What is Cystic Fibrosis Screen Positive Inconclusive Diagnosis (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 diagnosis. Children with CFSPID are expected to lead long healthy lives. However, it is not known if these children are at risk of developing a cystic fibrosis like disease later in life.

How do you diagnose CFSPID?

In Australia, most babies have a cystic fibrosis newborn screening test in the first few days of life. Cystic fibrosis 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 cystic fibrosis. The babies will then 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 cystic fibrosis 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 cystic fibrosis genes are found, and the sweat test results are borderline on more than one occasion.

How frequent is CFSPID?

The true frequency of CFSPID is still unknown. It is estimated that for every three babies diagnosed with cystic fibrosis through newborn screening, there is one CFSPID diagnosis.

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 cystic fibrosis disease. It is possible that some children may develop more significant evidence of cystic fibrosis disease later in life. Currently, there is not enough information available to predict who will remain healthy and who will develop cystic fibrosis disease.

If your child was to develop symptoms of cystic fibrosis disease, the 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 cystic fibrosis team 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. The cystic fibrosis team will use several 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 cystic fibrosis clinic, this may be 6-12 monthly depending on your child's age. One in 10 babies with CFSPID may meet criteria for cystic fibrosis as they get older. It is essential for the cystic fibrosis team to monitor your child so early treatment and interventions can begin if necessary. Your cystic fibrosis clinic will arrange regular appointments and assessments for your child. Your cystic fibrosis Clinic can provide you with more information.

Please report to your cystic fibrosis clinic if your child:

- stops gaining weight
- has loose stools, stools with abundant mucus or, very foul-smelling stools
- develops acute severe stomach pain and vomiting
- has frequent lung or sinus infections.
- has a wet or long-lasting cough

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 cystic fibrosis team or your GP.

Your Cystic Fibrosis Clinic

There is a large multi-disciplinary team at Queensland Children's Hospital, Brisbane, all of whom are involved in providing you and your child with the best possible care.

Switchboard 07 3068 1111

Appointment bookings 07 3068 2300

Acknowledgment to Cystic Fibrosis Clinics at British Columbia Children's Hospital Vancouver and Hospital for Sick Children Toronto.