NEWBORN SCREENING BC

Information sheet

My baby had a positive Newborn Screening Result for CFSPID

What does this mean?

What is CFSPID?

CFSPID stands for "Cystic Fibrosis Screen Positive Inconclusive Diagnosis". This means your baby has a positive newborn screening result but does not meet all the criteria for a Cystic Fibrosis (CF) diagnosis. Children with CFSPID are expected to lead longer and healthier lives than those with CF. However, it is not known if there is a risk of developing a CF-like disease later in life.

How do you diagnose CFSPID?

Your child may be diagnosed with CFSPID for the following reasons:

- Two CF gene variants are found and at least one of these variants has unknown or unclear consequences. The sweat test is normal or borderline.
- One or no CF gene variants are found and the sweat test results are borderline on more than one occasion.

How can CFSPID affect the body?

Although most people with CFSPID will remain healthy, some may experience milder symptoms of CF disease.

- Lungs: All children are at risk of developing chest infections, but for children with CFSPID, these may take longer to clear and may need treatment with antibiotics.
- Pancreas: children with CFSPID may develop inflammation of the pancreas (a digestive gland).
- Reproductive system: if they are male, people with CFSPID may be diagnosed with infertility as an adult.

The start of symptoms is unknown. We assume that this, if at all, may occur later in life. However, in some individuals, this may be earlier.

It is possible that some children may develop true CF disease later on in life. 1 in 10 babies with CFSPID may meet criteria for CF as they get older.

How are children with CFSPID monitored?

Your CF health care provider will use a number of tools to monitor the health of your child. This is important, as early treatment of potential symptoms will help to keep your child healthy. Your CF clinic will arrange regular appointments and assessments of your child. Your CF team can provide you with more information.

Please report to your CF team if your child has any of the following symptoms:

- Stops gaining weight
- Changes in your child's stool (poops) – loose, foul smelling, or has mucous
- Sudden severe stomach pain and vomiting
- A wet cough that is not going away

How frequent is CFSPID?

The true frequency of CFSPID is still unknown. It is estimated that for every three babies diagnosed with CF through newborn screening, there is one CFSPID diagnosis. About 20 babies per year are diagnosed with CFSPID in Canada.

There are other terms used across the world for a diagnosis like CFSPID. In the United States the Cystic Fibrosis Foundation (CFF) uses the term CRMS (CFTR Related Metabolic Syndrome).

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.

How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. If you are feeling overwhelmed or have questions, you can talk with your healthcare provider to discuss supports available or call the CF Newborn Screening Team at 604-875-2623. A list of supports is attached.

Where can I get more information?

Talk to your primary care provider. 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/**
- **CF Newborn Screening Parent Information Sheet**
- 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