

Newborn Screening Test

Helping you decide

You can also read this information on our website at smauk.org.uk/399e where you can follow all the links we give to further information.

This information is for any parents of a newborn baby born in a hospital or area offering spinal muscular atrophy (SMA) screening.

1. Your options

The NHS offers newborn blood spot screening when a baby is 5 days old. They are given a heel prick to take blood. If your baby has been born in one of the hospitals or areas that is offering screening for SMA, you can choose:

To have your baby screened for SMA.

If you choose this option, your baby's information will help with making decisions about:

- adding the test permanently to newborn blood spot screening
- if added, the best way to introduce NHS screening for SMA nationally.

In Scotland your baby will be automatically 'opted in' for this test.

Not to have your baby screened for SMA.

If you choose this option, your baby can still be screened for the other conditions covered by newborn blood spot screening.

In Scotland you will need to 'opt out' your baby from the SMA test if you don't want them to have it.

2. What is SMA?

SMA is a rare condition. There are different forms of SMA. The most common and treatable form is called 5q-SMA. This newborn bloodspot screening test is for 5q-SMA. To make it easier to read this information, we refer to it as SMA.

Recent studies suggest that SMA affects an estimated 1 in 14,000 births. This would mean that 47 babies are born a year with SMA in the UK^{1, 2, 3}.

The NHS estimates that the number is more likely to be around 66 babies a year. Exact numbers are not known.

The condition causes muscle weakness and wasting (atrophy), which gets worse over time. SMA affects movement, breathing and swallowing.

Symptoms can start at different ages, from newborn to young adult.

There are four main types of SMA broadly based on when symptoms started. The impact it has varies from person to person. It can be life-threatening in the most severe cases.

For more information see: [SMA – a Brief Summary](#) >

There is no cure for SMA but there are drug treatments that can help. If given before symptoms start, the drugs can stop them developing.

Early treatment can help babies with SMA have the best chance of a healthier future.

3. Newborn blood spot screening

The NHS offers newborn blood spot screening. This uses a heel prick to take blood when a baby is 5 days old. It currently looks for 10 rare but serious conditions.

Most babies will not have any of the conditions. For those who do, finding them early through screening means they can get earlier treatment. This can prevent severe disability or even save their life.

Detailed information about screening tests for you and your baby is available. You can speak to your midwife or search online for screening tests for you and your baby '[STFYAYB](#)' >

4. How SMA is screened for

We all have 2 copies of a gene called *SMN1* – one inherited from each parent.

If a baby inherits one **altered** *SMN1* gene from each parent (a gene that is different from how it should be), they may develop SMA.

To test for SMA, the same blood sample taken during the newborn screening heel prick test is used.

5. SMA test results

There are 2 possible results – ‘SMA suspected’ and ‘SMA not suspected’:

- **SMA not suspected (no altered *SMN1* genes)**

Most babies’ blood samples do not show the altered genes that indicate SMA.

- **SMA suspected (altered *SMN1* genes)**

If your baby’s blood sample shows altered *SMN1* genes this means your baby may have SMA. If these altered genes are present, a member of a specialist clinical team will call you to arrange an urgent appointment.

They will need to see your baby as soon as possible – usually the day after they call you.

They will assess your baby and take another blood sample to confirm the results and to determine what this may mean for your baby.

The specialist team will talk to you about available treatments and offer advice and support.

Newborn screening finds most babies with SMA. However, the test cannot detect very rare genetic changes in the *SMN1* gene. These are known as point mutations. Also, this test cannot detect the extremely rare forms of SMA caused by different genes. So, while it is very unlikely, a baby can still develop SMA even if newborn tests are negative.

[**If your baby’s test result shows suspected SMA read more >**](#)

6. References

1. Aragon-Gawinska et al. (2023) Spinal muscular atrophy treatment in patients identified by newborn screening—A systematic review. *Genes* 14: 1377.
2. Belter et al (2024) Newborn Screening and Birth Prevalence for Spinal Muscular Atrophy in the US” in *JAMA Pediatrics* on July 15, 2024.
3. Verhaart IEC, et al. (2017) A multi-source approach to determine SMA incidence and research ready population. *J Neurol* 264: 1465–1473



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