TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening FACT Sheet

Cystic Fibrosis (CF)

What is Cystic Fibrosis?

CF is a disease that affects the lungs, digestive system, and overall growth. People with CF have thickened secretions, which clog the lungs and cause breathing problems. It can also affect the pancreas and make it hard to break down and absorb food.

What Causes Cystic Fibrosis?

CF is inherited. This means it is passed from parents to child through genes. Each parent has two CF genes. CF happens when both the mother and the father pass a CF gene that does not work right to their child. A child only has CF if they have two abnormal genes. The disease affects the normal flow of salt and water in the body. This is what causes the body to make the thick mucous.

Screening Tests

All Texas newborns get two blood tests that screen for many rare disorders. High levels of immunoreactive trypsinogen (IRT) protein in the blood may be a sign of CF. A baby with high IRT levels, or an elevated IRT (if only one screen is obtained) will have a cystic fibrosis DNA study performed on the existing blood sample. If IRT levels are very elevated on both newborn screening tests, or the IRT levels are high on two tests and a change in the DNA (mutation) is detected, the baby will need a sweat test to determine if the high IRT is due to CF. It does not mean the baby has cystic fibrosis.

The sweat test checks the amount of salt in the baby's sweat. People with cystic fibrosis have too much salt in their sweat. During the test, a pad soaks up sweat from the baby's arm. It takes about an hour. The test result is sent to your doctor when it's ready.

A "negative" sweat test usually means the baby does not have cystic fibrosis. If the sweat test result is borderline (between normal and high), the baby may need further testing including a repeat sweat test.

A "positive" test means the baby has cystic fibrosis. Your child will need to be seen at a Cystic Fibrosis Center. Your pediatrician will help you to be seen there as soon as possible.

Newborn screening will not detect all cases of cystic fibrosis. If your child shows symptoms of CF as they get older, they should be re-evaluated for CF by a specialist.

What Symptoms or Problems Occur with Cystic Fibrosis?

Problems vary from child to child. These are some of the common ones:

- Coughing and wheezing;
- Lung infections;
- Shortness of breath;
- Salty-tasting skin and sweat;
- Bowel blockage;
- Greasy, large stools;
- Slow weight gain and growth;
- Inability to have children in males; and
- Diabetes in older children and adults.

What is the Treatment for Cystic Fibrosis?

People with CF must have treatment all during their lives. Treatments help with breathing, reduce lung infections, and help digest food. Children with CF also need to receive routine vaccinations from their pediatricians. At this time there is no cure for CF, but the current therapies have improved the survival. There are many drugs under development that will hopefully lead to increased life expectancy or possibly a cure for this disease.

Therapies and Medicines

Treatments for CF may be different for each person and are based on individual need.
These may include:

- Treatments to clear the airways;
- Antibiotics to prevent and treat infections; and
- Digestive aids and special vitamins to help digest and absorb food.

Diet

Your child should eat a healthy diet that is high in calories, protein, and fat. Extra vitamins (especially A, D, E, and K) may be needed. A dietitian can help you plan a good diet for your child.

Things to Remember

- Your child will need regular treatment all lifelong;
- Treatment can improve your child's quality and length of life; and
- Exercise helps loosen mucous and makes the heart and lungs stronger.

Note: QR code for sweat test education video is adapted from the Cystic Fibrosis Foundation website at cff.org.