WHAT IS CFSPID?

What does this mean for my Child?

Your Baby has been diagnosed with CFSPID. What does this mean?

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?

In Canada, most babies have a CF newborn screening test in the first few days of life. CF newborn screening involves measuring a marker (Immuno Reactive Trypsinogen: IRT) in a blood sample taken from the baby's heel. Those babies with a high IRT will go on to have a genetic test that looks for the genes responsible for causing CF. Depending on the province's newborn screening program, many infants will undergo a sweat test. There are three possible outcomes from the sweat test:

- A positive result (chloride >60mmol/L)
- A negative result (chloride <30mmol/L)
- A borderline result (30-60mmol/L).

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

a) Two CF genes are found and at least one of those genes has unknown or unclear consequences. The sweat test is normal or borderline.

b) One or no CF genes are found and the sweat test results are borderline on more than one occasion.

Diagnosis	CF	CFSPID	
Number of genes found	1 or 2	1 or 0	2
Sweat test result	Positive	Borderline	Normal or Borderline
Symptoms of CF	Classic	Variable	Variable

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).

Resources

www.cysticfibrosis.ca
www.cff.org

Find your local CF Clinic:

http://www.cysticfibrosis.ca/our-programs/healthcare/how-cf-care-is-delivered/cf-clinics-in-canada

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How Can CFSPID affect the body?

With widely available genetic testing, more is being discovered about the genes that are found with CFSPID. Although most people with CFSPID will remain healthy, some may experience milder symptoms of CF disease. It is possible that some children may develop more significant evidence of CF disease later in life. Currently, there is not enough information available to predict who will remain healthy and who will develop CF disease.

If your child was to develop symptoms of CF disease, following areas of the body can be affected: lungs, digestive and reproductive systems.

All children are at risk of developing chest infections, but for children with CFSPID, these may take longer to clear and require treatment with antibiotics. Additionally, children with CFSPID may develop inflammation of the pancreas (a digestive gland) or, if they are male, may be diagnosed with infertility as an adult. These are the main body areas that can be affected in CFSPID, but your CF health care providers can provide you with more information.

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

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.

How are children with CFSPID monitored?

It is important to regularly monitor children with CFSPID in a CF Clinic. People with CFSPID may have similar, milder symptoms of CF disease. Importantly, 1 in 10 babies with CFSPID may meet criteria for CF as they get older. It is essential the CF Clinic monitors your child so early treatment and interventions can begin if necessary.

Your CF clinic will arrange regular appointments and assessments for your child. Your CF Clinic can provide you with more information.

Please report to your CF Center if your child

- stops gaining weight
- has loose stools, stools with abundant mucus or, very foul-smelling stools
- Your child develops acute severe stomach pain and vomiting, which is not explained by a stomach flu
- Your child has a wet or long-lasting cough
- Your child has frequent lung or sinus infections

How do you feel?

Many parents say they feel shocked, scared, and confused when they are told their child has CFSPID. These are normal feelings. If you are feeling overwhelmed or have questions, you are encouraged to talk with your CF healthcare provider or local physician.

Developed by the CF Clinics at BC Children's Hospital Vancouver and Hospital for Sick Children Toronto, in collaboration with the CF Newborn Screening Working Group