
INFORMATION FOR PARENTS/GUARDIANS

My Baby Has a Positive Cystic Fibrosis Newborn Screening Result

What is Newborn Screening?

These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, like cystic fibrosis (CF).

What does it mean if my baby has a positive CF newborn screening result?

Based on how newborn screening for CF is done in Ontario, a positive screening result means that there is a high chance that your baby could have CF or develop some of the same health issues that people with CF can have, and more testing is needed to know for sure. Babies with CF can be healthier if treatment begins early, so it is important to have follow-up testing to find out if your baby truly has CF. It is natural to feel worried or have questions about your baby's positive screening result. Many parents in this situation feel this way.

What is cystic fibrosis (CF)?

Cystic fibrosis is an inherited (genetic) disease that causes thick mucus to build up in the lungs, digestive system and other organs. This leads to problems such as a persistent cough, frequent lung infections, and problems with gaining weight and growing. CF health problems can range from mild to serious. CF affects about 1 in every 3,600 babies born in Ontario.

How do I find out if my baby has CF?

A painless test, called a sweat test, is the best way of checking for CF. People with CF have a large amount of salt in their sweat, and measuring the amount of salt in the sweat can help determine whether or not your baby has CF. Some babies need other tests like blood tests.

How is the sweat test done?

A special machine causes a small part of the baby's arm or leg to sweat. The skin may feel warm and tingly for a few minutes while the machine is on. Your baby may cry during this part of the test, but it is not painful, and you can be with your baby for the whole test. The sweat is collected on a piece of filter paper or a plastic coil and sent to the lab to be tested. It takes about one hour from start to finish.

When can my baby have the sweat test?

The sweat test is usually done when your baby is at least two to three weeks of age. The test will be done later if your baby was born premature or had a low birth weight.

Do I need to do anything to prepare my baby for the sweat test?

Do not use any lotions or creams on your baby's arms or legs on the day of the test (including moisturizing soaps). You can bring an extra blanket or sweater and a hat to keep your baby warm during the test.

What do the results of the sweat test mean?

Usually, parents get the sweat test results either the same day, or the next day. There are four possible results:

- **Abnormal result:** An abnormal sweat test result means that your baby most likely has CF because a higher amount of salt was found in your baby's sweat. The sweat test result cannot tell us what symptoms your baby will have or whether the symptoms will be mild or serious.
- **Borderline result:** This means the sweat test result is in between the abnormal and normal range and more testing is needed to tell whether your baby has CF, or could develop some of the same health problems that people with CF have when they get older.



- **Normal result:** This means that it is unlikely that your baby has CF because a low amount of salt was found in your baby's sweat. Rarely, a baby can have a normal sweat test and still have CF. Some babies with a positive newborn screen who have a normal sweat test could develop some of the same health problems that people with CF can have when they get older.
- **Insufficient quantity:** This means there was not enough sweat collected to get a result. It is important to bring your baby back on another day to do the test again.

Babies who have an abnormal sweat test result should see a doctor who specializes in CF for care and to begin treatment and monitoring. For babies who have a borderline or a normal sweat test result, a doctor and/or health care professional who specializes in CF will discuss what other tests or follow-up is suggested for your baby.

Why screen for CF?

Screening means that babies with CF can be diagnosed and treated earlier. This can help people who have CF live longer, healthier lives.

How is CF treated?

Treatments for CF can include: physiotherapy and exercises to help keep the lungs clear, vitamins and supplements to help the digestive system absorb nutrients and for growth, and taking different medications. People with CF may take medications to prevent or fight infections, to help keep the lungs clear, and in some cases, to correct the non-working protein made by the CF gene.

Babies with newborn screen positive results and a normal or borderline sweat test result who are not diagnosed with CF may be offered extra check ups to monitor their child's growth and health.

How does a baby get CF?

CF is an inherited (genetic) disease. Genes provide instructions for our bodies to make proteins, and the CF gene (also called the CFTR gene) gives the instruction for the body to make CFTR protein. A person with CF has two non-working copies of the CFTR gene and as a result the CFTR protein that is made does not work properly. Each parent of a child with CF usually has one copy of a non-working CFTR gene and is considered a "carrier". Carriers of CF are usually healthy and often unaware they carry a non-working CF gene.

Where can I get more information?

For more information on newborn screening, please visit our website at www.newbornscreening.on.ca or talk to your health care provider.

For more information about CF, please visit the Canadian Cystic Fibrosis Foundation website at www.cysticfibrosis.ca or call 1-800-378-CCFF (2233).

NOTE TO PARENTS/GUARDIANS: This information is for parents whose baby has had a positive newborn screening result for cystic fibrosis. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

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