



**NEWBORN SCREENING
FOR CYSTIC FIBROSIS:
REPORT AND
RECOMMENDATIONS**

**OREGON CYSTIC FIBROSIS NEWBORN SCREENING TASK FORCE
MAY 2005**



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EXECUTIVE SUMMARY

Cystic Fibrosis (CF) is a lethal genetic disorder that occurs in one of every 3,700 births in the U.S. The diagnosis and treatment of CF are often delayed for months or years because CF symptoms are easily mistaken for other diseases. During this period, the disease can progress unchecked in infants and young children. In contrast, when CF is detected at birth by a laboratory test there is no delay in diagnosis and the progression of the disease can be prevented or deferred.

Fortunately, babies can now be tested for CF using blood samples already collected for other newborn screening purposes. In fact, nine states currently test all newborns for CF and four other states have pilot programs or limited screening. Recently, several national organizations and the federal government have recommended that all newborns be screened for CF.

The Oregon CF Newborn Screening Task Force was convened in September 2004 to review all available information and recommend to the Department of Human Services whether CF screening should be adopted for Oregon newborns. The Department's Public Health Laboratory currently tests all newborn infants in five states for 26 different disorders. After considering benefits, risks, costs, alternative program models, national trends and recommendations, and policy issues, the Task Force recommended the following:

- Implementation of a comprehensive newborn screening program for CF, integrated into the existing program for metabolic, endocrine, and hemoglobin disorders
- Immediate follow-up and tracking of infants with abnormal screening results, and assurance of adequate diagnostic evaluation
- Inclusion of CF Care Centers as an integral part of the CF newborn screening system, as a focal point for confirmatory testing, medical consultation, parent and practitioner education, and genetic counseling
- Genetic counseling offered to the families of all infants with positive screening results
- Expert medical consultation available to infants' physicians, as well as for program and policy decisions
- Parent and practitioner education
- Quality assurance of all program elements
- Self-funding of CF screening activities from newborn screening fees





INTRODUCTION AND BACKGROUND

The Oregon Cystic Fibrosis Newborn Screening Task Force was convened in response to a growing interest in newborn screening for Cystic Fibrosis (CF) among health professionals and the public. An expanding body of research has documented the effectiveness of newborn screening for this condition. The Task Force was co-sponsored by the Office of Family Health and the Office of Oregon State Public Health Laboratories (OSPHL) within the Oregon Department of Human Services (DHS).

The Task Force was charged with recommending whether newborn screening for CF should be implemented for Oregon infants, and, if so, defining the critical components of the system for screening, follow-up, referral, and counseling. Task Force members were asked to review and consider a wide range of information, and then make recommendations to DHS. Task Force members represented local and state public health programs, physicians, hospitals, insurers, and consumers (Appendix A). The Task Force held six meetings between September 2004 and April 2005.

This report reviews the major issues relating to newborn screening for CF and presents the recommendations of the Task Force.

NORTHWEST REGIONAL NEWBORN SCREENING PROGRAM

The Northwest Regional Newborn Screening Program (NRNSP) is a collaborative effort between the state public health agencies of Oregon, Alaska, Idaho, Nevada, and Hawaii, the Oregon Health and Science University (OHSU), parents, and health care providers for newborn babies throughout the region. The purpose of the NRNSP is to identify, and refer to medical care, newborn babies needing immediate treatment to prevent profound developmental problems, serious illness, or death.

Each infant is screened for 26 metabolic, endocrine, and hemoglobin disorders using a few drops of blood collected from the heel. Infants with these disorders appear normal at birth, but may become affected during the first few weeks or months of life unless they are treated promptly. Most of these disorders arise from an underlying genetic cause.

The OSPHL performs laboratory screening and administers the regional program. In addition to laboratory testing, the NRNSP provides medical consultation and education for practitioners, follow-up and tracking of infants with positive screening results, initial laboratory confirmation of diagnoses, and educational materials for parents. Each year, the OSPHL performs more than three million tests on 240,000 specimens collected from 130,000 infants, including 46,000 born in Oregon.

The Newborn Screening Program is supported entirely by fees that are authorized by state law (ORS 431.310) for Oregon infants, not to exceed \$30 per sample, with two samples mandated for each baby. Fees are set in Oregon Administrative Rule (OAR 333-024-0240); the fee is currently \$27 per sample. Alaska, Idaho, Nevada, and Hawaii each set their own newborn screening fee, and contract with the OSPHL to provide laboratory screening, initial follow-up, confirmatory testing, and medical consultation.



CRITERIA AND ASSUMPTIONS

Task Force members felt that the primary goal of CF newborn screening should be to identify babies with CF early and connect them with comprehensive care to prevent or delay serious health and developmental problems. The program should also strive to minimize any potential harm from the screening process itself.

In the past, various organizations, including the World Health Organization and the American Academy of Pediatrics, have developed criteria to guide decisions about which disorders should be screened for in newborns. However, technologies have evolved significantly since these criteria were developed, prompting many states to develop their own guidelines.

In general, all sets of newborn screening criteria are in agreement on the following:

- Newborns should be screened only if the screening provides clear benefit to the newborn
- Appropriate testing technology must be available
- Appropriate diagnostic follow-up and treatment must be available
- The disease incidence must be high enough to justify screening, and
- The benefits of screening must justify the cost.

Early in the deliberative process, Task Force members held mixed opinions about the value of CF newborn screening. However, they all agreed that if a program was implemented it must be comprehensive, assuring high quality community-based health care, accurate and reliable laboratory screening and diagnostic tests, ongoing guidance by expert medical consultants, and educational outreach for parents and practitioners.



CYSTIC FIBROSIS OVERVIEW

CLINICAL FEATURES

Cystic Fibrosis is a lethal genetic condition that usually manifests in infancy but is often difficult to recognize. The most common early symptoms are recurrent cough, wheezing, abdominal pain, loose stools, and failure to thrive. Pancreatic insufficiency leads to malnutrition and severe growth problems. Respiratory infections increase in frequency and severity with age, in association with progressive decrease in lung function. Respiratory failure is the cause of death in more than 90% of persons with CF.

Fortunately, with better understanding of the disease and improved treatment, life expectancy for CF patients has increased dramatically. In recent decades, the median predicted age of survival in the U.S. has risen from 14 years to 33 years. Until the 1980's, most deaths occurred during childhood or adolescence, but today most persons with CF survive into adulthood.¹

Currently, a small portion of infants with CF are diagnosed during pregnancy by prenatal testing. Another 15-20% become ill during the first days of life with a complete intestinal obstruction that leads to prompt diagnosis of CF. The remaining large majority of infants with CF often go unrecognized for some time. Without newborn screening for CF, the median age at diagnosis is 14.5 months; with newborn screening, the median age at diagnosis is two weeks.


In the absence of newborn screening for CF, diagnosis is often delayed because most CF symptoms are not specific to CF and many affected children are misdiagnosed as having food allergies, celiac disease, asthma, or bronchitis before their CF is finally recognized. This delay greatly increases the health care costs associated with CF. Also, misdiagnoses often result in a “diagnostic odyssey” involving multiple office visits, diagnostic tests, and hospitalizations which can take an emotional and economic toll on the family. In addition, the child's health may be compromised by long delays in diagnosis. Data from the national Cystic Fibrosis Foundation registry show that babies diagnosed based on clinical symptoms, versus newborn screening tests, are significantly more likely to suffer the effects of moderate to severe malnutrition, as well as crucial vitamin deficiencies. There is compelling evidence that these vitamin deficits can affect future cognitive function.² In addition, acquisition of frequent, severe respiratory infections in undiagnosed children correlates with more rapid progression of lung disease.

INHERITANCE

Cystic Fibrosis is caused by an inherited defect in the normally occurring cystic fibrosis transmembrane regulator (CFTR) gene, which codes for a protein that regulates transport of salts across cellular membranes. This defect leads to excessively thick, viscous secretions that cause blocked glands, chronic respiratory obstruction, and infection.

Cystic Fibrosis shows autosomal recessive inheritance. For a child to have CF, both parents must carry a CFTR gene with a CF mutation, and the child must inherit a CF mutation from each parent. However, the molecular diagnosis of CF is more complex than might be expected because there are more than 1,000 different mutations in the CFTR gene. The most commonly recognized mutations cause serious disease, while some of the rare mutations cause less severe health problems. In the U.S., at least 70% of CF cases are the result of one mutation ($\Delta F508$), and another 25% of cases can result from any of approximately 40 other mutations.³

On the average, one in 32 people in the U.S. carries a single CF mutation. These “carriers” do not have CF and



will not develop it. One in 3,700 newborns has two CF mutations, and therefore will develop CF. The frequency of disease varies by race/ethnicity, as shown below.

<u>Group</u>	<u>CF Cases/Live Births</u>
Caucasians	1/2,500-3,000
Hispanics	1/4,000-10,000
African Americans	1/15,000-20,000
Asians	1/30,000

Cystic Fibrosis is the most common lethal genetic disease in Caucasians, but CF can occur in any racial or ethnic group. In 2003, 95.3% of U.S. CF cases occurred in Caucasians, 6.5% in Hispanics, and 3.8% in African Americans.⁴ Some CF mutations are specific to certain racial and ethnic populations. Therefore, to improve detection of CF in all infants, the genetic marker panel used for newborn screening in a given state should be “panethnic,” reflecting the demographics of that state.

DIAGNOSIS AND TREATMENT

The diagnosis of CF is made on the basis of the presence of one or more of the following:

- One or more clinical findings suggestive of CF
- A history of CF in a sibling
- A positive newborn screening test

PLUS one or more of the following:

- Positive sweat chloride test performed by experienced personnel using standardized methodology
- Characteristic ion transport defects in nasal tissue
- Two CF-causing mutations

Early treatment of CF is crucially important in improving the quality and length of life. The intent is to minimize the long-term consequences of lung involvement and malnutrition. Treatment varies depending on the stage of the disease and the organ systems involved. Once the diagnosis is confirmed, treatment is focused on preventive and maintenance care, with acute care when needed. Pulmonary-specific treatment includes chest physiotherapy at least twice a day to keep the airway clear, and inhaled medications to improve lung function and combat infection. Pancreatic insufficiency results in malabsorption and malnutrition. Treatments for this include oral pancreatic enzymes with every meal, oral or tube feedings with high-caloric supplements, and repletion of essential vitamins. Many promising new therapies for CF are under investigation, including those that directly target the underlying mechanism of CF by improving salt transport across membranes.



GENETIC COUNSELING

Genetic counseling involves assessing the chance a condition will occur; educating about inheritance, testing, management, prevention, and research; connecting with resources; and assisting with adaptation to the condition. Genetic counselors help people understand the medical, psychological, familial, social, ethical, and reproductive implications of a condition.

Genetic counseling should be offered to the parents of all infants identified through newborn screening as having CF or being CF carriers, totaling approximately 150 Oregon infants per year. This number includes an estimated 130 infants who would be identified as carriers (one CF mutation) as well as approximately 15-20 affected infants (two CF mutations).

The parents of infants diagnosed with CF should be offered genetic counseling because they have a 25% chance of having a baby with CF in each future pregnancy, and should be informed of this risk and its implications. The parents of carriers should also be offered genetic counseling because at least one parent of each of these infants must also be a carrier. In fact, because of the high carrier frequency in the general population, for each CF carrier infant born, there is a three percent chance that both parents are carriers. These couples also have a 25% chance of having a baby with CF in each future pregnancy.

Available data suggest that without adequate counseling, many parents of babies who are carriers do not fully understand that their children do not have CF, and will not develop CF in the future. This misconception can lead to increased worry, anxiety, and overprotection.

CYSTIC FIBROSIS SERVICES IN OREGON

Oregon has two CF care centers, both located in Portland, that offer multidisciplinary care consistent with clinical practice guidelines delineated by the national Cystic Fibrosis Foundation. The center at OHSU is fully accredited by the Foundation, and the center at Kaiser Permanente is accredited as an affiliate center. The centers provide comprehensive outpatient management of CF including respiratory therapy, nutritional counseling, laboratory services (including sweat chloride testing), social work evaluations, and hospital inpatient care. Twenty-four hour physician and staff coverage is available. CF center professionals also provide consultation to the patient's primary care and other service providers. CF mutation analysis is available from the OHSU DNA Diagnostic Laboratory.

ROLE OF CYSTIC FIBROSIS CENTERS IN NEWBORN SCREENING

The Task Force felt strongly that Oregon's CF care centers should be an integral part of a CF newborn screening system. The centers would serve as a focal point for sweat chloride testing, medical consultation, parent and practitioner education, and genetic counseling. Infants with positive CF screening tests would need to be evaluated in one of the Portland CF care centers. Ideally, expert specialty care would be available to all Oregon CF patients within their own community, but this is not currently the case. In the future, DHS should work with health care providers throughout the state to develop high quality follow-up services for infants with positive newborn screening results and to increase the level of care that can be provided locally.





NEWBORN CYSTIC FIBROSIS SCREENING

SCREENING METHODS

Cystic Fibrosis newborn screening can be performed using the same dried blood spot sample collected for other newborn screening tests. Samples are first tested for immunoreactive trypsinogen (IRT), an indication of pancreatic obstruction that is present at birth in most newborns who have CF. Programs in five states then repeat the IRT test for infants whose initial IRT value is elevated. Those with repeatedly elevated IRT values are referred for diagnostic sweat chloride testing. Testing for specific CF mutations in the infant's DNA is not performed as part of the screening process.

Eight states employ an IRT/DNA testing protocol. All samples with elevated IRT levels are tested for one or more specific mutations in the CFTR gene. Some states test for only the most common mutation ($\Delta F508$), while others test for a panel of up to 40 different CFTR mutations. Regardless of the screening protocol – IRT/IRT or IRT/DNA – programs differ in their screening cutoff values, test algorithms, and diagnostic referral practices.

WEIGHING BENEFITS AND RISKS

The Task Force agreed with the following conclusions by the CDC/Cystic Fibrosis Foundation work group:

“On the basis of a preponderance of evidence, the health benefits to children with CF outweigh the risk of harm and justify screening for CF.... As a result, CDC believes that including screening for CF in state newborn screening programs is justified. The evidence of clinical benefits from newborn screening for CF is based on an extensive body of research, including two randomized clinical trials and multiple prospective cohort studies.... The net balance of benefits and risks is contingent on how newborn screening for CF is implemented. Consequently, newborn screening programs for CF, if initiated, should be of high quality and carefully monitored to ensure consistent quality and effectiveness.”¹

EVIDENCE OF BENEFIT

The Task Force found a substantial body of evidence that newborn screening for CF provides significant benefits. In 1997, and again in 2003, the Centers for Disease Control and Prevention (CDC) and the Cystic Fibrosis Foundation cosponsored workshops on newborn CF screening as a state public health policy issue. These workshops found that peer-reviewed scientific evidence supports the utility of newborn screening tests in identifying newborns with CF. Some of the major benefits include:^{1,5}


- Improved growth (by preventing or minimizing malnutrition)
- Improved cognitive development
- Reduced hospitalization
- Improved survival
- More rapid diagnosis
- Genetic counseling for family planning
- Reduction of psychosocial stress



EVIDENCE OF RISKS

In addition to considering evidence of benefit, the Task Force evaluated evidence for risk of harm from CF newborn screening. Potential harms exist for CF patients and their families, children who have false-positive or false-negative newborn screens, and for the health care system as a whole.¹ The following risks were considered by the Task Force.

- Infants with CF who are identified through newborn screening could acquire serious lung infections earlier than they would have otherwise, through exposure to other patients during follow-up of a positive newborn screening test, or while receiving preventive treatments. Procedures that isolate screen-positive and affected infants from older CF patients can minimize this risk.
- Parent-child relationships could be altered when a parent learns, via a newborn screening result, that an apparently healthy child may have a serious illness. However, the available research data do not suggest that early identification of CF negatively affects parent-child relationships.
- Although detecting CF carriers is not the primary purpose of CF newborn screening, the screening process will detect approximately 10% of carriers. Theoretically, families of infants identified as carriers might feel uncomfortable with this information, fear discrimination, or feel stigmatized. However, research has actually shown the opposite that most families consider carrier identification to be a useful by-product of newborn screening because it provides helpful information for future decisions about health care and reproduction.
- As a part of newborn screening for CF, some infants who do not actually have CF will have a positive screening test result, and will undergo a diagnostic evaluation. Many of these “false positive” babies will be carriers for CF, while others will not. The parents of a baby with a false-positive test may experience distress and anxiety while waiting for the results of a confirmatory sweat chloride test. Usually, these feelings resolve once the sweat chloride test is complete and the child is found not to have CF or to be a carrier.
- The genetics of CF are easily misunderstood and the implications are complex for the parents of infants with CF as well as the parents of carriers. In both situations, future children may be at risk of having CF. In addition, many parents of children who are carriers of a single CF mutation may not understand that a single copy of a CF gene will not cause disease. Consequently, it is essential that a CF newborn screening program offers genetic counseling to parents of all infants who have CF or are identified as carriers through screening. Educational materials must also be available to parents, both before and after screening.
- As with any screening test, false-negative test results (negative newborn screening results in infants who have CF) are possible. A well-designed and implemented Oregon CF newborn screening program should miss an average of less than one baby per year. Ironically, the implementation of a CF newborn screening program might decrease the index of clinical suspicion among health care providers who assume that all CF cases will be detected at birth. This could delay the diagnosis of the small number of infants with CF who are not detected by newborn screening. Practitioner education efforts should address this issue.

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- Newborn screening for CF poses a risk to the health care system as a whole. The increased need for sweat testing and genetic counseling may exceed the capacity of existing health care resources, including CF care centers. Therefore, implementing a newborn screening system carries associated opportunity costs if health care resources are diverted away from other potential uses.

NATIONAL TRENDS AND RECOMMENDATIONS

The findings and recommendations of the 2003 CDC workshop were reported in *Morbidity and Mortality Weekly Report (MMWR)* in October 2004. The report recommended that “The magnitude of the health benefits from screening for CF is sufficient that states should consider including routine newborn screening for CF in conjunction with systems to ensure access to high-quality care.” Additional recommendations included:

- Consider state priorities and national guidelines regarding CF screening, diagnosis, and treatment
- Collect follow-up data for use in monitoring and improving CF newborn screening
- Implement rigorous infection control policies to minimize the risk of person-to-person transmission of pulmonary infection
- Ensure effective and timely communication between the newborn screening laboratory, parents, and primary care providers to facilitate prompt referral to diagnosis centers. CF centers should be skilled in diagnostic testing and should provide effective education and genetic counseling

The Task Force also considered other national trends. The March of Dimes recently added CF newborn screening to its core panel of standard newborn screening tests recommended for implementation in all states.⁶ Nine states now screen all newborns for CF, and four other states have pilot programs or limited screening.

Most recently, the federal Health Resources and Services Administration commissioned the American College of Medical Genetics to evaluate the overall effectiveness of newborn screening, and to make recommendations, including a uniform panel of conditions for implementation across all states. Based on the best scientific evidence and rigorous analysis of the evidence, CF was one of 29 disorders recommended for inclusion in this uniform panel.⁷

PROGRAM ELEMENTS AND SERVICES

The Task Force reviewed the program elements and services included in the CF newborn screening programs of other states. Some Task Force members were already acquainted with other states’ programs. Task Force members were particularly impressed with both the implementation and evaluation strategies of the Massachusetts and Wisconsin programs. Detailed information about the Wisconsin program was reviewed by the Task Force.⁷ In addition, Dr. Richard Parad, Co-Director of the Massachusetts CF Newborn Screening Program, traveled to Oregon to describe the Massachusetts program for the Task Force. Elements of both programs were incorporated into the recommendations that follow.





RECOMMENDATIONS

The Task Force recommends implementing a comprehensive newborn screening program for CF in Oregon infants, provided that all necessary program elements are available and in place at the time the screening program is implemented. These elements include:


- Cystic Fibrosis screening of newborns integrated into the existing newborn screening program for metabolic, endocrine, and hemoglobin disorders
- Laboratory screening of dried blood spots for immunoreactive trypsinogen (IRT) (Appendix B)
- CFTR mutation analysis of infants with positive IRT screening results, using a panethnic panel of at least 40 different mutations
- Immediate follow-up and tracking of infants with abnormal screening results.
- Assurance of adequate diagnostic evaluation
- Confirmatory testing by a sweat chloride method approved by the Clinical and Laboratory Standards Institute⁹
- Referral of infants with a positive sweat chloride test to a CF Foundation-approved center
- Inclusion of CF Centers as an integral part of the CF newborn screening system, as a focal point for sweat chloride testing, medical consultation, parent and practitioner education, and genetic counseling
- Genetic counseling offered to the families of all infants receiving sweat chloride testing
- Expert medical consultation available to infants' physicians, as well as for program and policy decisions
- Parent and practitioner education
- Quality Assurance of all program elements

CF NBS Task Force Initially recommended screening be done using the first IRT, then DNA protocol. After further considerations, the Oregon CF NBS strategy was modified to first IRT, then follow up testing by IRT. Screening exclusively by IRT eliminates the disadvantage of inadvertently identifying children who are CF carriers. Infants with elevated IRT levels on newborn screens will be referred to a CF Care Center for diagnostic sweat chloride testing.

COST AND REVENUE

The costs of the CF NBS screening program would be paid from newborn screening fees adopted in Oregon Administrative Rules, and cannot exceed the statutory cap (ORS 431.310). The cost of CF screening, taking into account all of the recommended components, was estimated at \$7.87 per infant (Appendix C). Program funds would pay for sweat chloride testing and genetic counseling for those infants who are not covered by public or private health insurance.

Included in the \$7.87 estimate are the costs necessary to provide genetic counseling for the families of infants identified with hemoglobin and metabolic disorders, comparable to that recommended for CF. Although these



are not direct costs of CF screening, the Task Force felt that these services should be implemented along with CF screening. Genetic counseling is not currently provided for these families as part of the newborn screening program.

TIMELINE FOR IMPLEMENTATION

The Task Force recommends that CF screening be implemented as soon as it becomes possible to begin a comprehensive program. The newborn screening fee must be raised to accommodate CF screening. Because the estimated cost of CF screening (\$7.87 per infant) exceeds the \$6.00 currently available under the statutory fee cap, this may require changes in ORS 431.310 during the 2007 legislative assembly. The OSPHL should look for savings and efficiencies in other areas that could be used to pay part of the cost of CF screening and allow earlier start-up, provided these savings do not result in service reductions in existing programs, and a permanent, sustainable source of revenue can be identified. Once the necessary revenue becomes available, CF screening should begin within 9 months.



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APPENDIX A

Oregon Cystic Fibrosis Newborn Screening Task Force Members

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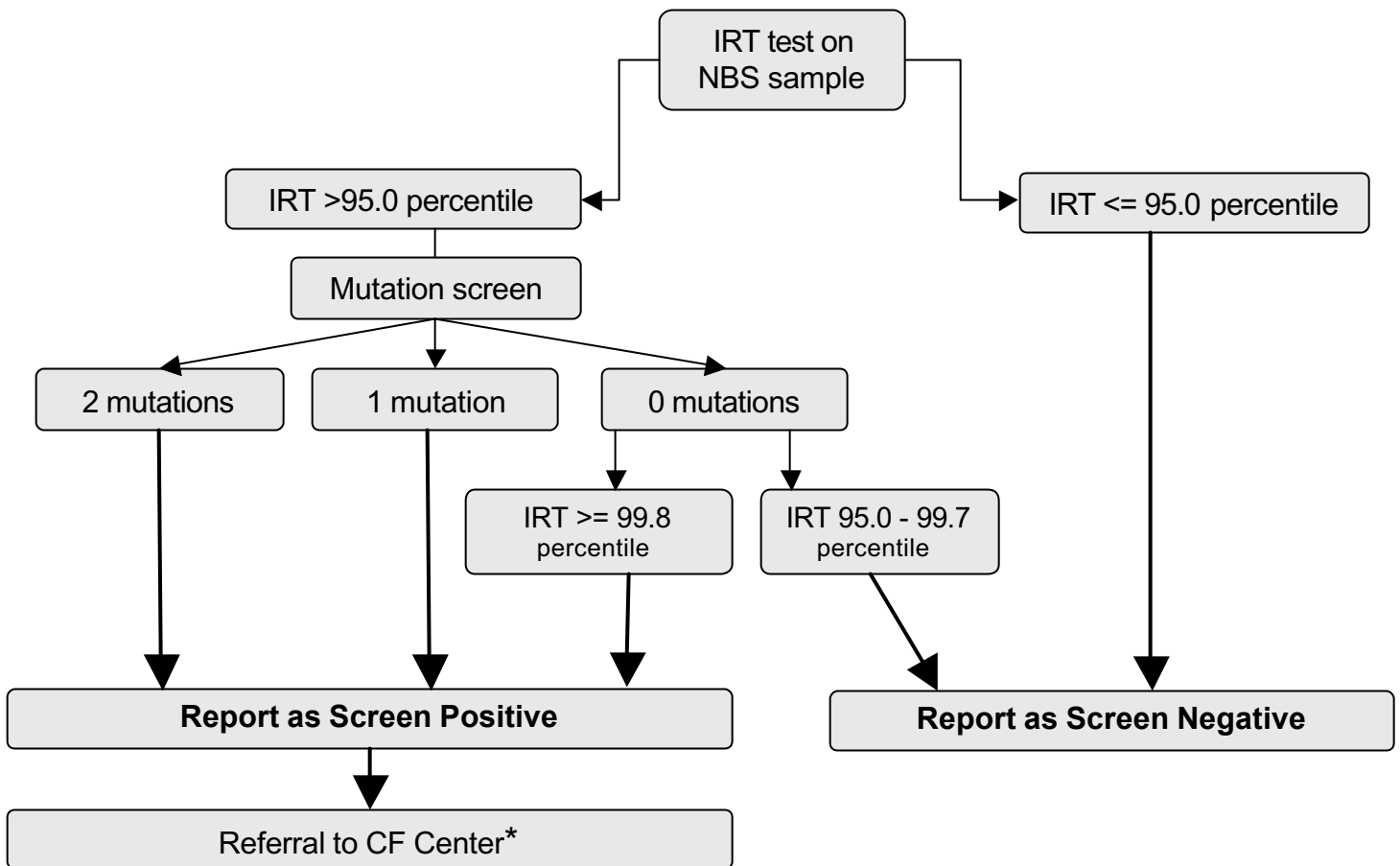
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APPENDIX B

Newborn Laboratory Screening Protocol For Cystic Fibrosis



*For sweat chloride testing, clinical evaluation, treatment, genetic counseling

IRT = Immunoreactive Trypsinogen

NBS = Newborn Screening

Adapted from Dr. Richard Parad, New England Regional Cystic Fibrosis Newborn Screening Program.

APPENDIX C

Cost Analysis for Newborn Cistic Fibrosis Testing for Oregon

	IRT + 40 mutations
Lab Testing at OSPHL	
Reagents (A)	
IRT (46,000)	\$49,000
DNA (2,000)	\$100,000
Equipment (A)	
IRT	\$10,000
DNA	\$4,000
Labor	
Microbiologist 2 (B)	\$62,000
Initial Follow-up and Tracking (C)	
Medical Consultation (D)	
program support (E)	\$13,248
physician (F)	\$36,018
Confirmation Test	
sweat chloride (G)	\$6,000
Practitioner & Parent Education (H)	\$29,203
Genetic Counseling (I)	
Cystic Fibrosis (J)	\$33,750
Hemoglobinopathies (K)	\$13,275
Metabolic (L)	\$5,625
TOTAL	\$362,119
Cost per specimen	\$7.87

Notes:

Assumption: 46,000 Oregon specimens per year (test first NBS specimens only)

- (A) Based on Wisconsin experience.
- (B) 0.5 FTE for IRT and additional 0.5 FTE for DNA testing.
- (C) Will be absorbed with existing OSPHL staff.
- (D) Based on current contract for medical consultation for endocrine disorders.
- (E) Program includes bi-monthly meetings & annual regional meeting.
- (F) Assumes 0.5 hr per presumptive positive plus 1.0 hr per confirmed positive.
- (G) Cost for uninsured only: 150 tests/year x \$200/test x 20% uninsured.
- (H) Based on current contract with OHSU CDRC for education and follow-up.
- (I) Alternately, 1.0 FTE genetic counselor would cost approximately \$62,572 per year.
- (J) Estimate all sweat chloride test families (150) at \$225/visit.
- (K) Estimate 10% of identified carriers (50) plus all confirmed cases (9) at \$225 per visit.
- (L) Estimate 25 metabolic cases at \$225 per visit.





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