Treating Cystic Fibrosis

Early Intervention, Team Approach Improving Quality of Life for Children with Disorder

by Sue Emond

Cystic fibrosis, a chronic, progressive disease that primarily affects the lungs and digestive system, afflicts 30,000 children and adults in the United States, and 70,000 worldwide. According to the American Lung Association, it's the second most common inherited, life-threatening, childhood onset disorder in the United States, just behind sickle cell anemia. About 1,000 new cases of cystic fibrosis are diagnosed each year.

Yet thanks to devoted research teams from leading organizations like The Children's Hospital Cystic Fibrosis Program, along with the ongoing dedication of national organizations, especially the Cystic Fibrosis Foundation, the lives of cystic fibrosis patients continue to improve. In fact, 50 years ago, children with cystic fibrosis typically failed to reach grade school. But now, those afflicted with the disease often live into their 30s, 40s, and even longer. In 2006, the predicted median age of survival for a cystic fibrosis patient was 37 years. Through innovative research, substantial improvements in measures of malnutrition and lung function, and a diverse and comprehensive approach to care, cystic fibrosis patients can today experience a quality of life unforeseen even a few decades ago by those with the disease.

Understanding Cystic Fibrosis

Cystic fibrosis occurs in children who inherit the gene from both parents—who are each carriers of the cystic fibrosis gene but who do not have cystic fibrosis themselves. Fortunately, Colorado state law requires that hospitals in Colorado screen all newborn babies for cystic fibrosis. Coloradoans can thank pioneers like Dr. Frank Accurso, director of the Cystic Fibrosis Program at The Children's Hospital, along with several of his colleagues, for the benefits of early diagnosis and treatment. Their aggressive research in support of newborn screening for cystic fibrosis back in 1987 led to Colorado,becoming the first state to adopt a statewide newborn screening requirement. Nationwide, more than 70 percent of patients are diagnosed by age 2. But now, the U.S. Department of Health and Human Services Centers for Disease Control and Prevention recommends that all states require newborn screening for cystic fibrosis.

Symptoms of cystic fibrosis can occur at birth or sometimes later, and can be mild or severe. Children with cystic fibrosis face various challenges, especially respiratory and digestive problems since the defective gene causes the body to produce abnormally thick mucus. The mucus clogs the lungs, causing breathing difficulties, as well as recurring lung and sinus infections. Cathy Lingard, MS RD, a registered dietician specializing in cystic fibrosis at The Children's Hospital, says the thick mucus also obstructs the ducts of the pancreas, leaving digestive enzymes unable to reach the intestines. So cystic fibrosis patients don't absorb food nutrients well and have difficulty gaining and maintaining weight—in stark contrast to today's pediatric obesity epidemic. Lingard notes that cystic fibrosis children particularly can't digest fats and the fat soluble vitamins, including A, D, E, and K. Other symptoms include reproductive problems, irregular nail beds of the fingers and toes, nasal polyps, and a high concentration of sodium in the sweat.

CYSTIC FIBROSIS SYMPTOMS

- Persistent coughing, at times with phlegm
- Frequent lung infections
- Wheezing or shortness of breath
- Poor growth/weight gain in spite of a good appetite
- Frequent greasy, bulky stools
- Very salty-tasting skin

Source: The Cystic Fibrosis Foundation

Dalyn Shkapich blows into a pneumotach, a device used to measure respiratory flow.

Aggressive Treatment for a Better Life

The Cystic Fibrosis Program at The Children's Hospital provides an agressive approach to cystic fibrosis. Both infants and older children receive extensive diagnostic testing at regular visits, and patients are monitored every two to four weeks initially, monthly for the first six months, then every two months up to one year. After that, most patients are monitored every three months throughout their lives.

According to Dr. Scott Sagel, associate director of the Cystic Fibrosis Program at The Children's Hospital, care providers regularly monitor the nutritional and pulmonary status of patients through in-depth testing, assessing problems like malabsorption, maldigestion, and wheezing. Dr. Sagel insists that "frequent assessment and early intervention are key. If there's a change in clinical status we can intervene early. This allows us to be as aggressive as possible in their care."

Kids with cystic fibrosis can't help but make lots of new friends at The Children's Hospital, given the program's multidisciplinary, collaborative approach. An experienced staff of doctors, nurses, dieticians, social workers, child life therapists, respiratory therapists, physical therapists, and others provide comprehensive evaluation and management of cystic fibrosis, while also encouraging family involvement. Today, the Cystic Fibrosis Center treats 600 patients throughout the region, including children at The Children's Hospital and adults at National Jewish Medical and Research Center in Denver.

Cystic Fibrosis—A Family Affair

Though diagnosis and evaluation for children with cystic fibrosis takes place at The Children's Hospital, ongoing treatment typically happens at home. Every day these kids need a variety of at-home treatments, including drugs, airway clearance techniques, special diets, and more. Sagel and Lingard describe some of the typical respiratory and dietary treatments prescribed by the Cystic Fibrosis Center at The Children's Hospital:

- Nutritional intervention – including digestive enzymes, antacids, and nutritional supplements
- A high-calorie diet – typically high in fats, that surpasses the minimum RDA for calories in normal children by 20-50 percent
- Antibiotics – to treat bacteria that frequently colonize and infect the airways, resulting in chronic bacterial bronchitis
- Airway clearance techniques – done one to four times per day to improve lung function and reduce the severity of lung infections, they include chest physiotherapy, where parents and others clap on the child's chest; handheld mucus-clearing devices; and external devices such as vests that vibrate externally to loosen phlegm
- Inhaled mucolytic agents – taken by nebulizers to loosen and liquefy mucus secretions

Without such treatments, cystic fibrosis was a fatal early childhood condition. Dr. Sagel says that improvements in survival are due to "better antibiotic options, enhanced mucus clearance technologies, and regular care at accredited cystic fibrosis care centers," like the Cystic Fibrosis Center at The Children's Hospital.

Instilling Hope through Promising Research

Churee Pardee, APN, an advanced practice nurse and research coordinator for the Cystic Fibrosis Program at The Children's Hospital, says the program currently is involved in close to 40 key cystic fibrosis studies—with particularly promising research in the areas of gene therapy and protein rescue therapy. "Children with cystic fibrosis are born with different proteins than those without cystic fibrosis, so we're investigating exactly what those differences are," says Pardee. The program also is involved in several important drug studies, such as trying to develop a drug that will help the defective cystic fibrosis gene work better.

The Cystic Fibrosis Center has no problem finding willing subjects for its research and drug trials. Pardee notes that almost all Cystic Fibrosis Program patients are involved in research, either directly or indirectly. Some simply provide their patient data, while others offer regular specimens, participate in extended drug trials, or are willing to try new procedures. These patients are critical to keeping this important research moving forward.

According to Dr. Sagel, all current treatments treat the consequences of cystic fibrosis, including airway infection, inflammation, and mucus plugging. And though we can't prevent cystic fibrosis today, The Children's Hospital Cystic Fibrosis Center research team, along with others dedicated to improving the quality of life for these patients or even finding a cure, is involved in early-phase human clinical trials of potential disease-modifying treatments. Excitedly, Dr. Sagel says these treatments have the potential to prevent the downstream consequences of cystic fibrosis. "They represent hope for these patients," he says.

Pulmonary specialist and Director of the Cystic Fibrosis Program at The Children's Hospital, Frank Accurso, visits with Devin, Daphne and Dalyn Shkapich