

Cystic Fibrosis Fact Sheet

-2 Mutations-

Initial Newborn Screening Results With High Immunoreactive Trypsinogen (IRT) and Two Cystic Fibrosis Mutations Identified

The California Newborn Screening Program has recently added cystic fibrosis (CF) to the panel of diseases newborns are routinely screened for. Newborns with high IRT values and 2 CF mutations are considered to have an initial positive screening result for CF.

The baby in your care was identified with 2 CF mutations. You should have received a phone call and materials from the local Newborn Screening Area Service Center (ASC). The next step is to immediately refer the baby for a confirmatory sweat test at a California Children's Services (CCS)-approved/Cystic Fibrosis Foundation (CFF)-accredited CF Center.

Cystic fibrosis is a genetic disorder that occurs when an individual inherits mutations in the cystic fibrosis gene on both chromosomes. It is a recessive condition, meaning that in order for a person to be affected, both of his or her parents must carry and pass on a cystic fibrosis mutation. People who have CF mutations on only one chromosome are unaffected by the disease. In California about 100 children are born with cystic fibrosis every year.

Symptoms result from faulty transport of sodium and chloride across the membranes of cells lining organs. The organs and systems that may be affected include the lungs, digestive tract, sinuses, sweat glands, and the male reproductive system. Lung secretions clearance can be impeded, causing inflammation and infection, leading to further deterioration of function. The digestive system, including the pancreas and liver, is affected in most individuals with CF. When pancreatic enzymes are prevented from reaching the intestines, malnutrition with poor weight gain and growth can occur. Symptoms of CF in the first few months of life may include some of the following: very salty sweat, recurring pneumonia, including severe respiratory syncytial virus (RSV) infections, failure to thrive, malnutrition, and frequent runny stools. Later there can be chronic wheezing or coughing, frequent greasy stools or difficulty with bowel movements. Symptoms can also include many other health problems such as sinusitis, nasal polyps, and abdominal pain and discomfort.

There is currently no cure for CF, but its symptoms can be treated. Early detection and appropriate treatment have been shown to improve growth and cognitive ability, to reduce hospitalizations, and may improve long-term survival. Current treatments focus on preventing malnutrition, keeping the lungs clear of thick mucous which obstructs the airways, and aggressively treating lung infections.

BENEFITS OF EARLY DIAGNOSIS

Once a child is found to have CF, treatment to prevent malnutrition and minimize lung damage can be started immediately. Imaging studies have shown that even if there are no symptoms present, lung damage occurs very early. Preventive treatment is critical in preserving lung function. Treatment typically includes nutritional supplements, respiratory therapy, and monitoring for and aggressively treating lung infections when they arise. New treatments are being explored and significant breakthroughs have been made in recent years.

Children can be diagnosed at birth with cystic fibrosis if they have meconium ileus. This occurs in about 15% of births of children with CF. For babies without symptoms at birth, identification through newborn screening will mean treatment can be initiated before significant health problems have occurred.

The sweat test is the standard diagnostic test for cystic fibrosis. All babies with two identified gene mutations should have a sweat test at a CCS-approved/CFF-accredited CF Center to confirm the diagnosis.

SCREENING STEPS IN CF IDENTIFICATION IN THE CALIFORNIA NBS PROGRAM

The initial NBS specimen collected in the hospital is used for all of the following tests:

- Immunoreactive trypsinogen (IRT) assay. IRT is an enzyme which is elevated when there is injury to the pancreas. Most newborns with CF have IRT elevations
- DNA analysis using the California CFTR mutation panel. Specimens with an elevated IRT are tested with this DNA panel. Greater than 90% of persons with CF have a mutation on the panel.
- DNA sequencing of the CFTR genes. Specimens which have only one mutation identified on the California CFTR panel are sent on for this detailed DNA analysis. This step can take 3 to 4 weeks. This testing can detect less common CF mutations and thus distinguish between those few babies in this group affected with CF and the majority who have only one mutation (carriers).

Newborn Screening Follow-up

All newborns referred to a CCS-approved Cystic Fibrosis Center by the California Newborn Screening Program are eligible for a diagnostic evaluation through the CF Center regardless of income. The Newborn Screening ASC Coordinator will work with the primary care provider in determining which CF Center to refer the baby to, based upon location and insurance plan coverage. Because the disorders screened for by the NBS Program require immediate follow-up, the CCS program has developed an expedited authorization process for the initial diagnostic evaluation at a CF Center.

Specialists at the CF Center will work closely with the primary care provider in determining what testing is needed and in the development of a treatment plan when necessary. When a disorder is confirmed, the NBS Program strongly recommends that newborns receive ongoing specialty care at a CF Center where a multi-disciplinary team (physicians, dietician, nurse, respiratory therapist, social worker, genetic counselor) can provide a comprehensive approach to assisting the family.

Additional Resources

- For more information about Newborn Screening and the most current list of CCS-CF Centers please visit our website at: { HYPERLINK "<http://www.dhs.ca.gov/nbs>" }
- For more information about cystic fibrosis and to find local resources, please visit the Cystic Fibrosis Foundation website at { HYPERLINK "<http://www.cff.org>" } and/or Cystic Fibrosis Research, Inc. at { HYPERLINK "<http://www.cfri.org>" }