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Newborn bloodspot test (Heel prick test)

Mothers of all newborn babies are offered testing for certain conditions by testing a blood spot taken from the baby's heel.

How is the blood test done?

This is usually done by your midwife at home when your baby is 5 days old. A very small sample of blood is collected by pricking your baby's heel, using a special device and squeezing out a few drops of blood on to a card. Although this test may be uncomfortable, it is over very quickly. The sample is sent away for testing, and you will get the results by letter or phone in due course.

It is important that you tell your healthcare professional if you, your partner or a member of your family have been diagnosed with one of the conditions below.

Which conditions might be tested for by the heel prick test?

Conditions which the heel prick test can screen for are as follows:

- Sickle cell disease - this is an inherited blood disorder. Treatment started early in life, can prevent complications. This may include taking regular antibiotics and having extra vaccinations.
- Cystic fibrosis - this is an inherited disorder that affects the internal organs, especially the lungs and digestive system. The heel prick test detects a chemical called immunoreactive trypsinogen. This is high in babies with cystic fibrosis. If it is high then a sweat test and genetic test are usually done to confirm the diagnosis. The earlier the diagnosis is made, the sooner treatment can begin which improves the outlook (prognosis).

- Phenylketonuria - this is a very rare condition in which the body is unable to break down a substance called phenylalanine, which builds up in the blood and brain. It is really important for this to be picked up early. Early treatment significantly reduces the risk of brain problems and complications in the future.
- Congenital hypothyroidism - this is a rare condition which can lead to impaired growth and mental development. However, treated babies can develop normally so it is important for it to be diagnosed early. If your baby was born prematurely (under 32 weeks) then your baby will need a repeat test when they are 28 days old.
- Medium-chain acyl-Co-A dehydrogenase deficiency - this is a very rare, but potentially life-threatening, inherited disorder, where fat cannot be broken down by the body as well as usual. Babies with this condition develop normally, but recognising the condition early enables parents to make sure their babies and children with this condition eat regularly. When people with this condition go for a long period of time without eating there is a build-up of medium-chain fats inside the body, which can have a poisonous effect. Also, a sudden and severe drop in blood sugar levels can occur.
- All four devolved nations of the United Kingdom now offer screening for four other serious but treatable conditions. These are:
    - Homocystinuria (HCU)
    - Maple syrup urine disease (MSUD)
    - Glutaric aciduria type 1 (GA1)
    - Isovaleric acidaemia (IVA)

If you do not want your baby to have some or all of these conditions tested for in the heel prick test then you should inform your GP or midwife.

Further reading

- Population Screening Programmes (England); GOV.UK
- Newborn screening; NHS Choices
- Newborn screening; NI Direct Government Services
- Postpartum care; NICE Guidance (April 2021)

- Newborn bloodspot screening NHS Scotland

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