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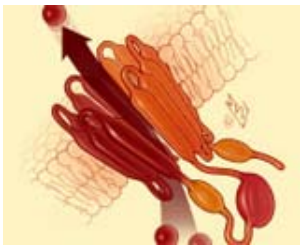

May 2008: VOLUME 1, NUMBER 1

Issues Related to Newborn Screening

Welcome...
The Johns Hopkins University School of Medicine is pleased to welcome you to this inaugural issue of *eCysticFibrosis Review*™. Over the course of this series, we will be reporting on issues critical to providing the most effective diagnoses and the safest evidence-based care management for patients with cystic fibrosis.

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In this Issue...

Cystic fibrosis (CF) newborn screening (NBS) has become an important topic because extraordinary advances in diagnosis, treatment, and prognosis are being achieved nationwide, with tests for immunoreactive trypsinogen (IRT) and DNA implemented at an unprecedented rate to screen infants for this autosomal recessive disorder. Currently, 40 states are routinely using either IRT/IRT (N= 9) or IRT/DNA (N=31) — an increase from 1 and 4, respectively, since the beginning of 2005. However, this is both "good news and bad news": generally good for CF patients and their parents, but potentially bad to have a complex system of care implemented at such a fast pace in regions that may not be optimally prepared, lack adequate understanding of NBS principles/practices, and/or proceed so quickly that errors limit the effectiveness needed to assure more good than harm.

In this issue, we report on important new information on the 6 interdependent components of NBS: a) education of professionals and parents; b) screening – specimen collection, submission, and testing; c) follow-up of abnormal and unsatisfactory test results; d) confirmatory testing and diagnosis; e) medical management and periodic outcome evaluation; and f) system quality assurance.

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THIS ISSUE

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Guest Faculty Disclosures

Philip M. Farrell, MD, PhD has disclosed no relationship with commercial supporters.

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The author has indicated that there will be no reference to unlabeled or unapproved uses of drugs or products in the presentation.

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LEARNING OBJECTIVES

At the conclusion of this activity, participants should be able to:

- Discuss with colleagues the six components of cystic fibrosis (CF) newborn screening (NBS) program
- Describe to colleagues the NBS-linked method and maximum acceptable age of CF diagnosis
- Explain to colleagues the medical and economic benefits of diagnosing CF through newborn screening

COMMENTARY

CF is an autosomal recessive disorder attributable to mutations in the CFTR gene that lead to impaired chloride transport and secondary abnormalities in the sweat glands (excessive salt loss), pancreas (defective digestion leading to malabsorption/ malnutrition), and the respiratory system (recurrent bronchopulmonary infections and obstructive lung disease). Because the diagnosis of CF following signs and symptoms is typically delayed for months or years, newborn screening is needed to ensure a healthy start. However, to maximize sensitivity, NBS tests in general are designed to minimize false negative results ("missed patients"), leading to hundreds more false positive tests that need to be explained to parents and primary care physicians. On the other hand, some NBS tests such as the IRT/DNA strategy add special value: because they employ CFTR multimutation analyses in the DNA tier, about three-quarters of CF infants are recognized as having 2 CF-causing mutations and can be readily diagnosed — indeed, genetically diagnosed within the first week of postnatal life — directly from the dried blood spot. The remaining NBS positives with 1 mutation detected following a high IRT level have <5% probability of CF, but need a diagnostic sweat test for resolution (i.e. determination if they have CF or if they are CF heterozygote carriers). Irrespective of the CF NBS strategy employed, all parents deserve expedited resolution of the NBS outcome and expert, effective counseling. Thus, CF centers have responsibilities for both the false positive families and those actually diagnosed through a positive sweat test. Excellent management of both categories will help ensure more good than harm.[1]

CF NBS programs are "only as strong as the weakest link in the chain." In October 2004, the CDC encouraged the States to proceed with screening programs, but included cautious recommendations such as: "Newborn screening systems should ensure parental and provider education" with comprehensive planning processes.[2] Indeed, the key question shifted from "should we screen" to "**how** we should we screen".[3] During the past 2 years, approximately 100 articles have been published on "how". The 6 NBS aspects listed above were originally published in a report prepared by the American College of Medical Genetics with the Maternal and Child Health Bureau.[4] Although there is an imbalance in the amount of new and important data with regard to the 6 components, it was possible to select 6 excellent, peer-reviewed articles that cover all these elements.

The articles selected encompass the entire NBS system, including the spectrum of processes/procedures and identification of all the populations we need to be concerned about while progressing from nationwide implementation to quality improvement in CF NBS. The article by Tluczek et al clarifies educational challenges and psychological issues by examining parental perceptions about their NBS follow-up experiences and genetic counseling. This study is remarkable for changing the usual focus from what clinicians regard as important (e.g. outcomes) to a parent-centered perspective (e.g. desiring factual information provided clearly and with emotional support). The CF NBS tests *per se* are in a state of evolution — both the IRT tier (due to cutoff value issues) and especially the CFTR panel of the DNA tier (due to uncertainties about what mutations to include reflecting the region's patients). Scotet et al applied an epidemiologic research strategy to evaluate children in Brittany, France with the R117H mutation and proposed "withdrawal of the R117H variant from the panels of CFTR mutations used in cystic fibrosis newborn screening". Other regions are considering expansion of the CFTR panel. In CF follow-up programs, a longstanding issue has related to the optimal age for evaluation of NBS-positive infants with a sweat test. Although the youngest age feasible might be 2 weeks, the article by Sims et al shows unequivocally that 2 months is the maximum acceptable age of diagnosis. The issue of how to proceed with the confirmatory diagnostic test was addressed by Doull et al in Wales, who suggest that the time from notifying parents about NBS positivity to performing a sweat test should be no more than 24 hours; a notification that in practice was accompanied by a high level of parental satisfaction. The economic benefits of early diagnosis through NBS were demonstrated by Sims et al, adding further evidence to the compelling nutritional arguments in favor of screening newborns for CF. Finally, the Farrell and Kuruvilla article addresses quality assurance in the risk communication (genetic counseling) interactions with parents.

[Editor's Note: This issue's author, Dr. Philip M. Farrell, recommends interested clinicians access an outstanding review article (Newborn screening for cystic fibrosis: evidence for benefit) recently published in ***Archives of Disease in Childhood***[5] *that provides a comprehensive description of the key issues in CF NBS].*

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EDUCATION OF PROFESSIONALS AND PARENTS

Tluczek A, Koscik RL, Modaff P, et al. **Newborn screening for cystic fibrosis: parents' preferences regarding counseling at the time of infants' sweat test.** *J Genet Couns.* 2006;15(4):277-291.

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The objective of this study was to clarify important psychological issues by addressing 3 goals: 1) understanding parents' perceptions about genetic counseling received while awaiting their infants sweat test results; 2) identifying conditions that may affect the quality of their experience; and 3) developing a model for genetics counseling under the conditions of newborn screening using CF as a prototype. Utilizing Ground Theory (which is an interpretive research method that applies systematic analyses to qualitative data for the purpose of developing a theory about the studied phenomena and is derived from the social psychology of theory of Symbolic Interactionism), the design involved one hour, audiotaped interviews with 33 families whose infants had abnormal IRT/DNA tests. The group consisted of 24 false-positive families and 8 sets of parents whose child was diagnosed with cystic fibrosis; in addition there was 1 infant with a "borderline" sweat test. Two dimensions were explored during the interview: factual information and emotional aspects of the NBS follow-up experiences. The audiotapes were transcribed and coded by themes before analysis.

These 33 parental interviews, done when the babies were 1.5-6 months old, revealed a mix of parents' preferences. An analysis of the interview data revealed two central dimensions in parents' preferences, namely factual information and emotional aspects of the NBS follow-up experiences. The parents clearly wanted factual information from a "CF expert", and especially appreciated learning about a "low probability" of the disease. The parents had interesting comments to make about the timing of their genetic counseling experiences in relationship to the sweat test: while some preferred that the communication be initiated before the results were available, more parents preferred to know after the sweat chloride concentration was reported and explained to them. The study also revealed that there is a great challenge in managing the terminology around genetic screening. Even simple terms that clinicians take for granted, such as "false-positive", may be misleading and confusing, particularly because "positive" is generally considered the good thing. Perhaps the most interesting observations relate to emotional support. Many parents explicitly identified preferences for emotional support and wished to have the counselor show empathy and instill a sense of hope. Parents also appreciated a personalized approach and a hospitable setting. In this study, 84% of parents experienced good matching of experiences to their needs, but — importantly — 16% reported a mismatch such as receiving "too much information too soon".

Genetic counseling involves providing information and support to families who have members with genetic disorders and to families who may be at risk for a variety of inherited conditions. Most studies in the past have been focused on the views of health care providers, rather than parents, and have been most interested in outcomes such as cognitive gains, reproductive decision-making, satisfaction, etc, rather than in processes. Only recently have researchers attempted to identify those aspects of counseling that are important to the counselee. Consequently, this study is quite valuable in the context of CF NBS. Perhaps the most significant practical revelation relates to the value of good matching. Indeed, counseling that matched with parents' preferences reduced parental stress, while mismatched counseling tended to increase parents' worry and anxiety about their infant. The implications of this study include an emerging view that we may need to

redesign models of counseling, placing greater emphasis upon emotional support for parents, placing greater emphasis upon assessing the emotional states as well as informational needs of parents so that the timing, amount and method of counseling may be tailored to better match their needs.

SCREENING—SPECIMEN COLLECTION, SUBMISSION, AND TESTING

Scotet V, Audrézet MP, Roussey M, et al. **Immunoreactive trypsin/DNA newborn screening for cystic fibrosis: should the R117H variant be included in CFTR mutation panels?** *Pediatrics.* 2006;118(5):e1523-1529.

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The objective of this epidemiologic study was to evaluate issues regarding the CFTR panel used for the DNA component of the IRT/DNA screening test, with particular emphasis on a relatively common class IV mutation that occurs in exon 4 (R117H). Recognizing that class IV and V mutations are associated with pancreatic sufficiency and presumably mild lung disease, these investigators in Brittany, France organized their study to describe the clinical outcomes of 9 children averaging 7 years of age who were diagnosed as compound CF heterozygotes with one R117H allele. They were identified from 360,466 newborns who had IRT/DNA screening with a 30 mutation panel from 1995 through 2004. The outcomes included a variety of nutritional indices, and pulmonary function testing was performed in 2 of the children; however, no description is provided of respiratory symptoms or chest imaging observations.

The results of evaluating these children revealed that 4 had sweat chloride levels less than 30 mmol/L, while the others were between 30 and 59 mmol/L. All of the children had pancreatic sufficiency, and thus it was not surprising that they had normal nutritional status and growth. In addition, spirometric data available for 2 children "seem very good". One patient did have *Pseudomonas aeruginosa* cultured on one occasion. In essence, these children appeared to be asymptomatic.

R117H was the second most common mutation detected in the French CF NBS program, as has also been the case in other regions such as Massachusetts and Wisconsin. However, as the authors point out, this mutation is not generally detected when CF patients are diagnosed as a result of signs and symptoms (i.e. in the prescreening era). The authors propose "withdrawal of the R117H variant from the panels of CFTR mutations used in cystic fibrosis newborn screening" because "it is of the utmost importance that only mutations that result in classical cystic fibrosis are included". Their proposal is currently being considered in France and has been discussed in other regions as well. However, concurrent studies in Massachusetts suggest that acquisition of *Pseudomonas aeruginosa* may occur with some regularity in patients with this mutation.[1] In addition, a collaborative study between the Brittany molecular genetics laboratory and Wisconsin revealed that even patients with pancreatic sufficiency may develop lung disease as children, based on longitudinal chest X-ray scores, even though its appearance occurs later than in F508del/F508del patients.[2] Further, it has become clear during recent years that spirometry in young children with CF is not sufficiently sensitive to reveal lung disease, and that either longitudinal quantitative chest radiography or chest HRCT scans are needed.[3] Consequently, as an alternative to the proposal of Scotet et al, if one accepts the assumption that we need to know more about such patients, prudence favors continuing to diagnose them early through NBS and evaluate them in even more detail for longer periods of time.

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FOLLOW-UP OF ABNORMAL AND UNSATISFACTORY TEST RESULTS

Sims EJ, Clark A, McCormick J, Mehta G, Connett G, Mehta A; United Kingdom Cystic Fibrosis Database Steering Committee. **Cystic fibrosis diagnosed after 2 months of age leads to worse outcomes and requires more therapy.** *Pediatrics.* 2007;119(1):19-28.

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This important article addresses the long-standing issue of determining the maximum acceptable age for diagnosing cystic fibrosis with a newborn screening program; i.e. when are the disease manifestations significant enough that a delay in diagnosis causes long-term adverse effects? A classic article[1] by Shwachman et al in 1970 suggested that diagnosis with symptoms after 3 months of age resulted in a worse prognosis based on survival statistics, but CF mortality has never been a satisfactory outcome measure. The objective of Sims et al was to compare outcomes and treatments of patients clinically diagnosed during the newborn screening "reporting window" (early clinically diagnosed group) with those presenting after this period (late clinically diagnosed), and with patients diagnosed by newborn screening. The key design elements included a cross-sectional analysis of patients homozygous for F508del who were registered in the UK Cystic Fibrosis Database and received care at "specialist cystic fibrosis centers". Patients were 1-10 years of age during the interval of 2000-2002. Two analyses were performed using a variety of outcome measures related to nutritional and pulmonary status. In addition, a novel comparison of the intensity of long-term therapy was performed. In the primary analysis, patients diagnosed through NBS were matched to the other 2 groups, yielding a total of 133 per diagnostic cohort – but this method may have skewed the analysis toward an older, and therefore more severely affected population (introducing a potential selection bias). Consequently, a secondary analysis was done with similar matching over 3 years of evaluation, which resulted in matched data sets that were evenly distributed across all ages.

The results of the primary and secondary analyses showed that patients diagnosed through NBS had significantly higher height Z-scores and clinical scores, suggesting that they were healthier, and indeed fewer NBS patients were below the 10th percentile for height compared with the late clinically diagnosed patients. There were no differences found in FEV^1. Patients in the NBS cohort received significantly fewer long-term therapies than the late clinically diagnosed patients. Although no significant differences were found in the prevalence of chronic or intermittent *Pseudomonas aeruginosa* infection between the cohorts, fewer 1 to 5 year-old patients diagnosed through NBS were chronically infected with *Pseudomonas aeruginosa*. Most interestingly, the patients diagnosed early received significantly less treatment.

One of the great strengths of this study is the focus on long-term evaluation of comparable cohorts who are homozygous for F508del. Because Sims et al defined "clinically late diagnoses" as after 2 months of age, it is reasonable to conclude that 2 months is the maximum acceptable age of diagnosis. In addition to being healthier, patients diagnosed before that time also required less of a "treatment burden". However, it should be noted that the choice of therapies is typically a physician-driven outcome, and that clinicians will vary in their habits with regard to long-term therapies of cystic fibrosis. While the authors point out that "policymakers have widely used the reasoning that CF screening is of no benefit because lung function is no better for patients with CF whether they have been screened at birth or not", this tendency is quite unfortunate and reflects a lack of understanding of current pulmonary function data reported for children with CF. In fact, spirometry is relatively insensitive to early lung disease, and even structurally irreversible lung disease (i.e. bronchiectasis) might occur before obstructive pulmonary dysfunction as detected by spirometry. Thus, investigations such as the study by Sims et al should utilize chest imaging techniques to enhance the sensitivity of their comparisons.

[Editor's Note: Please access Dr. Farrell's 2007 article in Pediatrics[2] for additional information.]

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CONFIRMATORY TESTING AND DIAGNOSIS

Doull IJ, Hall SJ, Bradley DM. **A sweat test centered protocol for the disclosure and diagnosis of cystic fibrosis in a newborn screening program.** *Pediatr Pulmonol.* 2007;42(9):773-778.

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The objective of this article was to report observations from the newborn screening program of Wales, examine the timing and follow-up process when IRT/DNA screening was positive, and assess parental satisfaction. The IRT/DNA test with a 32 CFTR mutation panel has been used in Wales since 1996, and the follow-up system evolved there with elements such as a "lead pediatrician" at each hospital and a protocol for the disclosure and diagnosis processes that was driven by the timing of the sweat test. In practice, Wales uses a follow-up sweat test as "the cornerstone of the process of diagnoses". Generally, a sweat test is arranged for the first possible date after a positive NBS test; the parents are then informed about the possibility of CF the day before the sweat test. The intent is to ensure the minimum waiting period in an effort to alleviate anxiety. In addition the authors report that "to prevent families being left unsupported over weekends, we do not contact families on Thursday or Friday". They further state that "on the day of the sweat test parents are met by the CF nurse specialist and the lead CF clinician prior to the test", and that after the sweat test is performed the result is given to the parent "within an hour or two by the lead CF clinician".

During the study period, a total of 295,247 newborns were screened, resulting in 121 being diagnosed with CF, 83 of whom had 2 mutations; thus 69% were presumptively or genetically diagnosed from the IRT/DNA test. There were at least 4 false negative tests (3%). Although the authors state that they "aim to diagnose a majority of our patients with CF within five weeks of birth", the actual age at diagnosis is not reported in the article; however, I contacted Dr. Doull and learned that the median age of the sweat test was 38 days during the 1996-2005 study period but apparently has decreased to 21-28 days after DNA methods were improved. The results of simple parental satisfaction surveys were quite encouraging, with questionnaire responses from parents of both affected and heterozygote carriers showing no significant differences in parental satisfaction.

This article illustrates the top priority given, whenever a positive IRT/DNA result occurs, to minimizing the delay from initial parental contact to the definitive resolution that occurs with a sweat test, and is a tactic that should be an essential component of all CF NBS programs to help ensure more good than harm. In fact, the follow-up program in Wales might be considered an international role model with regard to minimizing the anxiety/anger-provoking delay between notification of the positive NBS test and the definitive sweat test. Further, the screening test methods in Wales are state of the art: IRT by the Autodelphia method of Perkin Elmer and 31 mutation analysis. These aspects are critical for all regions to pay attention to as they undertake their quality improvement efforts.

[Editor's Note: For additional information, please access Dr. Farrell's Pediatrics article[1] on "shifting the key question"]

References

1. Farrell PM. Cystic fibrosis newborn screening: Shifting the key question from "Should we screen?" to "How should we screen?" *Pediatrics.* 2004;113(6):1811-1812.

MEDICAL MANAGEMENT AND PERIODIC OUTCOMES EVALUATION

Sims EJ, Mugford M, Clark A, et al; UK Cystic Fibrosis Database Steering Committee. **Economic implications of newborn screening for cystic fibrosis: a cost of illness retrospective cohort study.** *Lancet.* 2007;369(9568):1187-1195.

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The objective of this study was to determine the therapies and their annual costs in 184 cystic fibrosis patients diagnosed through newborn screening, and to compare those data with 950 patients who were clinically diagnosed at 1-9 years old during 2002. The group used the UK Cystic Fibrosis Database with one subgroup analysis (examination of F508del homozygotes). Their goal was to test the hypothesis that the cost savings from early diagnosis through NBS could offset the incremental cost of the screening *per se*. Thus, this is a cost-of-illness retrospective cohort study, although it provides only a partial cost analysis because it did not include supplemental feedings, counseling, collection of newborn blood spots, and health promotion visits.

The results provide valuable data on the long-term therapies given for 3 months or more to CF patients and their pharmacologic expenses. It was determined that the costs of therapy for patients diagnosed through NBS were significantly lower than those for clinically diagnosed patients: $7,228 compared to $12,008. For the F508del homozygotes, the savings were even more impressive. Indeed, the mean or median drug cost savings could have offset the estimated cost of adding CF to the UK national newborn screening service.

This study demonstrates that in addition to nutritional benefits, which provide the strongest evidence in favor of CF NBS, there are significant economic benefits associated with early diagnosis. It is indeed remarkable that a method of diagnoses would pay for itself through savings in subsequent therapies. Clearly, any region that is uncertain about whether or not to implement newborn screening for CF should pay close attention to this article and its implications, even though more research of this type is needed because the costs of caring for CF patients long term have not been adequately studied.

SYSTEM QUALITY ASSURANCE

Farrell MH, Kuruvilla P. **Assessment of parental understanding by pediatric residents during counseling after newborn genetic screening.** *Arch Pediatr Adolesc Med.* 2008;162(3):199-204.

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The objective of this study was to perform a baseline evaluation of an aspect of the quality of communication about positive NBS results by 32 pediatric residents. This investigation also developed new quality assurance communication methods to quantify physician behavioral skills in educating parents regarding genetic screening results. After attending a brief educational session about CF and sickle cell hemoglobinopathy, residents were placed in a familiar office setting where the investigators audiotaped 2 separate encounters with a simulated mother. The tapes were then transcribed and analyzed by an explicit-criteria abstraction technique modeled after medical chart review. The analysis of communication quality focused on “assessment of understanding questions” (AUs), which are important when a physician needs to verify a parent’s knowledge about a topic or that a message has been successfully conveyed. The investigators developed a taxonomy of four types of AUs, ranging from ambiguous acknowledgement (“OK?”) at the lowest end to close-ended questions (“Does that make sense?”) to open-ended questions (“What parts of this are hard to understand?”) to requests for the parent to reiterate the content (“To make sure I was being clear enough, could you please share with me the main points you got from our discussion?”).

Interabstractor reliability was excellent, with a kappa statistic of 0.93. Only 44.1% of the transcripts met the criteria for at least 1 definite (not a simple “OK?”) AU. Among these,

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the mean (SD) number of AU's per abstract was 5.5 (7.6). There were no statistically significant differences related to seniority, sex, or duration of the counseling session (range=5-20 minutes). Based on a A to F grading system, the average score was D+, and the best grade was B, which was achieved by only 3 transcripts. Furthermore, 94.9% of the transcripts were determined to be in need of "targeted feedback" quality improvement.

Although generalizability may be limited by small size, this study suggests that pediatricians in advanced GME training are in need of quality improvement with regard to conveying screening test results as part of a risk communication session with simulated mothers. In particular, it was observed that there was a preponderance of missed opportunities to assess and confirm understanding, i.e. a paucity of AUs. While there might be some concern that the project focused on physicians in training rather than more experienced pediatric practitioners, it can be posited that this group might be the perfect study population because they are the new generation of primary care physicians who will be the first to enter the era of molecular medicine. A larger implication of this paper comes from its demonstration of a method to assess what the authors call a "communication quality indicator" that will be affordable and quantitatively reliable to use over entire geographic regions. An Editorial[1] accompanying this article stated that "it would be wonderful to have a replicable marker for good communication or a series of markers that could be applied over time and incorporated into a health care organization's routine feedback to clinicians". While the authors admit that further study of such a population-scale approach to communication quality assessment after NBS screening needs to be done, it should be noted that communication about CF carrier and other false positive NBS results is so challenging for primary care providers and specialists that this sort of work should be a very high priority. In addition, it would be ideal to have methods that assess and improve communication while addressing the psychosocial challenges of parents.

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Learning Objectives — back to top

At the conclusion of this activity, participants should be able to:

- Discuss with colleagues the six components of cystic fibrosis (CF) newborn screening (NBS) program
- Describe to colleagues the NBS-linked method and maximum acceptable age of CF diagnosis
- Explain to colleagues the medical and economic benefits of diagnosing CF through newborn screening

Internet CME/CNE Policy — back to top

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Respiratory Therapists

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RESPIRATORY THERAPIST POST-TEST

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