Expanded newborn screening in Texas: a survey and educational module addressing the knowledge of pediatric residents

Amy Stanford Wells, MS[1], Hope Northrup, MD[1], Sharon S. Crandell, MD[2], Terri M. King, PhD[3], Neena L. Champaigne, MD[5], Michael Yafi, MD[4], Bradford L. Therrell, PhD[6], and Sarah Jane Noblin, MS[1]

Purpose: To assess the effectiveness of an educational module as a tool for improving the knowledge of pediatric residents about newborn screening and its expansion in Texas. **Methods:** The study population consisted of 63 pediatric residents from the University of Texas at Houston, Baylor College of Medicine in Houston, and the University of Texas Medical Branch in Galveston. Residents were invited to participate in the study during daily scheduled didactic lectures in their respective residency programs. Questionnaires were distributed to the residents both before and after the presentation of an educational module about newborn screening in Texas to assess whether knowledge was gained from the presentation. **Results:** Analysis of questionnaires from the full group of participants showed a substantial increase in knowledge about newborn screening in Texas after the presentation of the educational module. This included a 45.4% increase in knowledge about pre-expansion newborn screening conditions and a 308.4% increase in knowledge about expanded newborn screening conditions ($P \leq 0.001$). **Conclusions:** Our results suggest that an educational module about newborn screening like the one we created for this study would be an effective tool for improving the knowledge of pediatric residents on a larger scale. ***Genet Med*** **2009:11(3):163–168.**

Key Words: *newborn screening, expanded newborn screening, pediatric residents, education, Texas*

Newborn screening has a rich and interesting history that began more than 70 years ago with the discovery of the inborn error of metabolism, phenylketonuria (PKU).[1] Almost 30 years after this discovery, Dr. Robert Guthrie reported his development of a newborn screening test for PKU using a bacterial inhibition assay performed on blood dried onto filter paper.[2,3] His process for collecting, transporting, and analyzing blood in this way provided a mechanism for screening newborns within the first days of life that eventually led to a world-wide effort to include PKU testing as a routine preventive public health strategy.[4]

The evolution of newborn screening in Texas

Newborn screening for PKU was mandated by Texas law in 1965.[5] A 1977 law mandated screening for congenital hypothyroidism, however no funding was provided. The funding issue was remedied by the 1979 legislature and screening for congenital hypothyroidism, galactosemia, and homocystinuria officially began on February 1, 1980 (although limited pilot testing for galactosemia and homocystinuria had been ongoing since 1979).[6,7] No cases of homocystinuria were detected during the next 3 years (~1 million screens), and as a result on November 1, 1983, screening for homocystinuria was discontinued in favor of screening for sickle cell anemia and other hemoglobinopathies.[8] Screening for congenital adrenal hyperplasia was added on June 1, 1989.[9] This panel of five disorders detectable from dried blood spot screening remained unchanged until December, 2006 when Texas expanded this area of the state newborn screening program. Actions by the Texas Department of State Health Services (DSHS) in 2006 meant that Texas newborns would be routinely screened for 28 of the 29 conditions recommended by the American College of Medical Genetics in their March 2005 report, "Newborn Screening: Toward a Uniform Screening Panel and System."[10,11] Cystic fibrosis was the only condition not included in the expansion. Table 1 provides a list of conditions included in the Texas expanded newborn screening program along with their commonly used abbreviations.

The addition of certain metabolic conditions to the newborn screening panel required the use of tandem mass spectrometry, a sophisticated and complex technology previously reserved for diagnostic applications. This technology allows for the simultaneous analysis of many metabolic conditions concurrently, as opposed to the previous method of analyzing each condition individually.[12] The recent newborn screening expansion in Texas and in other states has highlighted a knowledge gap in medical practice in which clinicians are generally unprepared for the rapidly expanding number of detectable conditions from newborn screening. Improving the knowledge of health care professionals in regards to screening procedures and referral practices is essential in order for the programs to reach their full potential. It is important for health care providers to not only have knowledge of the conditions being screened for, but to also have knowledge of referral procedures when abnormal results arise. It was our hope that an educational module focused on newborn screening in the state of Texas would improve the knowledge of pediatric residents included in our study. This should in turn lead to increased knowledge of practitioners in the state, because many of these residents will practice here after completion of their residency programs. In this study, we

From the [1]Division of Medical Genetics, Department of Pediatrics; the [2]Division of Medical Education, Department of Pediatrics; the [3]Children's Learning Institute, Department of Pediatrics; and the [4]Division of Endocrinology, Department of Pediatrics, The University of Texas Medical School at Houston, Houston, Texas; the [5]Division of Medical Genetics, Department of Pediatrics, The University of Texas Medical Branch, Galveston, Texas; and the [6]National Newborn Screening and Genetics Resource Center, The University of Texas Health Science Center at San Antonio, San Antonio, Texas.

Hope Northrup, MD, Division of Medical Genetics, Department of Pediatrics, The University of Texas Medical School at Houston, MSB-3.144, 6431 Fannin Street, Houston, Texas 77030. E-mail: Hope.Northrup@uth.tmc.edu.

Disclosure: The authors declare no conflict of interest.

Submitted for publication July 2, 2008.

Accepted for publication October 25, 2008.

DOI: 10.1097/GIM.0b013e3181930787

Table 1 Conditions included in the Texas Expanded Newborn Screening Program

Group	Condition	Abbreviation
Organic Acidemias	3-Methylcrotonyl-CoA carboxylase deficiency	3MCC
	Beta-ketothiolase deficiency	BKT
	Glutaric acidemia type 1	GA I
	Hydroxymethylglutaric aciduria	HMG
	Isovaleric academia	IVA
	Methylmalonic academia, Cbl A/Cbl B forms	MMA CblA/B
	Methylmalonic academia, mutase deficiency form	MMA mut
	Multiple carboxylase deficiency	MCD
	Propionic academia	PROP
Fatty Acid Oxidation Disorders	Carnitine uptake defect	CUD
	Medium-chain acyl-CoA dehydrogenase deficiency	MCAD
	Long-chain hydroxyacyl-CoA dehydrogenase deficiency	LCHAD
	Very-long-chain acyl-CoA dehydrogenase deficiency	VLCAD
	Trifunctional protein deficiency	TFP
Amino Acid Disorders	Argininosuccinic academia	ASA
	Citrullinemia	CIT
	Homocystinuria	HCY
	Maple syrup urine disease	MSUD
	Phenylketonuria	PKU
	Tyrosinemia type 1	TYR I
Hemoglobinopathies	Sickle cell anemia (Hb SS)	
	Sickle beta thalassemia (Hb S/β Th)	Heme
	Sickle-hemoglobin C disease (Hb S/C)	
Others	Galactosemia	GAL
	21-OH deficient congenital adrenal hyperplasia	CAH
	Congenital hypothyroidism	CH
	Biotinidase deficiency	BIOT
	Hearing deficiency	Hearing

aimed to determine the effectiveness of our educational module and to identify strengths and weaknesses in general knowledge about newborn screening among pediatric residents.

MATERIALS AND METHODS

The study population consisted of 63 pediatric residents from the University of Texas at Houston, Baylor College of Medicine in Houston, and the University of Texas Medical Branch in Galveston. Residents were invited to participate in the study during their daily scheduled didactic lectures in their respective residency programs. Because the study was educational, it was ruled exempt by the Institutional Review Board (IRB) at the University of Texas at Houston. In accordance with the IRB process, a letter stating the purpose and volunteer nature of the study was read to all participants before any data were collected. Medical students attending the presentations were excluded from the study.

An educational module in the form of a PowerPoint presentation was developed to provide the pediatric residents with information about newborn screening and the expansion of the newborn screening program in Texas. The presentation was designed as an overview, focusing on main points in the areas of general information about newborn screening in Texas, referral procedures for abnormal screening results, information about newborn screening technology, and a listing of the conditions screened for before and after expansion of the state's newborn screening program. During the months of December,

2006 and January, 2007, the educational module was presented to pediatric residents from the three different residency programs. Presentations were given at a total of five locations. Presentation sites for the University of Texas at Houston residency program included Memorial Hermann Hospital—Texas Medical Center and Lyndon B. Johnson General Hospital. Presentations to residents from the Baylor College of Medicine program were given at Texas Children's Hospital and Ben Taub General Hospital. The fifth presentation was given to residents at the University of Texas Medical Branch in Galveston.

To measure the residents' base knowledge of newborn screening in Texas, a questionnaire was distributed before the presentation of the educational module. Another questionnaire containing the same questions in mixed order was given to the participants immediately after the presentation of the educational module. The pre- and postpresentation questionnaires were matched from each participant, allowing for a matched data analysis. A copy of the letter required by the IRB was attached to the front of each questionnaire packet, and the packets were numbered as a means of identification. No personal identifiers were obtained from the participants of the study.

Each questionnaire included a demographic section, followed by a section with general questions about newborn screening. These sections were followed by two additional sections containing questions about the conditions included in pre-expansion newborn screening and expanded newborn screening in Texas.

Data collected from the questionnaires was number-coded and entered into a Microsoft Excel file in preparation for analysis. Participants' responses from the pre- and postpresentation questionnaires were compared to assess whether or not the educational module had improved their knowledge. Most of the questions included in the surveys were in a closed format (yes/no or true/false). Two score-based questions required participants to choose from a list the correct conditions included in the Texas newborn screening program both before and after the expansion. Average scores from the score-based questions on the pre- and postpresentation questionnaires were standardized to a "blank test normal" to allow for a clearer representation of the results. A blank test normal centers the score around a score of zero and avoids including a large number of "negative corrects" in the final score for these questions. Negative corrects in this case are defined as conditions that were not chosen by the participant and are not included in pre-expansion or expanded newborn screening. A positive standardized score indicates that the participant chose more correct conditions than incorrect conditions whereas, a negative standardized score indicates that they chose more incorrect conditions than correct conditions.

All statistical tests were performed electronically using Statistical Package for the Social Sciences (SPSS).[13] Closed format questions were analyzed using the Wilcoxon signed ranks test, which measures whether there was a significant change in residents' responses from the pre- to the postpresentation questionnaire. Because the Wilcoxon test measures paired data, only those individuals who answered a particular question on both the pre- and the postpresentation questionnaire were included in the analysis. Therefore, the number of participants analyzed varied from question to question, as some residents left a particular question blank on either the pre- or the postpresentation questionnaire and thus were excluded from Wilcoxon analysis.

Average scores from the score-based questions were analyzed using paired-*t*-tests to assess whether or not there was an improvement in the residents' knowledge about conditions included in both pre-expansion and expanded newborn screening in Texas after the presentation of the educational module. Results with a *P*-value less than or equal to 0.05 were considered significant.

The educational module and questionnaire used in this study are available by request through correspondence with the address provided.

RESULTS

Table 2 summarizes demographic information for the 63 residents who participated in the study. In this publication, we will focus on analysis of the entire group of these participants. Figure 1 shows the six conditions included in pre-expansion newborn screening in Texas, and graphically illustrates the pre- and posteducation changes in answers among the participants (refer to Table 1 for condition abbreviations). Participating residents showed an increase in knowledge about 5 of the 6 pre-expansion conditions after the presentation of the educational module. Interestingly, the percentage of residents who correctly chose PKU decreased slightly from 96.8% on the prepresentation questionnaire to 95.0% on the postpresentation questionnaire. Figure 2 shows all conditions included in the expanded newborn screening program in Texas, and the percentage of participants who correctly chose each condition on their pre- and postpresentation questionnaires (refer to Table 1 for condition abbreviations). The three hemoglobinopathies included in the expanded panel were grouped as one disorder (Heme), whereas, the different forms of methylmalonic acidemia (MMA) were listed separately. Residents showed an increased knowledge about all conditions in the expanded program after the presentation of the educational module.

Closed format questions

Of the 62 closed format questions included in the questionnaires, 40 (66.7%) returned significant results based on Wil-

Table 2 Demographic information for study participants ($n = 63$)

	Number	%
Male	22	34.9
Female	41	65.1
Age (yr)		
≤25	4	6.4
26–35	58	92.1
36–45	1	1.6
≥46	0	0.0
Residency Program		
UTH	16	25.4
BCM	27	42.9
UTMB	20	31.8
Attended Medical School in Texas		
Yes	31	49.2
No	32	50.8

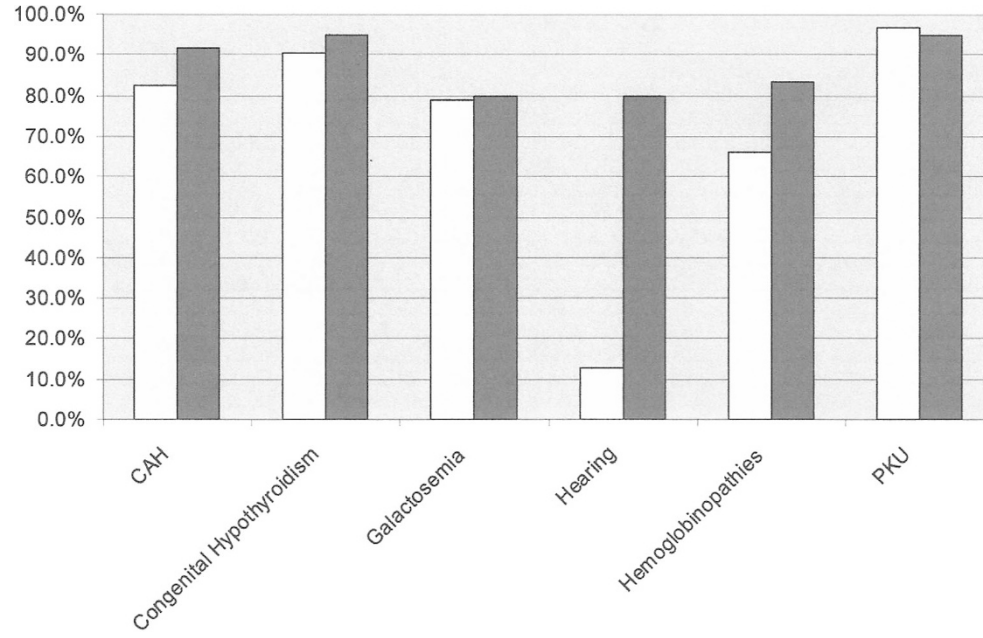

Fig. 1. Percentage of residents who correctly chose each pre-expansion NBS condition before and after the educational module. □, Before educational module; ■, after educational module. Note: Refer to Table 1 for condition abbreviations.

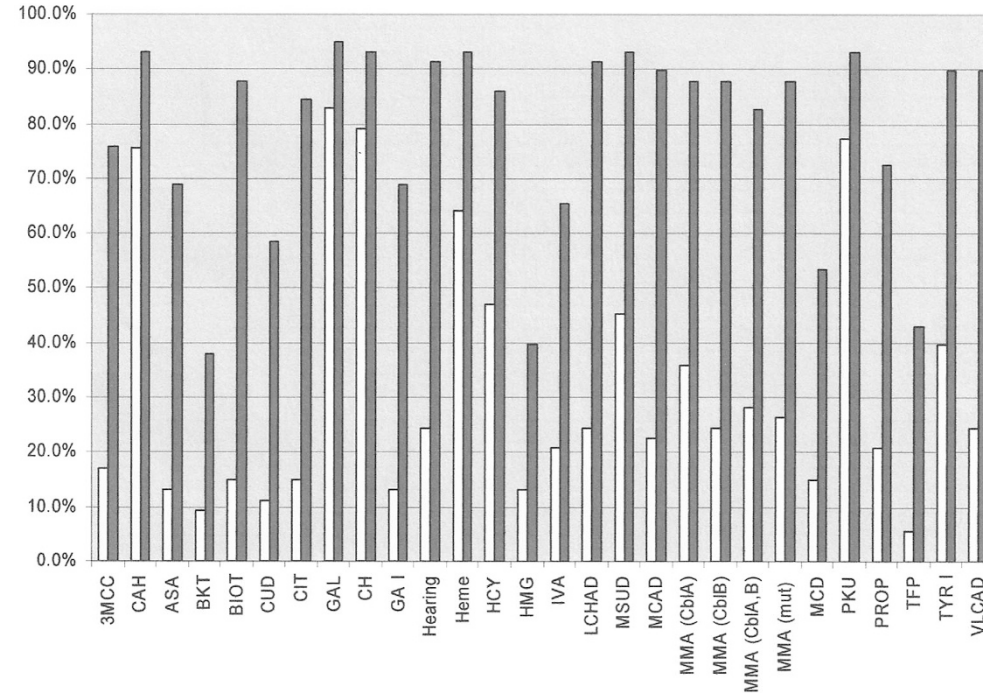

Fig. 2. Percentage of residents who correctly chose each expanded NBS condition before and after the educational module. □, Before educational module; ■, after educational module. Note: Refer to Table 1 for condition abbreviations.

coxon signed ranks analysis ($P \leq 0.05$). Thirty-nine of the 40 questions that returned a significant result were positively significant, meaning that the participating residents learned the answers to those questions from the educational module. In other words, a significant number of residents answered 39 of the 62 closed format questions incorrectly on the prepresentation questionnaire and then changed their answer to the correct choice after the presentation of the educational module. The one question that returned a negative significant result was a true/false question about pre-expansion newborn screening which stated that there is "no need to contact the screening laboratory if no results are reported, as only ab-

normal newborn screens are reported" (i.e., no news is good news). Although only a few residents switched to the incorrect answer, "true," after the presentation of the educational module, there was enough of a shift in responses to result in a positive Wilcoxon test. Despite the negative significant result for this question, the overwhelming majority of residents answered "false" correctly on both the pre- and the postpresentation questionnaire.

Score-based questions

A paired *t*-test performed on the average scores from the question asking residents to choose conditions included in pre-expansion newborn screening in Texas revealed a 45.4% increase in the average score after the presentation of the educational module, a significant change ($P \leq 0.001$). A paired *t*-test performed on the average scores from the question asking residents to choose conditions included in expanded newborn screening in Texas also yielded a significant result, showing a 308.4% increase in the average score from pre- to postpresentation questionnaires ($P \leq 0.001$).

DISCUSSION

With ever-evolving technology, more conditions are being screened for during the newborn period than ever before. The majority of states in the United States have implemented an expansion of their newborn screening programs due to the introduction of tandem mass spectrometry as a new and efficient means of screening methodology.[12,14] Texas' expansion of its newborn screening program requires all newborns in the state to be screened twice for 27 disorders detectable through a blood sample, and for hearing deficiency.[15] The extent to which clinical practitioners in Texas are aware of the expanded newborn screening requirements or even the pre-expansion program is not known. This study was conducted to assess the effectiveness of an educational module as a tool for improving the knowledge of pediatric residents about newborn screening and the expansion of newborn screening in Texas.

First, we looked at the baseline knowledge of residents in regards to the Texas newborn screening program through a questionnaire given before the educational module was presented. After their participation in the educational module, their knowledge was once again assessed using a randomized version of the same questionnaire. Improved knowledge was apparent in 5 of the 6 pre-expansion conditions after the presentation of the educational module. The decrease in residents who correctly chose PKU as a pre-expansion condition after the presentation of the educational module may have occurred because some of the residents were concentrating more on the conditions that they had not known before the educational module and perhaps mistakenly left out PKU, a more familiar condition, on the postpresentation questionnaire. All 28 conditions included in the expanded newborn screen saw a substantial increase in the percentage of residents who chose them correctly on the postpresentation questionnaire, a good indication that these conditions were unfamiliar to most participants before the presentation of the educational module.

When we looked for significant changes in knowledge within the closed format questions, we were impressed with the number of questions answered incorrectly on the prepresentation questionnaire and then changed to the correct answer after the presentation of the educational module. Thirty-nine of the 62 closed format questions on the questionnaire showed a positive significant shift in knowledge after the presentation of the educational module. Interestingly, there were no specific subject areas in which residents showed a particular lack or proficiency of knowledge before the educational module was presented. Information known or not known by the residents before the presentation of the educational module was distributed relatively evenly among the subjects of general newborn screening knowledge, newborn screening technology, pre-expansion newborn screening in Texas, and expanded newborn screening in Texas. Consequently, knowledge gained from the educational module was also distributed relatively evenly among the same subjects.

The score-based questions allowed us to focus specifically on the residents' knowledge of conditions included in both pre-expansion and expanded newborn screening in Texas. As expected, we saw a significant score increase, and thus increase in knowledge, for both the pre-expansion and expanded conditions from pre- to postpresentation questionnaire. Because the conditions included in pre-expansion newborn screening are also included in the expanded panel, these conditions appear graphically in both Figures 1 and 2. Interestingly, the number, and thus the percentage of residents who correctly chose the pre-expansion conditions differ between the two figures. This is because residents were first required to choose conditions included in pre-expansion newborn screening only, and then in a separate question were asked to select all conditions included in the expanded panel. The significant increase in knowledge seen especially with the expanded conditions is again consistent with the idea that the conditions included in expanded newborn screening in Texas are not generally well known, and were consequently learned from the educational module.

CONCLUSION

Our study showed that an educational module about newborn screening and the expansion of newborn screening in Texas was an effective and efficient tool for improving the overall knowledge of the pediatric residents who participated in the study. It seems, based on the results of this study, that an educational module like the one we created would be beneficial to pediatric residents on a larger scale. Not only is it important for pediatric residents to be aware of the conditions for which screening is required in their state's newborn screening program, but it is also important to understand the infrastructure of the program to provide appropriate and optimal medical care to newborns. It is our hope that the pediatric residents who participated in this study not only have a better understanding of newborn screening in Texas, but now have a better understanding of the purpose of genetics professionals and when to make referrals to geneticists, genetic counselors, and other specialists.

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