Cystic Fibrosis (CF)

Also known as:

- cystic fibrosis of the pancreas
- fibrocystic disease of the pancreas
- mucoviscidosis

What is CF?

CF is an inherited condition that predominantly affects the lungs, pancreas, intestine, liver, sweat glands and male reproductive tract.

What causes CF?

CF is caused by mutations in the cystic fibrosis conductance regulator (CFTR) gene which results in absent or deficient CFTR protein. CFTR is responsible for chloride transport in the body. Defective chloride transport leads to thick mucous secretions in the lungs, resulting in airway obstruction and recurrent respiratory infections. Viscous secretions in the pancreas lead to pancreatic duct blockage, resulting in malabsorption and nutritional deficiencies.

How common is CF?

The incidence of CF is about 1 in every 3,600 infants born in Canada. CF is more common in Caucasians and people of northern European ancestry.

What are the clinical features of CF?

CF usually presents in infancy with failure to thrive secondary to malabsorption and/or respiratory symptoms, such as cough and wheeze. A minority of CF patients present in the newborn period with intestinal obstruction due to meconium ileus. The clinical course of CF is variable. Patients are prone to recurrent respiratory infections and may develop lung damage. Later complications may include diabetes and liver disease. Almost all males with cystic fibrosis have infertility due to congenital bilateral absence of the vas deferens.

What is the screening test for CF?

Screening for CF is done in several steps including immunoreactive trypsin (IRT) testing and DNA analysis. When IRT levels on the newborn blood spot screen are elevated, the sample is sent for DNA analysis for the most common European mutations in the CFTR gene. Newborn blood spot screening is not 100% perfect. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of CF is confirmed by sweat chloride testing and may involve other testing or more extensive DNA analysis. Most children with CF have an increased sweat chloride concentration. Sweat chloride testing involves a special device that stimulates a small part of the infant's arm or leg to sweat over a 30 minute time period and results are then analyzed. Specialists at the clinics listed below will arrange for diagnostic testing.

How is CF treated?

Treatment of CF involves improving nutrition and treating respiratory problems. Pancreatic enzyme supplements are given to improve absorption of nutrients, and additional vitamins and salt are added to the child's diet. Respiratory problems are treated by anti-microbial agents and medications to clear the thick mucous secretions. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. When respiratory infections occur, prompt treatment is necessary to prevent lung damage. The treatment is life-long.

What is the outcome of treatment for CF?

Early introduction of treatment for CF is associated with improved growth, lung function and has a long-term beneficial effect on quality of life for the child and family.

Is CF inherited?

CF is inherited as an autosomal recessive trait. Parents of a child with CF are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. CF carriers are healthy. Genetic counselling and prenatal testing is available to all families with CF.

For additional resources, please call:

Edmonton Pediatric Cystic Fibrosis Clinic

Stollery Children's Hospital
2E2.24
8440 – 112 Street NW
Edmonton, AB T6G 2B7
Phone: 780-407-8341
Fax: 780-407-4927

Calgary Pediatric Cystic Fibrosis Clinic

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7319
Fax: 403-955-7527

Molecular Diagnostic Laboratory

8-26 Medical Sciences Building
8440 – 112 St. NW
University of Alberta
Edmonton, AB T6G 2H7
Phone: 780-407-1434
Fax: 780-407-1761