Information on a Positive Newborn Screening Result for

Cystic Fibrosis (CF)

MARITIME Newborn Screening
Dépistage néonatal DES MARITIMES

Summary:
- The baby may be more or less likely to have cystic fibrosis (CF) based on the newborn screen result.
- **More testing is needed** for a final answer. The baby needs to have a **sweat test**.
- **There is treatment for CF.**
- Maritime newborn screening connects the baby with the IWK CF clinic for follow-up testing.

What is newborn screening?

These are routine tests done soon after birth. A few drops of blood from a baby's heel are put onto a piece of absorbent paper (blotter). The blood is tested for rare, treatable conditions. These tests are done because a newborn can look healthy but have one of these conditions and need treatment. We want to find babies with these conditions so that we can start treatment to keep them as healthy as possible.

What are the possible results from newborn screening?

Most babies have a "screen negative" (normal) result. This means a baby has a decreased chance to have the conditions on newborn screening, and no follow-up is needed. Some babies (~10%) require a repeat of the newborn screen and another sample to be collected and tested. A "screen positive" result means a baby has an increased chance to have a condition from newborn screening, but it does not mean the baby has this condition. Follow-up testing is needed to give a final answer.

What happens when there is a positive newborn screen result?

A healthcare provider from Maritime Newborn Screening (MNBS) will contact the primary care provider and the family to discuss the result. MNBS will connect the baby to the clinical team providing the follow-up. MNBS and/or the clinical team will arrange follow-up appointments and testing to happen as soon as possible. Families may feel worried about the baby's newborn screen result. Many families in this situation feel this way. The healthcare teams are here to support families during the next steps.

What does it mean if a baby has a positive screen result for cystic fibrosis (CF)?

This result does not mean that the baby has cystic fibrosis (CF). It means that more testing is needed because the baby **may** have CF. Babies with CF grow and develop better if treatment begins as soon after birth as possible. Follow-up testing is done quickly to find out if a baby has CF. This is done for the baby's best possible outcome.

What is cystic fibrosis (CF)?

Cystic fibrosis (CF) is an **inherited** condition, meaning a baby is born with it. CF happens when there are no working copies of a gene called *CFTR* (cystic fibrosis transmembrane conductance regulator). Everyone has two copies of the *CFTR* gene, and they get one from each parent. Someone with CF inherited two non-working genes. When both *CFTR* genes are non-working, it affects how water moves in and out of cells in the body. This causes the mucus in the body to be too thick. The main places this causes problems are the lungs/respiratory system and digestive system. CF can also affect other body systems/organs such as the reproductive system, pancreas, and liver. It is hard to predict the exact symptoms of someone with CF, and how severe or mild they might be. Babies with CF might have a bowel blockage called "meconium ileus", trouble gaining weight, and stools that are oily and foul-smelling, or they may have no symptoms.

How many babies have CF?

CF is a rare disease. CF is most common in people with white European ancestry, but anyone of any ancestry can have CF. CF affects approximately 1 in 3,000 people of white European ancestry; 1 in 17,000 people of African ancestry; and 1 in 31,000 people of Asian ancestry.

Why screen for CF?

Babies with CF look normal at birth and may not show any health problems right away. If we screen for CF, we can find babies with CF to start treatment before symptoms start. Early treatment can help with better growth and lung function.

How is a baby diagnosed with CF?

A baby with a positive newborn screen for CF needs to have a **sweat chloride test** and will be seen by the **CF clinic at the IWK**. A sweat test is the main diagnostic test for CF. People with CF have salty sweat, and a sweat test measures how much salt is in the sweat using a device to stimulate sweating on a small patch of skin. Genetic testing (bloodwork) also helps confirm a diagnosis of CF. Families may be connected with a genetic counsellor from Maritime Medical Genetics Service. It can take a few days or weeks to confirm a diagnosis of CF. This waiting period can be hard for families. The healthcare teams are here to support families.

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How is CF treated?

The treatments for CF have greatly improved over the past decade. Because of this, someone born with CF today has a much better outcome than someone with CF born 10-20 years ago. Most treatments for CF focus on the symptoms such as enzyme replacement to help with digestion and growth, and antibiotics, other medications, and airway clearance to keep the lungs healthy. Many people with CF are also eligible for medications that treat the underlying cause of CF. These medications are called **modulators**, and they help the product of the non-working *CFTR* genes become functional again. These modulators are incredibly effective and may stop the symptoms of CF from happening.

How does a baby get CF?

CF is an **inherited condition**. This means the baby is born with it. When a baby has CF, we know that the biological parents are likely carriers of the condition. Carriers do not have the condition but can have a baby with the condition if the baby inherits a non-working copy of the CF gene (*CFTR*) from each parent. This is called autosomal recessive inheritance (see Figure 1).

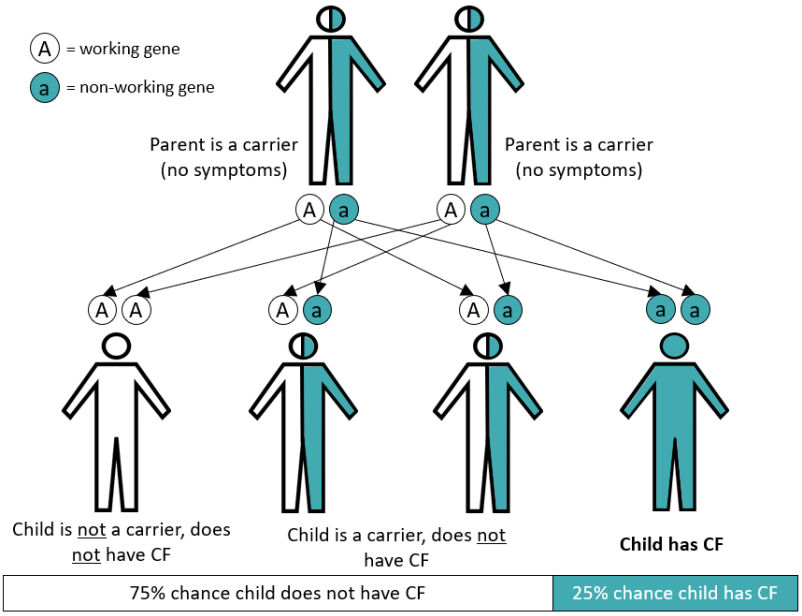

Figure 1. Autosomal recessive inheritance when both parents are carriers of cystic fibrosis (CF).

Could a family have another baby with CF?

Yes, if the biological parents are carriers there is a 25% (1 in 4) chance in each future pregnancy to have a child with CF. We offer genetic counselling through **Maritime Medical Genetics Service at the IWK** to talk about the following:

- Inheritance of CF
- Chance of recurrence of CF in future pregnancies
- Testing options for other family members

Where can I get more information?

- For more information on newborn screening, please visit our website at www.maritimenewbornscreening.ca or call the newborn screening genetic counsellor at 902-470-2783.

- Websites for families:
  - KidsHealth: https://kidshealth.org/en/parents/cf.html
  - CF Canada: https://www.cysticfibrosis.ca/

- Websites for healthcare providers:
  - MedlinePlus Genetics: https://medlineplus.gov/genetics/condition/cystic-fibrosis/
  - UpToDate: https://www.uptodate.com/contents/cystic-fibrosis-clinical-manifestations-and-diagnosis/print
  - GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1250/

NOTE TO PARENTS/GUARDIANS: *This material is provided for informational purposes and provides basic information only. This material is not intended to be and does not take the place of medical advice, diagnosis, or treatment. Please talk to your health care provider if you have any questions or concerns.*

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