INTRODUCTION
Cystic fibrosis (CF) is an autosomal recessive disease that affects the lungs and digestive system of about 30,000 children and adults in the U.S. (70,000 worldwide). A defective gene and its protein product cause the body to produce unusually thick, sticky mucus that:

• Obstructs the airway and leads to life-threatening bronchiectasis
• Affects the pancreas and stops enzymes from helping absorption of food
• Impacts other organ systems affecting quality of life
In the 1950s, few children with CF lived to attend elementary school. Today, advances in research and medical treatments have further enhanced and extended life for children and adults with this disease. Many people with CF can now expect to live into their 30s, 40s and beyond.

The Cystic Fibrosis Foundation is a nonprofit donor-supported organization dedicated to the development of new drugs to fight the disease, improve the quality of life for those with CF and ultimately to find a cure. The foundation has supported a better understanding of the disease and improved patient care through the development of a national database, PortCT. More than 110 institutions accredited by the foundation (including Children’s Hospital of Wisconsin) gather important information from every patient at every visit. This data is entered into the database and outcomes are tracked and trended. The database provides the ability to compare one center to another and to indicate which areas of care need improvement.

**DIAGNOSIS**

Newborn screening for CF in Wisconsin began in 1994. Although many newborns will have a positive newborn screen for the disease, fewer than 10 percent will actually have it. Research has proven that infants diagnosed with CF through newborn screening and treated before symptoms begin are healthier.

The newborn screen for CF is a two-tier process. The first tier is the analysis of immunoreactive trypsinogen (IRT) using the Guthrie card. If the IRT is elevated, the second tier is diagnostic for CF with either gene analysis by the State Lab of Hygiene or sweat chloride testing at a CF center. This report is then sent to the primary care provider. Positive screening with either an elevated IRT or CF gene identification needs follow up at a CF center and facilitated by the primary care provider. (See Chart 1.)

**Chart 1 – Abnormal Newborn Screen**

- Elevated IRT with no mutations
  - Primary care provider to monitor for clinical signs of CF:
    - Persistent diarrhea
    - Poor weight gain
    - Chronic cough
    - Respiratory problems
  - If these signs appear or there is a family history of CF, contact a CF specialist.

- Elevated IRT with one CF mutation
  - Sweat test at CF care center

- Elevated IRT with two CF mutations
  - Diagnosis of CF
  - For infants up to 6 months, a sweat chloride of:
    - < 29 mmol/L - CF unlikely
    - 30 to 59 mmol/L - possible CF
    - > 60 mmol/L - diagnosis of CF
  - Refer to CF care center for follow-up
MANAGEMENT

The treatment of CF requires a collaborative and interdisciplinary approach. The aspects of CF treatment include airway clearance, inpatient and outpatient antibiotics, pancreatic enzyme replacement, proper nutrition and psychosocial support. Anticipatory guidance and routine well-child care, including immunizations, are managed by the primary care provider. Treatment should follow a collaborative care model with open communication between the family, primary care provider and the CF center. Care provided by the primary care provider and CF center should complement each other.

Treatment recommendations for the primary care setting

• Infants with CF need supplemental salt and older children are taught to salt their food
• The goal for infants with CF is to be at or above the 50th percentile weight-for-length
  ◦ Encourage a high-fat diet, including use of whole milk
  ◦ Inform the CF center if there is any lack of weight gain or weight loss
• Early symptoms can be subtle; call the CF center for any pulmonary or gastroenterology symptoms, such as cough or wheeze
  ◦ Antibiotics are initiated at a lower threshold in children with CF and continued for a longer period of time than in other children
• Life expectancy is steadily increasing, but this depends on daily preventive care
  ◦ Convey hope: We expect these infants to lead full adult lives
  ◦ Inquire about adherence to the prescribed CF regimen at each primary care visit
  ◦ Reinforce limit-setting (especially important in children who require daily treatments but may be perceived as vulnerable)
• The diagnosis creates psychosocial challenges
  ◦ There should be no person-to-person interactions with others with CF to prevent cross-infection
  ◦ CF places the entire family under stress
• Ask how unaffected siblings feel about CF
• Share insights on family functioning with CF center team

References


REFER A PATIENT

To refer a patient, visit chw.org/refer or call toll-free (800) 266-0366.

CALL FOR AN APPOINTMENT

To make an appointment, call Central Scheduling toll-free at (877) 607-5280.

For more information, visit chw.org/cf