Newborn Metabolic Screening Program

Cystic Fibrosis Added to Screening Panel on September 2007

INFORMATION ALERT

WHAT A PEDIATRIC PROVIDER NEEDS TO KNOW

Implications For Your Practice

1. Cystic fibrosis (CF) was being added to the state newborn screening (NBS) panel on **September 1, 2007.**

2. NBS for CF is a **screening** test. **DO NOT** base a diagnosis of CF on one or two positive NBS results. Diagnosis of CF needs to be confirmed with additional diagnostic tests and genetic evaluation. The Hawaii Newborn Screening Program can assist you with the coordination and costs of the follow-up to confirm a diagnosis of CF in the newborn.

3. Health care providers **should not** begin medical intervention until confirmatory testing is completed. About one-third of children with positive screening results are confirmed to have CF.

4. Families and infants diagnosed with CF should receive genetic evaluation and counseling, and ongoing care provided by a pediatric pulmonologist and gastroenterologist.

5. NBS **does not** detect all cases of CF in newborns. Up to 5% of CF cases may not be detected so **all symptomatic** children for CF should receive evaluation regardless of their NBS test result.

6. Questions regarding the NBS for CF or any other NBS questions can be directed to the Newborn Metabolic Screening Program Coordinator, Gwen Palmer, R.N., M.Ed., at (808) 733-9069.

ADDITIONAL INFORMATION

Background

Every state in the U.S. has now adopted newborn screening protocols for cystic fibrosis (CF). The Hawaii Newborn Metabolic Screening Program Advisory Committee recommended the addition of CF to the state newborn screening (NBS) panel and CF was added to the NBS panel on September 1, 2007.

Rationale

Cystic fibrosis occurs once in every 3,700 births in the United States and is one of the most common genetic disorders among Caucasians. CF is a severe multi-organ disease, primarily affecting the lungs and pancreas.

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The frequency of CF varies by race and ethnicity (see table below):

Group	CF Cases/Live Births
Caucasian	1 in 2,500 – 3,000
Hispanic	1 in 4,000 – 10,000
African American	1 in 15,000 – 20,000
Asian	1 in 30,000

It is caused by a defect in the cystic fibrosis transmembrane regulator (CFTR) gene, which encodes a protein that regulates salt transport across cell membranes. This defect leads to excessively viscous secretions that cause blocked glands, chronic respiratory obstruction, and infection. Death from pulmonary infections and respiratory failure may occur. Although there are over 1,000 different mutations of the CFTR gene, approximately 70% of the CF cases in the U.S. are caused by ΔF508 mutation; while the remaining 30% of CF cases can result from any of the 1000+ mutations.

The average age of diagnosis in the U.S. is 14 months without newborn screening. With newborn screening, diagnosis can be made as early as two weeks after birth. Children diagnosed earlier through newborn screening tend to have improved nutritional status and cognitive development. Future innovations in clinical management may significantly improve survival and further increase the benefits of newborn screening.

Screening Methodology

Hawaii will follow the testing algorithm used by the Oregon State Public Health Laboratory (OSPHL), Hawaii's contracted NBS laboratory. The algorithm involves initial immunoreactive trypsinogen (IRT) testing of the blood spot specimen routinely collected after birth ≥ 24 hours.

If the initial IRT is positive, a repeat IRT test will be requested. Trypsinogen is the inactive precursor of pancreatic trypsin and is elevated in infants with CF. Newborns with a second positive IRT test will receive confirmatory testing, genetic evaluation, and genetic counseling.

NOTE:
This testing algorithm does not identify carriers, and has a lower proportion of false-positive cases than an algorithm that performs IRT testing with second tier DNA testing.

Questions?

For more detailed information about NBS for Cystic Fibrosis, please go to http://www.newbornscreening.info/Parents/otherdisorders/CF.htm

or

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