

# Cystic Fibrosis Care

*at Stamford Hospital's Tully Health Center*





## Our Team:

Our multi-disciplinary medical team has the specific expertise you need. Led by Dr. Hossein Sadeghi, Director, Stamford Hospital's Cystic Fibrosis Center provides specialized pediatric nursing care and access to a medical team, which includes: a Pediatric Pulmonologist, Gastroenterologist, Endocrinologist and Infectious Disease Specialist. The team also includes a social worker, dietitian, respiratory therapist and pulmonary technician as well as genetic counseling.

An accredited sweat laboratory performs sweat testing three days per week. Stamford Hospital will begin neonatal screening on October 1, 2007. Pediatric pulmonary function testing includes spirometry, plethysmography and exercise testing.

## What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disease caused by a recessive gene. Both parents have to be carriers of the gene for a child to get the disease. It is characterized by abnormally thick secretions caused by the body's high concentration of salt. There is an abnormality in the function of a cell protein called the Cystic Fibrosis Trans Membrane Regulator (CFTR). CFTR controls the flow of chloride in and out of the body's cells. As the movement of salt and water in and out of the body's cells is altered, mucus becomes thickened. This thickened mucus can affect many organs and body systems including:

- Respiratory—sinuses, lungs and nasal passages
- Digestive—intestines, pancreas, liver, gallbladder
- Reproductive—absence of vas deferens in males and thickening of cervical mucus in females
- Sweat glands

Cystic fibrosis is a chronic, progressive and usually fatal disease. Median life expectancy for children with CF has been increasing and is currently close to 37 years of age.

Approximately one in 31 people in the US are carriers of the cystic fibrosis gene. These people are asymptomatic and usually do not know that they are carriers.

There are about 30,000 people in the US who are affected with the disease, and about 1,000 babies are born with it each year. It occurs more commonly in Caucasians with northern European heritage and less frequently in African-Americans, Asian Americans and Native Americans.

Cystic fibrosis is a complicated illness that requires clinical care by a physician and other healthcare professionals. Multi-disciplinary teams organized into CF centers, like the one here at Tully Health Center, are recommended for the care of CF patients and have been shown to result in improved health and longer lives for CF patients.

### **What are the signs and symptoms of cystic fibrosis?**

Infants born with cystic fibrosis usually show symptoms within the first year of life. Some children, though, may not show symptoms until later in life. The following signs and symptoms are suspicious of CF, and infants and children having these signs may be tested for CF:

- Malnutrition and failure to thrive
- Poor growth
- Diarrhea that does not go away
- Foul-smelling or greasy stools
- Frequent respiratory infections, persistent cough, frequent wheezing or episodes of pneumonia
- Salty tasting skin
- Dehydration and fluid loss especially on hot summer days

### ***Other medical problems may include:***

- Sinusitis
- Nasal polyps
- Clubbing of fingers and toes—a condition where the ends of the fingers and toes become enlarged; more prevalent in the fingers

- Pneumothorax—the presence of air or gas in the pleural cavity causing the lung to collapse
- Hemoptysis—coughing blood
- Abdominal pain and gas in the intestines
- Rectal prolapse
- Liver disease
- Diabetes
- Pancreatitis

Individuals may experience symptoms differently and the symptoms of cystic fibrosis may resemble other medical conditions. Consult your physician for a diagnosis.

### How is cystic fibrosis diagnosed?

CF is diagnosed by the presence of clinical symptoms as well as either the presence of genetic mutations from blood or cheek scraping cells or a positive sweat test.

In addition to a complete medical history and physical examination, diagnostic testing for cystic fibrosis may include the following:

- Sweat chloride test—a test to measure the amount of chloride in the sweat. The test is performed by stimulating sweat with a mild electric current, which does not cause pain or harm to your child. The sweat is collected and analyzed. Higher than normal amounts of chloride may suggest cystic fibrosis.
- Genetic testing (DNA analysis)—blood or cheek scraping cells can be tested for mutations in the CFTR gene.

### Treatment for cystic fibrosis

The goals of treatment are aimed at maintaining normal growth and lung function as well as easing the severity of symptoms and slowing the progression of the disease. In general, treatment is focused on controlling mal-absorption with pancreatic enzyme supplementation, vitamins and appropriate diet and improving or maintaining airway clearance with chest physical therapy and inhaled medications.

Specific treatment for cystic fibrosis will be based on your child's age, overall health and the extent of the disease but may include:

*Management of problems that cause lung obstruction may include:*

- Chest physical therapy (to help loosen and clear lung secretions; this may include postural drainage and devices such as a percussor or flutter, which vibrate the chest wall and loosen secretions)
- Exercise (to loosen mucus, stimulate coughing and improve overall physical condition)
- Medications such as bronchodilators and anti-inflammatory agents to reduce mucus and inflammation and improve breathing
- Antibiotics (to treat infections)

*Management of digestive problems may include:*

- Appropriate diet
- Pancreatic enzymes to aid digestion
- Vitamin supplements
- Treatments for intestinal obstructions

### *Psychosocial support:*

- Helping patients and their families deal with issues such as independence, compliance with treatments, explaining CF to children, family, friends and teachers, finances, relationships, sexuality and sterility.

With medical and psychosocial support, many children with CF can thrive and lead a productive life.

### **Clinical Research**

At the present time, there is no cure for CF; however, research in gene therapy is being performed. The gene that causes CF has been identified and there are hopes that this will lead to an increased understanding of the disease. Studies are underway to find a cure for CF by modifying the defective gene at multiple levels. Medications and other therapies are being researched to help slow the progression of the disease until a cure is discovered. These efforts have resulted in greatly improved life expectancy over the past 20-30 years.

As a Cystic Fibrosis Center, Stamford Hospital will participate in multi-center research projects in order to improve the care of CF patients and work toward finding a cure.

## Contact Us

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