Newborn Bloodspot Screening Test

What is the newborn bloodspot screening test?
This is a blood test recommended for newborn babies between 48 hours and 72 hours of age. It can pick up rare but serious medical conditions. If these conditions are picked up (diagnosed) early, it can help prevent serious health and development problems for your baby.

Babies with these rare conditions may not show any signs and may not seem sick. There may also be no family history of the condition.

The test is very important to help find out if your baby has one of these conditions. Finding these early is the best way to prevent health problems later on. If you are worried about the test, talk with your healthcare provider.

What does it test for?
There are more than 25 different genetic and metabolic conditions that the test looks for. The blood from the test won’t be used for any other reason without your consent.

Common conditions the newborn blood spot screen tests for are:

- **cystic fibrosis**—this affects digestive juices, mucous and sweat. Cystic fibrosis causes breathing problems, lung infections and poor growth if not treated with medicines, diet and other therapy.

- **PKU (phenylketonuria)**—the body can’t breakdown a protein (called phenylalanine) properly. This causes the protein (phenylalanine) to build-up in the blood. If not managed with a special diet, it can cause brain damage.

- **hypothyroidism**—the body doesn’t make enough thyroid hormone. If not treated with medicine (to replace the thyroid hormone), it can cause slow intellectual development (slow ability to think and reason).

- **galactosaemia**—the body can’t breakdown a sugar (called galactose) properly. This can lead to a build-up of galactose in the blood. Galactose is found in many foods including in breastmilk. If not treated with a special diet, it can cause problems with the liver and kidneys, and intellectual disability and cataracts.

- **congenital adrenal hyperplasia**—the body doesn’t make enough of a hormone called cortisol. Cortisol helps the body deal with illness and stress. If not treated with medicine, it can change how the body develops and matures. It may also result in severe dehydration needing treatment in hospital.
Will the test hurt my baby?
You baby may feel brief pain when the needle pricks their heel, and some discomfort while the blood is being collected.

You can help reduce your baby’s pain or discomfort by having skin to skin contact, swaddling your baby and breastfeeding just before or during the test. Blood flow to their feet can be helped by keeping their feet warm, putting socks on before the test and by cuddling them with their feet hanging down.

Your baby may be able to have a few drops of a mild pain relief called sucrose in their mouth, a couple of minutes before the test is done.

When is the newborn bloodspot screening test done?
The test is best done when your baby is between 48 and 72 hours of age (2–3 days).

A repeat test is sometimes needed if there is a problem with the sample
Some other reasons a repeat test may be needed— if your baby:
• was not having any milk feeds at all before the test was first done
• had a glucose (sugar) drip
• was preterm or low birth weight
• was a same sex twin or triplet
• had a blood transfusion

What happens if your baby goes home before 48 hours of age?
Before you go home, you will be given information about how and where your baby can have the test. It is very important this is done between 48 and 72 hours of age.

The test may be performed just before you go home from hospital. However, because some conditions don’t show up before 48 hours of age, a second sample is needed so that all the conditions can be tested for.

When will we get the results?
You will only be contacted if another sample is needed, or if there is a result that needs to be looked at more closely.

What treatment will your baby need?
If the test shows your baby could have a problem, further tests and treatment will be needed. The extra tests are to be sure your baby has the condition. The treatment is different for each condition. Your baby may require a special diet, medicine or other care such as physiotherapy.

Does your baby have to have this test?
Your healthcare provider will provide you with information about the test. Your permission is needed before it can be done, and they will ask if it is ok to collect your baby’s blood.

What if you decide not to have this test?
Talk to your healthcare provider about your concerns. If you decide you don’t want your baby to have the test, tell your GP and child health nurse that the test has not been done. They will know to check your baby for signs of the conditions (listed above) if they become unwell or are not developing normally.