



**Department of Health and Social Services**

Karleen Jackson, Commissioner

3601 C Street, Suite 540, PO Box 240249

Anchorage, Alaska 99524-0249 <http://www.epi.Alaska.gov>

**Division of Public Health**

Deborah L. Erickson, Acting Director

Local (907) 269-8000

24 Hour Emergency 1-800-478-0084

**Editors:**

Jay C. Butler, MD

Joe McLaughlin, MD, MPH

Bulletin No. 01 January 5, 2007

## Introduction of Newborn Screening for Cystic Fibrosis in Alaska

### Background

Twenty-one states currently screen for cystic fibrosis, and five additional states are planning to implement screening soon. The Alaska Newborn Metabolic Screening Program will add cystic fibrosis to the newborn screening panel beginning on February 1, 2007. This *Bulletin* describes the rationale for the decision to add cystic fibrosis to the newborn screening panel, the testing procedure involved, and implications for Alaska health care providers.

### Rationale

Cystic fibrosis occurs once in every 3700 births in the United States and is one of the most common autosomal recessive disorders among Caucasians. 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. Approximately 70% of people in the United States with cystic fibrosis have a single mutation ( $\Delta F508$ ), while the remaining 30% have any of over 1000 different gene defects.

Cystic fibrosis is a severe multi-organ disease, primarily affecting the lungs and pancreas. Death before adulthood from pulmonary infections and respiratory failure may occur. Better pulmonary care and nutritional support have resulted in higher quality of life and prolonged survival.

The average age of diagnosis in the United States is 14 months without newborn screening, and as little as 2 weeks in states with newborn screening. Children diagnosed earlier through newborn screening tend to have improved nutritional status and cognitive development. Nevertheless, studies have not yet found conclusive evidence that newborn screening results in increased survival, long-term improvements in pulmonary function, or decreased *Pseudomonas* colonization. Future innovations in clinical management may significantly improve survival and further increase the benefit of newborn screening.

### Screening Methodology

Alaska follows the testing algorithm used by the Oregon Public Health Laboratory, Alaska's designated newborn screening laboratory. The algorithm involves initial immunoreactive trypsinogen (IRT) testing of the screening blood spot specimen routinely collected at birth, followed by repeat IRT testing on a sample collected at age 2 weeks for newborns who are positive by the first test. Trypsinogen is the inactive precursor of pancreatic trypsin and is elevated in infants with cystic fibrosis. Newborns with a second positive IRT test receive a confirmatory sweat chloride test and genetic evaluation.

This testing algorithm does not identify carriers, and has a lower proportion of false-positive cases than an algorithm used in some states that perform IRT testing

followed by DNA analysis. Furthermore, two-step IRT testing is less costly than follow-up testing with DNA analysis.

### Implications for Health Care Providers

#### *Relevant Numerical Estimates*

- Approximately 10,000 live births occur in Alaska each year.
- 1 in 3000 infants born in Alaska have cystic fibrosis.
- 8% of infants with a single positive test and 39% of those with two positive tests will have cystic fibrosis confirmed.
- Over the next 10 years in Alaska, the initial IRT screening test will be positive in approximately 400 newborns, of whom 82 will have a positive second test. Of these 82 infants, 32 will have cystic fibrosis confirmed.
- During this same period, two children with cystic fibrosis will not be detected by the screening test program.

#### *Access to Care*

In Alaska, specialist care for children with cystic fibrosis is available through the Pediatric Subspecialty clinics at The Children's Hospital at Providence in Anchorage.

### Recommendations

1. Health care providers should be aware that newborn screening for cystic fibrosis will begin on 2/1/2007.
2. Health care providers should be aware that the fee per newborn for the two screen panel will increase to \$75, and this will be covered by Medicaid. Confirmation by sweat chloride test is not included in this fee.
3. Health care providers should not base a diagnosis of cystic fibrosis on one or two positive IRT screening tests. Diagnosis of cystic fibrosis can be confirmed only with a sweat chloride test.
4. Health care providers should remember that newborn screening will miss up to 5% of cystic fibrosis cases, and therefore should evaluate symptomatic children for cystic fibrosis despite a negative screening test.
5. Health care providers should not begin medical intervention or counseling until confirmatory testing is completed, because only about one-third of children with positive screening results have cystic fibrosis.
6. Infants diagnosed with cystic fibrosis should receive genetic evaluation and counseling, and ongoing care provided by a pediatric pulmonologist.
7. Questions regarding the newborn screening program should be directed to the Newborn Screening Program Director, Thalia Wood, at 907-269-3499.

Additional information for providers and parents is available from the Pediatric Subspecialty Clinics (907-261-4824).