

Cystic Fibrosis Newborn Screening (CF NBS) Frequently Asked Questions (FAQ)

1. What is a “positive CF NBS” and what does it mean?

It means that the immunotrypsinogen assay (IRT) tested in the upper 97.8% percentile for the day of the test. This triggered DNA analysis for the 46 most common CF genes in our population. That test was positive for at least 1 CF gene mutation. **This test is only done on the first sample.**

2. Does that mean my patient has CF?

That means that child possesses at least 1 common gene for CF gene mutation. It also means one of their parents also must carry the gene. Your patient is at least a carrier of one CF gene mutation.

3. I thought you needed 2 gene mutations to have CF?

This is true CF is an autosomal recessive disease. **A child could have one common CF gene mutation and one uncommon CF gene mutation.** Only the common CF gene mutation would be detected in the newborn screen. This panel of 46 common CF gene mutations accounts for about 92% of CF disease. At this time there are over 1400 known CF gene mutations. That’s why the positive NBS needs further evaluation.

4. Why don’t we just send a genetic test for DNA?

Unless you test for all the genes with expanded testing (which takes weeks and is very expensive) you will not have a definitive answer. Sometimes after the genetics are obtained the answers still remain unclear, the patient may have a new novel gene and may even involve testing other family members as well. It is an expensive process that can delay an accurate diagnosis.

5. So the patient has a +NBS; what should I do now?

At a minimum the child needs to obtain a sweat chloride test at a qualified testing center facility. Currently, those centers are at Phoenix Children’s Hospital and Saint Joseph’s Hospital in Phoenix and University Medical Center in Tucson.

6. What will happen at the NBS clinic visit?

After you receive a call from the CF center regarding the +NBS, a test can be arranged, including follow up management of the patient and testing. The child may be seen at the time of the sweat chloride exam. During this visit the child is assessed for early subtle signs of CF. The family is also given accurate information about CF and given genetic counseling for family regarding the CF gene mutation. This service includes information back to your office about the testing and any further implications for your patient and

their family. Providing accurate and rapid testing for your patient and information to you and your patient's family will follow.

Please have your referral coordinator obtain an authorization for a both a visit/consultation and testing. The ICD-9 code is 796.6.

7. If a child had a positive CF NBS what should we do about a sibling?

The CF NBS in Arizona started in 10/07. If a child has a + NBS for CF, any siblings that are symptomatic (including growth issues, asthma, recurrent pneumonias or sinus disease) should have a sweat chloride.

8. Does the NBS detect all CF patients?

CF NBS the detection rate is about 95%. *Meaning 8 % of children could be lost to care through a variety of reasons.* The program detailed above is an attempt to limit some of the extraneous factors causing a delay to diagnosis or misinformation about CF.

9. Now that there is a CF NBS can I take CF out of a patient differential diagnosis?

Absolutely not, as mentioned above CF NBS could miss 5 % of diagnoses. This means that any child with, for example, failure to thrive, recurrent pneumonia, chronic diarrhea or malabsorption it should be left high in the differential. *CF is a common illness present in 1/3100 in the Caucasian population.* Please obtain a sweat chloride for that child; it is an easy, inexpensive test that can aid your diagnostic process.

10. If I have further questions who can I call?

- Pediatric CF and NBS director Dr Adrian O'Hagan Children's Hospital, Department of Pediatric Pulmonology (602) 546-0985.
- Dr. Wayne Morgan, Tucson Cystic Fibrosis Center at (520) 626-7780.
- A pediatric pulmonologist of your choice

Another source of accurate information regarding CF is the Cystic Fibrosis Foundation (www.CFF.org).

Thanks to Dana Valletta from PCH CF Center for creating this valuable resource. She can be reached at 602-546-0985