University Hospitals
Case Medical Center

Rainbow Babies
& Children's Hospital

Provider Fact Sheet and Action Steps for the Cystic Fibrosis Newborn Screen

Interpretation of a Newborn Screening Report

A positive newborn screen by itself does not mean that an infant has cystic fibrosis (CF). The result means that the infant is at increased risk for CF.

If an infant has a positive immunoreactive trypsinogen (IRT) the sample will be sent for mutation analysis. An infant with a positive IRT and two identified mutations most likely has CF. An infant with a positive IRT and one mutation identified either has a second mutation which is not identified on the screening mutation panel (and might have CF), or the infant is only a carrier of CF. A sweat test will help determine whether the infant has CF or not. If the sweat test is inconclusive, additional testing can be done to confirm or rule out the diagnosis of CF.

In Ohio, it is expected that approximately 500 newborns will have positive screens; 50 will subsequently be diagnosed with CF, the others will most likely be carriers.

Rainbow Babies & Children's Hospital
(216) 844-RAINBOW
www.rainbowbabies.org

Recommended steps if a newborn has a positive result.

Contact the family and refer the infant for follow-up at the Cystic Fibrosis Center at Rainbow Babies & Children's Hospital in Cleveland. Assure the family that most babies with positive screens do not actually have CF.

Parents can call (216) 844-3936 to schedule a Friday morning appointment at Rainbow for a sweat test and genetic counseling. For the most accurate results, infants will be sweat tested 2 – 4 weeks after birth.

Information on the sweat test, a cystic fibrosis diagnosis, and what will happen during the family's visit to Rainbow along with a map is provided under the "For Parents" section tab at www.rainbowbabies.org.

Rainbow's follow-up to the pediatrician and parents.

An infant found to have CF following the sweat test, will be seen that day by a physician from the CF Center. If the infant is found to be a carrier, the genetic counselor will discuss testing options for parents and other family members. You will be notified that day by fax of the result of the sweat test. A more detailed letter summarizing the genetic counseling session will follow. Further genetic testing can be performed to identify other disease-causing CF mutations, and a nasal potential difference (NPD) test can be performed to aid in a definitive diagnosis. Rainbow's CF Center is the only Center in Ohio to offer NPD as a clinical test for infants and older individuals suspected of having CF.

For more information, please call the ODH lab at (888) 634-5227 or the Center for Human Genetics at University Hospitals at (216) 844-3936.

Diagnostic facts on Cystic Fibrosis

What is cystic fibrosis?
Cystic fibrosis (CF) is a life-threatening genetic disease with an overall incidence of approximately 1/3500 live births. The disease is most common in Caucasians, but affects all races and ethnic groups. The disease primarily affects the lungs and sinuses, gastrointestinal tract, sweat glands, and reproductive system. Common complications include progressive lung disease, chronic sinusitis, pancreatic insufficiency with growth failure, dehydration due to salt loss through sweat, and male infertility. Lung disease is the most common CF-related cause of severe disability and premature death.

What causes cystic fibrosis?
CF is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene which encodes the CFTR protein. Individual mutations are associated with a wide range of clinical symptoms. CF is inherited as an autosomal recessive disease. This means an infant must have a mutation in both copies of the CFTR gene to have CF. Heterozygote parents are asymptomatic "carriers." When two carriers have children together, each infant has a one in four (25%) chance of having CF.

In patients with CF, CFTR, which is a chloride channel protein that regulates the movement of salt and water into and out of cells, does not work properly. The cells that line the passageways of the lungs, pancreas, liver, reproductive tract and other organs produce abnormally thick secretions. Chloride is not well-absorbed from the sweat glands, leading to salty sweat; thus the diagnostic test for CF, the sweat test.

Management
Individuals who have CF are best treated at an accredited Cystic Fibrosis Center where they can be managed by a multidisciplinary team of physicians, nurses, respiratory therapists, dietitians, social workers, and genetic counselors. Medical interventions include treatment and prevention of pulmonary complications, nutritional therapy, and management of male infertility later in life.

Important note
Immediate follow-up of a positive newborn screen is important. Early diagnosis allows for immediate intervention with specialized therapies such as pancreatic enzymes to aid digestion and a high-calorie, high-fat diet. These therapies have been shown to result in improved height, weight and cognitive function. Early intervention for the lung disease maintains pulmonary function, decreases hospitalizations and leads to improved life expectancy. Equally important, false positive cases should be ruled out as soon as possible to relieve parental anxiety.

The LeRoy W. Matthews Cystic Fibrosis Center at Rainbow Babies & Children's Hospital

The LeRoy W. Matthews Cystic Fibrosis Center at Rainbow is world-renowned for its care of children and adults with CF, and is fully accredited with the highest possible rating by the CF Foundation. The model of care — a multidisciplinary team approach that includes pulmonary specialists, nurses, respiratory therapists, nutritionists, social workers and other specialists — was first developed by Dr. LeRoy Matthews in the 1950's, and adopted nationwide by CF Care Centers.

Scientists at Rainbow and its academic affiliate, Case School of Medicine, are among the world's most innovative researchers, developing new treatments and leading the search for a cure for CF. The Willard A. Bernbaum Cystic Fibrosis Research Center strives to understand the basic defect of the disease and its consequences, the reasons for the lung infection and inflammation in CF and how to treat it, and methods by which the corrective gene can be introduced into the lung. The fruits of these discoveries come to the patients via The LeRoy W. Matthews Cystic Fibrosis Center and the newly opened KC and Ginny Bryan Pulmonary Diagnostic Center. At these facilities, living a long life is the goal. So successful have the Center treatments become, that Rainbow recently opened a 10 room in-patient unit which provides specialized care for its large population of adult CF patients, who are among the oldest in the nation.

The LeRoy W. Matthews
Cystic Fibrosis Center at
Rainbow Babies & Children's Hospital
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