TEST

Neonatal cystic fibrosis screening test

A newborn heel-prick blood test that identifies babies who may need further testing for cystic fibrosis.

WHAT IT MEASURES
Blood IRT, sometimes followed by CFTR gene variants

SAMPLE
Blood from a heel prick

FASTING NEEDED?
No

HOW LONG IT TAKES
A few minutes

RESULTS IN
Several days to weeks, varying by laboratory

USED FOR
Screening newborns for cystic fibrosis

MAIN LIMITATION
Positive screens need confirmation; some cases are missed

Overview

Neonatal cystic fibrosis screening is part of newborn blood spot screening in many countries. A few drops of blood are collected from your baby's heel and placed on a special card.

The laboratory first measures immunoreactive trypsinogen, usually shortened to IRT. This substance is made by the pancreas, an organ that helps with digestion. Newborns with cystic fibrosis often have a higher IRT level because thick secretions can affect the pancreas.

A raised IRT level does not mean that your baby has cystic fibrosis. Premature birth, a difficult delivery and other newborn health problems can also raise it.

Depending on the screening programme, the laboratory may then look for selected changes in the CFTR gene. CFTR is the gene associated with cystic fibrosis. Some programmes repeat the IRT test instead, or combine IRT, genetic testing and a second blood sample.

This is a screening test, not a diagnostic test. It estimates whether further testing is needed.

Why the test is ordered

Screening aims to identify cystic fibrosis before clear symptoms develop. Early identification allows a specialist team to assess feeding, growth, digestion and lung health promptly.

Without screening, cystic fibrosis may first become apparent through poor weight gain, frequent chest infections, unusually salty sweat or difficulty absorbing food. However, these signs can have many other causes.

The test may also show that a baby carries one CFTR gene variant without having cystic fibrosis. Whether carrier results are reported varies between screening programmes.

Who should have the test

Where cystic fibrosis is included in routine newborn screening, it is usually offered to all newborns. The exact collection day differs by country and programme. In some places the sample is taken during the first two days of life; elsewhere it is collected several days after birth.

A baby may need separate assessment even after screening if there is:

Tell the newborn screening team if your baby was born elsewhere, moved between hospitals or did not have a blood spot sample. They can check whether screening was completed.

- A sibling or close relative with cystic fibrosis
- A known CFTR gene variant in the family
- A bowel blockage at birth called meconium ileus
- Symptoms that continue despite a screen-negative result

How to prepare

Your baby does not need to fast. Milk feeds can continue as usual, and you should not stop any medicine unless your baby's healthcare team has told you to do so.

Feeding, cuddling or skin-to-skin contact may help settle your baby during the heel prick. Keeping the foot warm can make it easier to collect enough blood.

Tell the person collecting the sample if your baby was premature, seriously unwell, received a blood transfusion or has a family history of cystic fibrosis. These details can affect the timing or interpretation of newborn screening.

What happens during the test

A healthcare professional cleans your baby's heel and makes a small prick with a sterile device. They collect several drops of blood onto marked circles on a filter-paper card.

The card is allowed to dry and is sent to a newborn screening laboratory. The same card is usually used to screen for several conditions.

If the IRT result meets the programme's threshold, the laboratory may test the same sample for selected CFTR gene variants. In programmes that use repeat IRT testing, you may be asked to bring your baby back for another heel-prick sample.

The collection itself takes only a few minutes. Your baby may cry briefly, but you can usually hold and comfort them.

Understanding your results

Results and wording differ between screening programmes. The main result categories are:

A screen-positive result is not a diagnosis. Many babies with a positive result do not have cystic fibrosis.

The usual confirmatory test is a sweat test. This measures chloride, a type of salt, in sweat. It is performed at a centre experienced in testing babies. Genetic testing may also be expanded beyond the limited set of variants used in screening.

Sometimes follow-up results remain uncertain. You may hear terms such as CF screen positive, inconclusive diagnosis or CFTR-related metabolic syndrome . These describe babies who need specialist follow-up but do not currently meet the diagnostic criteria for cystic fibrosis.

Laboratory thresholds and interpretation vary by programme, laboratory, age, sex, pregnancy status, medicines and clinical context. Pregnancy status is not relevant to the newborn, but maternal and birth information may form part of the overall assessment.

- Screen negative: The result did not meet the programme's threshold for further cystic fibrosis testing
- Screen positive: The result suggests that confirmatory testing is needed
- Repeat or unsuitable sample: Another blood spot is needed because of timing, sample quality or an unclear result

What affects the results

IRT is not specific to cystic fibrosis. It can be raised when a newborn is premature, unwell or has experienced physical stress around delivery. Low oxygen levels, surgery and some serious newborn illnesses may also affect it.

The baby's age at collection matters because IRT levels naturally change after birth. A sample taken too early or later than the programme expects may require different interpretation or repetition.

Sample quality also matters. Too little blood, blood applied in several layers, contamination or a card that was not dried correctly can make the result unreliable. Heat and delays during transport may reduce measurable IRT.

A baby with meconium ileus, a bowel blockage sometimes associated with cystic fibrosis, can occasionally have an IRT result that does not trigger further screening. Such babies need direct clinical assessment regardless of the screening result.

Genetic screening panels test only selected CFTR variants. They may miss rare variants, and the chance of this can differ with family ancestry because variant frequencies are not the same in every population.

A recent transfusion can affect parts of newborn blood spot screening and may change the collection schedule. It is not usually the main influence on the IRT measurement, but the screening team still needs to know about it.

Feeding, the parent's diet and ordinary activity do not usually affect the result. No common medicine is known to require routine stopping specifically for cystic fibrosis screening.

Accuracy and limitations

No single accuracy figure applies to every neonatal cystic fibrosis screening test. Programmes use different collection times, IRT thresholds, repeat-testing methods and genetic panels.

The test finds many babies with cystic fibrosis, but it can miss some. This may happen if IRT is not high enough, if it has fallen before testing, or if the baby's CFTR variants are not included in the screening panel. A screen-negative result therefore does not completely rule out cystic fibrosis.

The test can also be positive when the baby does not have cystic fibrosis. IRT can rise for reasons unrelated to the condition, and genetic screening may identify a healthy carrier.

A sweat test is generally the better test for confirming or excluding cystic fibrosis after a positive screen. Broader genetic analysis may be useful when the sweat result is unclear or the

suspected variants are not covered by the screening panel.

Risks and complications

A heel prick can cause brief pain, a small bruise or minor bleeding. Babies occasionally become unsettled or feel faint, but serious complications are uncommon.

What happens next

A screen-negative result usually requires no cystic fibrosis follow-up. Routine newborn and child health appointments should continue.

If the sample was unsuitable or collected at the wrong time, the screening service will arrange a repeat. This is common and does not by itself suggest that your baby has cystic fibrosis.

After a screen-positive result, your baby should be referred promptly to an experienced cystic fibrosis or paediatric service. The team will explain the result and arrange a sweat test or other follow-up. This is important, but a positive screening result is not usually an emergency.

Seek urgent medical advice regardless of the screening result if your baby is feeding poorly, repeatedly vomiting, producing fewer wet nappies, not gaining weight or having ongoing breathing problems.

Call emergency services if your baby has severe difficulty breathing, develops blue or grey lips or skin, is difficult to wake, or has a swollen abdomen with green vomit. These symptoms need immediate assessment and should not wait for screening results.

Common questions

Can my baby feed before the test?

Yes. Breast milk and formula do not interfere with cystic fibrosis screening, so fasting is not needed.

Can I drink coffee before my baby's test?

Yes. What you eat or drink before the heel prick does not affect your baby's result.

Does the time of day matter?

Usually not. Your baby's age in hours or days matters more than the time on the clock. The sample should be collected within the schedule used by your local programme.

Can my baby exercise or have tummy time beforehand?

Normal movement, crying and tummy time do not require special restrictions. Unlike some adult blood tests, recent exercise is not a recognised major influence on newborn IRT screening.

Why does my baby need another heel prick?

The first card may have contained too little blood, been collected outside the recommended window or produced a result requiring repeat IRT testing. A repeat request does not confirm cystic fibrosis.

Does a positive result mean my baby has cystic fibrosis?

No. It means the chance is high enough to justify a sweat test or further genetic testing. Some positive results are caused by a temporary rise in IRT or identify a healthy carrier.

Sources

- Cystic Fibrosis Foundation: Newborn Screening for CF
- Cystic Fibrosis Foundation: Sweat Test Clinical Care Guidelines
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- National Institute for Health and Care Excellence: Cystic fibrosis-diagnosis and management, NG78
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