

BC Newborn Screening Program

Information Sheet

Cystic Fibrosis (CF)

What is Cystic Fibrosis (CF)?

Cystic fibrosis (CF) is an inherited condition. Problems arise as certain organs become clogged with thick mucous secretions. CF occurs when a protein called the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR), is not working properly. CFTR is a chloride channel that allows chloride (salt) and water to move out of certain cells of the body.

What is its incidence?

CF affects about 1 in every 3,600 babies born in BC.

What causes the disease?

Alterations, or changes, in the CFTR gene result in chloride channel protein that is not working well or is absent.

What are the clinical features of the disease?

Children and adults with CF can develop breathing problems because the lungs are clogged with mucus. This makes individuals more susceptible to respiratory infection, which can cause inflammation and damage to the lungs.

The ducts of the pancreas that the body needs to release digestive enzymes can also be blocked with thick mucous secretions. Therefore, people who have CF may have trouble getting enough nutrition from the food that they eat. Babies with CF can have large, greasy bowel movements. Children with CF who do not have adequate nutrition do not gain weight or height at a

normal rate and may be thinner or smaller than other children the same age.

The sweat glands are also affected in CF; individuals with the disease will have sweat that is much saltier than usual. Individuals with CF (especially males) may have reduced fertility.

The presentation of CF is variable, with mild to severe forms of the disease. For example, there are individuals with the disorder who have breathing problems only, or who have digestive problems only, or are seemingly symptom-free until their reproductive issues become apparent.

How is the diagnosis confirmed?

BC uses a multistep approach for the screening and diagnosis of CF. Initial testing of bloodspot immunoreactive trypsinogen (IRT) is followed by DNA testing for a panel of common mutations using the same bloodspot. Depending on the results of both of these tests a second bloodspot card may be requested for IRT testing at day 21 of life. The final diagnosis of CF is confirmed by doing a sweat test. This involves collecting sweat from a person's arm and measuring the amount of salt (chloride) in the sweat. If required, this testing is arranged by the Cystic Fibrosis Clinic at BC Children's Hospital and will require travel to BC Children's Hospital in Vancouver.

What is the treatment of the disease?

Breathing exercises and physical therapy are done regularly to help clear the lungs of

thick mucous secretions. Antibiotics may also be given as a preventive measure against respiratory infections.

Replacement of pancreatic enzymes, usually by taking capsules with meals, is an effective means of improving digestion and ensuring adequate growth and nutrition. Extra supplemental vitamins are often taken as well.

What is the outcome of treatment?

Infants who have CF can benefit significantly from early treatment, and their condition requires lifelong management and monitoring. Although some people with CF may have a relatively good prognosis, others - depending on disease severity - may have more serious medical issues or complications, and a shortened life expectancy. Those who have severe lung complications may require lung transplantation.

Can a family have more than one child with CF?

CF is inherited as an autosomal recessive disease. Parents of a child with CF are assumed to be 'carriers' for the disease and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with the condition. Prenatal testing for CF can be done as early as 10-12 weeks of pregnancy. Since there is effective treatment, and as the prognosis of individuals who undergo treatment is more favourable, genetic counselling may be recommended to discuss the possible benefits of prenatal testing.

Unaffected siblings of a child with CF have a 2/3 chance of being carriers. CF carriers are healthy and do not have symptoms of CF.

Resources

<http://www.cysticfibrosis.ca/>

<http://genetests.com>

<http://pediatrics.aappublications.org/cgi/content/full/119/2/e495>

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