NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Cystic Fibrosis

What is Cystic Fibrosis?

Cystic fibrosis (CF) is an inherited disorder that causes thick mucus to build up in the lungs, digestive system (and pancreas) and other organs. Most people with CF get frequent chest infections. They may also have problems digesting their food and, as a result, may not gain weight as well as they should. The presentation of CF is variable, with mild to severe forms of the disease.

How common is CF?

About 1 in every 4,300 babies born in BC is affected.

What causes the condition?

People's bodies are made up of tiny building blocks called cells. Inside the cells are tens of thousands of instructions called genes. A gene is a set of instructions (like a recipe) on how to make a protein. If the gene/recipe has a major change in it then the protein is no longer made or does not work properly. These gene changes (called disease-causing variants) can affect how our body grows, develops, and functions.

We all have two copies of each gene, including the CF gene (CF Transmembrane Conductance Regulator - CFTR), one copy from each parent. Children and adults who have CF have changes (disease-causing variants) in both copies of the gene.

What is the screening test for CF?

The initial test is a protein in the blood spot called immunoreactive trypsinogen (IRT). If the IRT result is high (3% of results), a DNA test is performed on the same blood spot card. The DNA test looks for a panel of common disease-causing variants in the CFTR gene. Most babies who have the DNA test will not have any CFTR variants and no further testing is required. If one or two variants are found in the CFTR gene, a second bloodspot card may be requested for IRT testing at day 21 of life, or other testing will be arranged to rule CF in or out.

What is the treatment and outcome of CF?

Infants who have CF can benefit significantly from early treatment. Treatment for CF may include airway clearance and physiotherapy, pancreatic enzyme supplements, medications to help fight or prevent infections and in some cases, medications to correct the non-working protein made by the CF gene.

Where can I get more information?

Talk to your family doctor. You may also call the CF Newborn Screening Team at 604-875-2623.

Information about the BC Newborn Screening Program can be found at
www.newbornscreeningbc.ca

Information about CF can be found at:

- Canadian Cystic Fibrosis Foundation
  www.cysticfibrosis.ca/
- Canadian Cystic Fibrosis Foundation (CCFF), Vancouver & Lower Mainland Chapter
  www.cfvancouver.ca/
- For a listing of contacts for other CCFF chapters in BC:
  www.cfvancouver.ca/
- GeneTests (hosted by the National Center for Biotechnology Information (NCBI))
  www.genetests.org

This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis or treatment. Always talk to your health care provider about specific health concerns.

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.