How is Cystic Fibrosis Diagnosed?

Newborn Screening

The newborn screening test is a blood test taken on the fourth or fifth day of life from all babies born in Australia. A small heel prick is done to obtain a few drops of blood to be dropped onto a special card [also known as a Guthrie card] to be sent away for testing. Screening is performed to identify uncommon, but serious medical conditions, including CF. It is an extremely successful public health program that is conducted in many countries worldwide.

If a baby has high levels of trypsin on the newborn screening test, they then undergo CF gene mutation testing from the same blood spot on the Guthrie card. If two mutations are identified the baby is affected with CF. Genetics Victoria will inform the baby's treating doctor and then the baby will be referred to a CF service at either Monash Children's Hospital or The Royal Children's Hospital, depending on their home address or their family's preference.

If only one mutation is identified the baby may only be a carrier and will not be affected with CF or they may be affected with another mutation that is not detected on the screening program. Remember there are over 1600 possible mutations but the test will only test for some of the most common mutations. If none of these mutations are found, another test called a sweat test will be performed to rule out or confirm diagnosis of CF.

Clinical Features

While newborn screening is very good at detecting babies with CF, we still miss about one or two cases per year in Victoria. These children may develop symptoms of CF such as poor weight gain, persistent diarrhoea, abdominal bloating and a productive cough. These symptoms usually lead to the suspicion of a possible diagnosis of CF. A sweat test will be performed to confirm the diagnosis of CF.

Sweat Test

Children with CF have high amounts of salt in their sweat, therefore a sweat test can be used to diagnose the CF condition. The sweat test is performed by the biochemistry laboratory here at Monash Health. It is a safe procedure that involves putting some sweat stimulation cream onto the arm or leg before applying a very small electrical current [which is painless] to the area to induce sweating. The sweat produced will be collected and tested for the level of chloride present. The diagnosis of CF can be made if the sweat chloride level is greater than 60 millimoles per litre.

Meconium Ileus

Meconium is a baby's first stool or faeces. The meconium is usually passed within the first 24 hours of life. About 15-20% of babies with CF present with a bowel blockage known as meconium ileus. The symptoms that your baby may have are a swollen abdomen and vomiting. An x-ray may be needed to confirm the obstruction. An enema can be used to clear the blockage but most often, an operation is required. An ileostomy or a colostomy bag may be required for a number of weeks after the operation depending on the severity of the obstruction. Eventually the bowel will recover allowing the reversal of ileostomy or colostomy to be reversed and the function of the bowel will return to normal.

During the first week after diagnosis

Dealing with the news that your baby has a chronic illness is often difficult and distressing. Some parents feel overwhelmed or have feelings of grief and guilt. You will have many questions and it is important to discuss your questions and feelings with your partner, friends and family.

We will always make time to be available for your concerns. Newly diagnosed families routinely meet with our social worker during the first week to help you through the adjustment to the diagnosed and the emotions that you're experiencing. Our team provides ongoing support and education. This starts from diagnosis and it is important for both parents to attend these appointments and ask any questions you may have.

Feelings of grief, guilt, helplessness, disbelief, anxiety, confusion, stress, fear and worry are all very common after the diagnosis of CF. These feelings are all normal and valid but most importantly they will ease with time. Some families experience the diagnosis as a relief, especially when their baby has been unwell or unsettled before a diagnosis without a cause found.

Most parents feel intense grief during the first few months, but say that it lessens over time.

How to grieve

- Grieve your way. No one can tell you how to feel.
- Understand that grief takes time. You'll sometimes be surprised by how you're feeling.
- Share your feelings with someone you trust, and don't be afraid to show your emotions.
- Be aware that difficult events, like going back to the hospital, may set off memories and sadness and anxiety.
- Tell people who offer to help exactly what you need – a shoulder to cry on, a helping hand with the children or making meals. Don't be afraid to ask for help or accept it as it comes.

"Focusing in what you can control, and accepting that some things in life are not under your control, often helps"

For more information visit **www.cysticfibrosis.org.au**
To hear from other families sharing their stories, visit
www.cysticfibrosisbabysteps.org.au

Notes

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FAQ

What does the Newborn Screening test in my baby's blood?

The blood test is analysed for levels of trypsin, which is one of the digestive enzymes produced by the pancreas. In those without CF, the pancreas cells produce trypsin, which then flows along small tubes (or ducts) within the pancreas, until the main pancreatic duct which connects to the gut. In this way, the digestive enzymes produced by the pancreas mix with food and allow digestion to occur. However, in babies with CF, the pancreatic secretions are abnormal, which causes blockage of the pancreatic ducts. Thus, trypsin produced by the pancreas is prevented from entering the gut, and instead spills over into the blood stream, resulting in greatly elevated blood levels of trypsin, which is then detected in the heel prick blood spot.

How will I know if I am a carrier of the gene that causes cystic fibrosis?

Most people do not know if they are a carrier. If you have cystic fibrosis in your extended family, you should ask your GP to be tested as you are more likely to be a carrier than the general population.