Newborn Screening Education for Health Care Providers

YOUR PATIENT HAS HAD A POSITIVE CYSTIC FIBROSIS NEWBORN SCREEN TEST

Testing for cystic fibrosis (CF) has been added to your state's Newborn Screening (NBS) panel. While many newborns may have a positive NBS for CF, it is expected that fewer than 10% (about 1 in 10) of newborns with a postive CF screen will actually have CF. The time between a positive screen and testing to confirm a CF diagnosis is stressful for parents and families. Therefore, consider informing parents that a positive screen for CF means the newborn may have CF but that most do not. Healthcare providers need to be aware of this stressful time as they encourage or assist parents to obtain the appropriate follow up tests, without causing undue alarm.

NEWBORN SCREENING PROCESS

CF NBS is a two tiered process. The first tier is analysis of immunoreactive trypsinogen (IRT) and is performed on a blood spot from the standard Guthrie card. If the IRT is elevated then the second tier is a diagnostic test for CF. This is either a sweat test or PCR for gene mutations.

All infants with a positive IRT should be referred to a CF Foundation-accredited care center for further evaluation and diagnostic testing. Sweat testing for elevated chloride levels should be done at a CF care center. For infants up to 6 months of age, a sweat chloride level of: $\leq$29 mmol/L, CF unlikely; 30 to 59 mmol/L, possible CF; and $\geq$60 mmol/L, diagnosis of CF. Babies with a sweat chloride level of 30 and above should be referred to a CF care center for follow-up. If two known CF mutations are identified, this is consistent with a diagnosis of CF and the family should be referred to a CF Foundation-accredited CF care center. Keep in mind that a negative NBS does not necessarily mean that the child is negative for CF. Children with whom you have concerns should be referred to a CF care center for further evaluation. To locate the nearest CF Center go to the CF Foundation Web site (www.cff.org) and click on Care Center Network found in the Quick Links menu.

FOLLOW UP FOR CF NEWBORN SCREENING

Any baby who has a positive NBS for CF should be assessed for symptoms of malabsorption and respiratory problems and referred to a CF Foundation-accredited care center for further diagnostics. Sometimes a baby can have a positive NBS for CF even if the parents had a negative prenatal screen for cystic fibrosis. CF is most common in Caucasians, but it is present in all ethnic groups. In non-Caucasians, there may be CF mutations that have not been identified. Thus, sweat testing is a more sensitive test than DNA analysis in ethnic groups other than Caucasians. Even though CF is inherited, up to 80% of newly diagnosed people do not have a family history of CF.

When informing parents that their infant's CF NBS is positive, indicating the need for further testing. It is important to reassure them that most babies who screen positive do not have CF. A sweat test or genetic test should be scheduled within a week or two after newborn screen results are received. Parents may be understandably quite anxious and may wish to have the sweat test done as soon as possible.

On the day of the sweat test, instruct parents not to use any lotions, creams or moisturizing soaps on the baby's skin. Also, advise them to bring a blanket and hat to keep the baby warm.

WHY SCREEN FOR CF?

Studies have shown that babies who are diagnosed with CF through NBS and treated before symptoms appear are healthier. Their nutritional status can be monitored closely and interventions started as soon as necessary. Evidence from the CF Foundation's Patient Registry (http://www.cff.org) shows that the better the nutritional status is for the child with CF, they will likely grow and develop like a child without CF. In addition, there are indications that the better the nutritional status the more likely the person with CF will have healthier lungs. While there is

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not yet a cure for CF, early symptom management results in better nutritional outcomes, slower progression of lung damage and an improved quality of life. More than 50% of people with CF today will survive through their third decade, and many survive decades longer.

CF newborn screening also has implications for parents as they consider more children. Parents may choose to be genetically tested for a CF mutation even if their child does not have CF. If both parents are CF carriers, then each pregnancy has a 1 in 4 risk of being a child with CF. Parents who either have a child with CF or are carriers of a CF mutation should be referred to a genetic counselor familiar with CF.

RESOURCES FOR PARENTS AND FAMILIES

To learn more about testing for cystic fibrosis and Newborn Screening for CF, visit the CF Web site (www.cff.org) or you can contact the CF Foundation at 800 FIGHT CF (800-344-4823).

There are two fact sheets available from the CF Foundation that address the issues of newborn screening and babies who are identified as carriers:

My Baby's CF Newborn Screen is Positive

My Baby is a CF Carrier: What does that mean for my baby? What does it mean for me?

Resources for healthcare providers:

Guidelines for Diagnosis of Cystic Fibrosis in Newborns through Older Adults: Cystic Fibrosis Foundation Consensus Report available on the CF Foundation's Web site—http://www.cff.org/treatments/CFCareGuidelines/Diagnosis/#CF_Diagnosis.