), parents will be contacted before the baby is **4 weeks old**.
- If a baby is thought to have a sickle cell disorder (SCD), the parents will be contacted before the baby is **6 weeks old**.

If a baby is thought to have one of the conditions, he or she will need further tests to confirm the result.

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

Sickle cell disorders

About 1 in 2,500 babies born in the UK has a sickle cell disorder (SCD). These are inherited disorders that affect the red blood cells. If a baby has a sickle cell disorder, their red blood cells 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 that babies with SCD can receive early treatment, including immunisations and antibiotics, which, along with parent education, will help prevent serious illness and allow the child to live a healthier life.



Cystic fibrosis

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

Screening means that babies with CF can be treated early with a high-energy diet, medicines and physiotherapy. Although a child 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.

Will screening for these conditions show up anything else?

Screening for cystic fibrosis (CF) includes testing some babies for the most common gene alterations that cause CF. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have CF.

Screening identifies babies who are genetic carriers of sickle cell or other unusual red blood cell disorders. Carriers of sickle cell disorders are healthy and will not be affected by the condition.

Rarely, other conditions such as beta thalassaemia major can be identified. In this condition, the baby does not make enough red blood cells, and needs treatment for severe anaemia.



How will the midwife take the blood spots?

About a week after birth the midwife 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:

- By making sure your baby is warm and comfortable
- Being 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, or the result was unclear. Usually the repeat results are 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.

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