

Improving the Care of Infants Identified through Cystic Fibrosis Newborn Screening

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On behalf of the Cystic Fibrosis Foundation, we are pleased to share the enclosed clinical practice guidelines for the care of infants identified through cystic fibrosis (CF) newborn screening (NBS). The widespread implementation of NBS for CF throughout the USA presents an incredible opportunity to improve health outcomes for this patient population. The evidence documenting the magnitude of the opportunity is growing.^{1,2} We are entering a new era in which the devastating symptomatic presentations of CF such as failure to thrive will become a thing of the past. The CF Foundation Patient Registry data shows that a growing proportion of newly diagnosed infants are identified by NBS (Figure). If we are to fully capitalize on this opportunity, we must develop a rigorous approach to these newly diagnosed infants including frequent follow-up, institution of appropriate preventive measures to maintain health and early intervention when problems arise. The clinical practice guidelines published herein are aimed at promoting a standardized approach to the care provided for these infants.

The development of evidence-based clinical practice guidelines is a core component of the CF Foundation's quality improvement (QI) initiative. Although we believe the CF Foundation's system of care is a model for the care of individuals with a complex, chronic disease, variability in practice patterns and outcomes among CF care centers uncovered in the CF Foundation Patient Registry data indicated that we could do even better. We developed and implemented a strategic plan to identify and enable "best practices" across our care center network. Many of the more than 100 accredited care centers and more than 50 affiliates across the United States have incorporated QI into their daily work. Registry analyses since the launch of the QI initiative in 2002 suggest that the CF Foundation's investment in this area is having a substantive impact on key pulmonary and nutritional outcomes (manuscript in preparation). The current focus on the care of infants identified by NBS is a natural extension of this work.

The first article in this supplement contains the recommendations for management of infants identified by NBS who have a clear diagnosis of CF. A care grid with recommended frequency of follow-up and what should be done and when is included in the article. The important partnerships between the family and the health care team and between

the primary pediatrician and the CF care team are also highlighted in this document.

The second article describes the method used to develop the guidelines recommendations including the search and selection criteria, the data collection process, and production of a narrative report of the evidence and evidence tables to facilitate the deliberations of the guidelines committee. The CF Foundation has moved from a consensus-driven approach to an evidence-based approach for most of our practice guidelines work. The clinical expertise of the guidelines committee is important in putting the evidence into the proper context and in developing recommendations on important clinical questions for which there is little or no evidence.

The third article addresses infants who are identified by NBS as possibly having CF but do not fulfill the standard diagnostic criteria.³ The uncertainty surrounding the diagnosis, prognosis, and optimal medical care of this important but relatively small number of infants has created a great deal of angst among families and clinicians. As opposed to older individuals with cystic fibrosis transmembrane conductance regulator-related disorders who present for evaluation with symptoms but do not meet diagnostic criteria for CF, these infants identified by NBS are typically symptom-free. This article defines this "CFTR-related metabolic syndrome" group and provides recommendations on their follow-up and care. Given the paucity of evidence in this area, we used a Delphi approach to arrive at consensus recommendations from a panel of experts. A standardized approach to the care of these infants, coupled with collection of genotype-phenotype data in our patient registry will hopefully advance knowledge on this important patient population, ultimately allowing a more rapid diagnosis in some patients and reassurance for others.

We hope that these articles are helpful to CF care teams, general pediatricians, and families. As with all practice guidelines, they provide a framework that must be customized to the care setting. Individual decisions on specific interventions must be made on the basis of the specific circumstances of the individual patient and family. We are thankful to those who provided input on earlier draft versions and eager to hear from you about your experience in implementing the guidelines, which will be incorporated into updates as new data and treatments become available. ■

CF	Cystic fibrosis
NBS	Newborn screening
QI	Quality improvement

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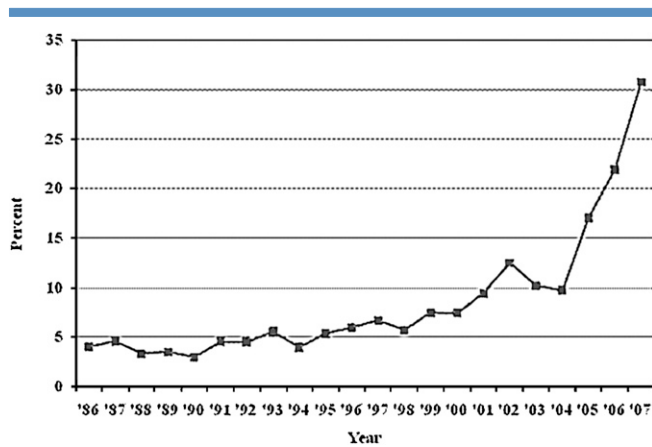


Figure. Proportion of new diagnoses by newborn screening reported to the CF Foundation Patient Registry.

Author Disclosures

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