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# Maternal Knowledge and Attitudes about Newborn Screening for Sickle Cell Disease and Cystic Fibrosis

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## Abstract

Illinois introduced mandatory newborn screening (NBS) for sickle cell disease (SCD) in 1989 and for cystic fibrosis (CF) in 2008. We examined maternal understanding of NBS for SCD and CF, and their knowledge of the genetics, symptoms and treatments of both conditions. Our methods consisted of conducting interviews of inpatient post-partum women (>18 years and English speaking). Our results showed that of the 388 eligible participants, 34 self-identified as sickle cell carriers, 1 with SCD and 1 as a CF carrier. Almost 3/4 were African American (282/387). Although all but 5 women had prenatal care, only 35% (133/378) recalled their prenatal care provider mentioning NBS, and only 56% (217/388) of participants recalled nursery staff mentioning NBS. There was more self-reported familiarity with SCD (3.32/5) than CF (1.97/5, $p<0.001$). Over 2/3 (260/388) of participants could not answer CF knowledge questions because they had never heard of CF. Among those who had heard of the conditions, mean knowledge scores were 66% for SCD (n=372) and 63% for CF (n=128). Bivariate analysis identified education, age, race, marital status, and insurance status as statistically significant. After linear regression education remained significant for both conditions. We conclude that in a sample of predominantly African American post-partum women, we found poor understanding of NBS, greater familiarity with SCD, and significant knowledge gaps for both SCD and CF. There are many missed educational opportunities for educating parents about NBS and specific conditions included in NBS panels in both the obstetric clinics and the nursery.

### Keywords

newborn screening; sickle cell disease; cystic fibrosis; parental understanding; knowledge; attitudes and beliefs

## INTRODUCTION

Newborn screening (NBS) for phenylketonuria (PKU) became possible in the early 1960s when Guthrie developed both the bacterial inhibition assay to measure phenylalanine from blood and the filter paper on which to collect and test the blood samples [Guthrie and Susi, 1963]. In 1963, Massachusetts became the first state to pass mandatory NBS legislation for PKU screening. Guthrie and the National Association for Retarded Children (NARC now known as the ARC) lobbied for mandatory screening to ensure that all children with preventable retardation were identified and most states adopted such legislation [Koch,

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1997]. Today, mandatory NBS exists in 48 states; parental consent is needed in the other two.

One concern with a mandatory screening program is what parents understand. When consent is needed, health care providers must ensure, at minimum, that parents know that screening is being done and that the parents agree to its performance. Ideally, health care provider would also explain why the test is being done, what the risks and benefits are, and what follow-up the parents can expect. Although mandatory screening does not preclude such discussions, data show that they often do not occur [Campbell and Ross, 2003; Davis et al., 2006; Detmar et al., 2007].

Discussions about NBS are important because lack of parental understanding may cause harm. If parents do not understand that an abnormal NBS result requires diagnostic testing to confirm or refute a presumptive diagnosis, they may fail to obtain appropriate follow-up in a timely manner and threaten the success of reducing morbidity and mortality [Arn, 2007; James and Levy, 2006; Miller et al., 1990].

In Illinois, NBS has existed since 1965 and has always been mandatory. In this manuscript, we examine parental understanding of NBS for two conditions, sickle cell disease (SCD) (introduced in Illinois in 1989) and cystic fibrosis (CF) (introduced in Illinois in 2008). NBS for both these autosomal recessive genetic conditions reveals some information about heterozygote carriers. We examined maternal attitudes and knowledge about NBS for SCD and CF, about the genetics, symptoms and treatments for SCD and CF, and how women learn about NBS. We hypothesized that there would be greater knowledge of SCD than CF; and that most women would be unfamiliar with specific details about NBS, but that they would be supportive of NBS.

## METHODS

We interviewed women in the post-partum unit of the University of Chicago Hospitals whose infants were admitted to the general care nursery. Interviews (conducted by CL and AS) took place Monday to Friday over 17 consecutive weeks (June-October 2008). Women who were less than 18 years old, were non-English speaking, or had DCFS (Department of Child and Family Services) involvement were not approached. Women were also excluded if the obstetricians, pediatricians, or social workers indicated they were inappropriate for inclusion. At initial contact, women were read a brief description of the study and offered a $5 incentive for participation. They were offered an opportunity to participate immediately or at another time prior to discharge. Those that were not interested were not recontacted. Oral consent was obtained prior to participation. In order to maximize uptake, interviews were conducted whenever the women were available, even if the child had not yet had blood taken for NBS.

The survey addressed knowledge of and attitude towards NBS, personal experience with genetic testing, and knowledge about the genetics, symptoms and treatments for SCD and CF. The questions about SCD were adopted from Adewuyi [2000] and a website sponsored by the Centers for Disease Control and Prevention (CDC) [2009]. Parallel questions were developed for CF.

If participants had never heard of SCD or CF they were not asked subsequent questions about those conditions. A separate section asked all participants their familiarity with and their perceptions of the severity of SCD and CF, using a 5 point Likert Scale (1=never heard of it to 5=very familiar and 1=not serious to 5=very serious, respectively). Participants had the option of saying they “did not know” how serious a condition was, and their response was excluded. Survey items were read aloud to all participants. Questions about carriers of

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SCD were phrased as “carriers of SCD” and repeated as “individuals with sickle cell trait (SCT)”. Throughout the survey, participants could ask for clarification of the questions but were not provided with correct answers for factual questions. If unsure of the answer to True/False questions, participants were encouraged to guess. If they refused to guess, the response was coded as “not sure” (<0.5% for each question). Demographics were also collected. At the conclusion of the survey, participants were offered some basic information about NBS, and the genetics, symptoms and treatments of SCD and CF. Participants were also given the opportunity to review the correct answers to knowledge questions and to ask questions about topics covered in the survey.

Descriptive statistics were performed. Responses to True/False knowledge questions were scored as correct or incorrect. A “not sure” was coded as incorrect. Percent correct (mean score) for all 20 questions was calculated. T-tests were performed comparing demographics against the mean scores with significance set at $p<0.05$. Because of the significant colinearity between 5 demographic variables (age, marital status, education, insurance statue, and race), linear regressions were conducted with mean scores as the dependent variable and the 5 demographics as independent variables. Statistics were performed using SPSS 16.0.

University of Chicago Institutional Review Board approved this project and allowed for oral consent and the waiver of written informed consent.

## RESULTS

Of the 477 women who were in residence on interview days, 51 (11%) were not approached for study participation due to exclusion criteria: <18 years old, non-English speaking, DCFS involvement, or medical team/social work exclusions. Eleven (3%) women were approached but refused participation. Fifteen (4%) were approached and expressed interested in study participation but were discharged before being interviewed.

In total, 400 interviews were conducted. Of those, 12 were excluded (2 because they did not meet inclusion criteria, 1 was interrupted for patient care and not completed, and 9 were physicians). Physician-mothers were excluded after their responses were reviewed and were found to be significantly different from responses of the remaining lay participants (data not shown) and to be more similar to responses in a parallel survey of physician knowledge and attitudes towards NBS that was being conducted (Stark and Ross, manuscript under review).

Of the remaining 388 participants, the average age was 27±6 years and slightly more than 1/3 of participants (149/388, 38%) were first time mothers (Table I). Sixty-six percent (254/386) of participants were not married, 73% (282/386) were African-American, and 59% (220/374) had public health insurance. Less than 30% (114/387) had graduated college.

Seventy-nine percent (307/387) of women had attended 10 or more prenatal visits, and only 5 (1%) did not receive any prenatal care. Data were not collected about the type of prenatal care provider (obstetrician versus midwife) nor whether the provider was affiliated with the University of Chicago. Thirty-eight percent of participants thought they had received some prenatal genetic testing (146/383) and 25% (97/383) were “not sure” if they had received prenatal genetic testing. Sixty-seven percent of participants had never heard of CF (260/388), compared to 4% (16/388) having never heard of SCD ($p<0.05$). Participants who had never heard of CF were more likely to be African American, younger than 27 years old, not a college graduate, not married and not privately insured ($p<0.001$ for each demographic variable, data not shown).

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Among women who had heard of SCD or CF, more women reported having been tested for SCD (171/372, 46%) than for CF (42/128, 33%). Of the participants who had heard of SCD, 21% (79/372) of participants were aware of a family history of SCD or SCT (heterozygote for sickle cell disease) and 51% (191/372) knew someone with either SCD or SCT. Of the participants who had heard of CF, only 4% (5/128) were aware of a family history of CF or CF carrier and only 21% (27/128) knew someone with CF or a CF carrier. In total, 34 women self-identified as having SCT, 1 with SCD, and 1 as a CF carrier. Approximately one-third of the participants who were confident that “I am not a carrier” for SCD or CF report never having been tested for SCD (100/313 or 32%) or CF (36/99 or 36%) respectively.

Only 35% (133/378) of participants recalled their prenatal care provider mentioning NBS and slightly more than half (217/388, 56%) recalled NBS being mention by medical staff in the nursery. However, only 25% (97/388) of participants remembered ever having NBS explained to them. This low recall was true for both first-time mothers and women with more than one child. Sixteen women stated that they had learned about NBS from reading prenatal materials, often procured outside of the medical setting. Although 27% (104/388) of respondents thought they knew a condition included in the NBS blood spot; only 49 of the 104 (47%) correctly named one or more of the conditions. Although a few women (6/239, 3%) reported that their previous child(ren) had a positive NBS, upon further questioning, 2 of these women were referring to conditions that are *not* included in the NBS (jaundice or a heart condition), and 12 women who did not report having a child with a positive NBS, later mentioned having a child with SCT. Despite the small percentage who actually know what NBS tests for, nearly all women (321/388, 83%) think NBS is a good idea.

Average SCD knowledge score for the 96% of women who had heard of SCD was 66% and the average CF knowledge score for the 33% of women who had heard of CF was 63% (Table II). We found several important knowledge gaps. For example, many women thought that it was possible for carriers to develop the disease (60% for SCD; 52% for CF), or that carriers have a mild form of the disease (49% for SCD; 41% for CF). Similarly, the majority of women thought that a person with SCD (71%) or CF (68%) could transmit the disease to a child even if the partner was not a carrier. Most women thought that it was important to know if one was a carrier (96% for SCD; 91% for CF) because of potential health implications, but this is only true for SCT. Likewise most women thought there was no cure for either condition (78% for SCD; 80% for CF), although bone marrow transplant does exist as a cure for SCD.

In bivariate analysis, 5 demographic variables correlated with higher knowledge scores for both SCD and CF: 1) being 27 or older; 2) being married, 3) having a college degree, 4) having private insurance, and 5) being non-African American (data not shown). Demographic variables were evaluated for colinearity using linear regression. Having a college degree remained significant for both SCD (72% vs. 65%, $p<0.05$) and CF (68% vs. 55%, $p\leq0.05$). Being non-African American (70% vs. 55%, $p<0.001$) and having private insurance (68% vs. 51%, $p<0.05$) remained significant for CF knowledge but not for SCD knowledge.

There was more self-reported familiarity with SCD (3.32/5) than CF (1.97/5) (n=388, $p<0.001$), and both were perceived as being serious health conditions (4.25 for SCD and 4.17 for CF out of 5). Women who self-identified as being moderately or very familiar with SCD scored significantly better on SCD knowledge questions than those who self-identified as being less familiar (69% vs. 65%, $p<0.001$), but women who perceived SCD as serious or very serious were not more knowledgeable (67% vs. 65%). Women who self identified as being moderately or very familiar with CF and women who perceived CF as serious or very

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serious scored significantly better on CF knowledge questions (76% vs. 60% and 66% vs. 55% respectively, $p<0.001$ for both).

## DISCUSSION

NBS is one of the most successful public health programs in preventing morbidity and mortality. Although the vast majority of our participants are very supportive of NBS, parental lack of awareness and understanding regarding the importance of follow-up testing and treatment threaten its success [Arn, 2007; James and Levy, 2006; Miller et al., 1990].

We found that virtually all women in our study had heard of SCD (96%) but that only a third of women (33%) had heard of CF. This may be due in part to the high percentage (74%) of African American women in our survey. SCD is more common in persons of African ancestry (one in 400–500 African Americans) [Smith et al., 2006]. However, a study published in 2005 from St Louis by Boyd et al. [2005] found that 69 of 231 (30%) African American women contacted to participate in a survey were excluded because they had not heard of SCD, whereas virtually all of our respondents (both African American and individuals of other ethnicities) were able to complete the SCD portion of the survey. In contrast, CF is more common in non-Hispanic Caucasians (one in 3300) and less common in other ethnic communities including African Americans (one in 15,300) [National Institutes of Health, 1999]. In our survey, the women who were able to complete the CF portion of our survey were more likely to be more highly educated or non-African American. The lack of familiarity with CF in the African American community needs to be rectified in order to ensure that African American children benefit from the therapeutic advances promised by early diagnosis of CF [Grosse et al. 2006].

One reason that our participants should have heard about SCD and CF is that all but 5 had received some prenatal care. The American College of Obstetrics and Gynecology (ACOG) recommends prenatal testing for sickle cell trait in individuals of African, Southeast Asian, and Mediterranean descent [ACOG Committee of Obstetrics, 2007]. ACOG also recommends routine prenatal screening for CF in Caucasian families and that all women be informed of, if not offered, prenatal testing for CF [ACOG Committee on Genetics, 2005]. Given that less than 40% of women reported receiving any prenatal genetic testing and a quarter were unsure whether they had received prenatal genetic testing, it seems that women are not being tested, are not being offered testing, or are not aware that prenatal testing was done. ACOG policy also recommends that obstetricians provide some basic information about NBS to all pregnant women [ACOG, 2007]. However, only 35% of our participants recalled any mention of NBS prenatally despite ACOG policy recommendations. While it may be that they were informed about NBS but did not recall the discussions, our data are consistent with previous studies that find that health care professionals are not discussing NBS with pregnant or post-partum women [Campbell and Ross, 2004; Faulkner et al., 2006; Larsson and Therrell, 2002].

Obstetricians are not the only health care providers failing to discuss, or at least failing to effectively discuss, NBS. Only 56% of the women recalled mention of NBS by medical staff in the post-partum period, and very few women could correctly name a condition included in the screening program despite the importance that the American Academy of Pediatrics (AAP) places on newborn screening as integral to the medical home [AAP Newborn Screening Task Force, 2000]. Data show that pediatric providers lack knowledge of NBS or take a passive approach, assuming that the public health department will ensure follow-up [Davis et al., 2006; Desposito and Lloyd-Puryear, 2001; Gennaccaro et al., 2005; Kemper et al., 2006]. The mandatory nature of NBS allows pediatric providers to avoid meaningful discussions of what NBS entails.

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Overall knowledge of the two newborn screening conditions that we focused upon, CF and SCD, is poor. One explanation may be the lack of familiarity with both of these conditions (average scores were 3.32 for SCD and 1.97 for CF). Lack of familiarity may be explained by the lack of visibility of these conditions in the general population, [McClaren et al., 2008; Mitchell et al., 1993; Poppelaars et al., 2003; Treadwell et al., 2006; Watson et al., 1991] and the lack of public awareness of NBS programs more generally [Campbell and Ross, 2003; Davis et al., 2006; Detmar et al., 2007]. Less familiarity with CF may also be due to relative recent introduction of CF NBS in Illinois and the racial/ethnic composition of our patient population.

Despite the low familiarity with both SCD and CF, both conditions were perceived to be quite serious (SCD=4.25; CF=4.17). Gustafson et al. [2007] found that knowledge of SCD directly correlated with perceived severity of SCD. Although we did not find this correlation to be true for SCD, we did find this correlation with CF.

The most important factor to correlate with increased knowledge of SCD and CF was parental educational status. Greater efforts to promote genetic knowledge and health literacy earlier in the school years will be necessary to ensure greater understanding of NBS across all educational, socioeconomic, and ethnic communities. In addition alternatives to school-based programs should be developed including church-, clinic-, or community-based multi-media programs, [Sobel et al., 2009; Gason et al., 2004; Jeste et al., 2008]or home-based educational videos [Klandy et al., 2005; Paholpak et al., 2006; Rowley, 1989]

One limitation of our study was the racial/ethnic distribution (almost 3/4 were African American) which may have biased knowledge in favor of SCD and against CF given the relative frequency of these conditions in the African American community. The large majority of women had not heard of CF (n=260) whereas only 16 women had not heard of SCD. A second limitation is that maternal reports about what they were offered and told prenatally and postnatally is subject to recall bias. It may be that obstetricians and pediatricians discussed NBS more frequently than the women remember or that the terminology used in the clinical setting and in our survey were slightly different yielding more negative responses. We did not collect information about where or from whom participants received prenatal care, so we are not able to determine whether these providers have policies or practices in place to address NBS. We also interviewed post-partum women whenever it was convenient for them, and so in some cases their child had already had blood collected for NBS and at other times they had not. Although we did not specifically ask the women if the NBS blood spot had been collected or if they knew whether the NBS blood spot had been collected, both first-time mothers and women with previous children (who could have been aware of NBS from their earlier deliveries) expressed low awareness of NBS procedures and purpose. Our finding that limited information about NBS is communicated to pregnant and post-partum women is also consistent with other research studies that documented minimal conversations about NBS by both obstetricians and pediatricians [ACOG Committee on Genetics, 2005; Campbell and Ross, 2004; Faulkner et al., 2006; Larsson and Therrell, 2002].

## CONCLUSIONS

NBS in Illinois includes mandatory screening for SCD and CF. In a sample of predominantly African American post-partum women, we found greater familiarity with SCD compared with CF. However there are significant knowledge gaps for both conditions. Post-partum women report inadequate education about NBS, but none-the-less, they are supportive of it. There are many missed opportunities to educate mothers about NBS in general and about specific conditions included in NBS panels in both the obstetric clinics

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and the nursery. Creative educational efforts are needed to promote health literacy in mothers of all educational backgrounds.

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**Table I**

Demographics

| **Demographic*** | **n (%)** |
|---|---|
| Age (N=388) Mean: 27 ± 6 Range: 18–46 | |
| Married (N=386) | 132 (34) |
| First time mother's (N=388) | 149 (38) |
| Education (N=387) | |
| Did not graduate High School | 61 (15) |
| High School Graduate | 68 (17) |
| Some College | 144 (37) |
| College Graduate | 46 (11) |
| Graduate School/Degree | 68 (17) |
| Health Insurance (N=374) | |
| Public | 220 (41) |
| Private | 154 (57) |
| Number of Prenatal Care Visits (N=387) | |
| 0 | 5 (1) |
| 1–9 | 74 (19) |
| 10+ | 307 (79) |
| Race/Ethnicity (N=387) | |
| African American | 282 (73) |
| Asian/Pacific Islander | 16 (4) |
| Caucasian/European | 50 (13) |
| Mexican/Hispanic/Latin American | 24 (4) |
| Other/Mixed | 14 (6) |
| Religion (N=386) | |
| Christian | 274 (70) |
| Other | 29 (7) |
| None | 83 (20) |
| Has heard of … | |
| SCD (N=388) | 372 (96) |
| CF (N=388) | 128 (33) |
| Has a family history of … | |
| SCD/SCT (N=372) | 79 (21) |
| CF/CF-carrier (N=128) | 5 (4) |
| Self-Reported … | |
| SCT (N=372)^ | 35 (9) |
| CF-carrier (N=128) | 1 (<1) |
| Know someone with … | |

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| Demographic* | n (%) |
|---|---|
| SCD/SCT (N=372) | 191 (51) |
| CF/CF-carrier (N=128) | 27 (21) |

Abbreviations: CF=Cystic Fibrosis; SCD=Sickle Cell Disease; SCT=Sickle Cell Trait

*N varied from 388 due to non-responses.

^One individual had SCD.

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**Table II**

Knowledge Questions

| Question | Correct Answer | SCD N=372^ % correct (95% CI) | CF N=128^ %correct (95% CI) |
|---|---|---|---|
| [SCD/CF] can be transmitted by physical contact with an affected person. | False | 99 (97–100) | 98 (95–100) |
| [SCD/CF] is a genetic condition.* | True | 97 (95–99) | 88 (82–93) |
| It is important to know if you are a carrier for [SCD/CF] even if you don't have any symptoms, because of possible health risks associated with being a carrier.* | SDC = True<br>CF = False | 96 (94–98) | 9 (4–15) |
| There are things a person with [SCD/CF] can do to avoid some of the complications. | True | 90 (86–93) | 89 (84–95) |
| Individuals with [SCD/CF] are usually of normal intelligence. | True | 89 (85–92) | 81 (74–88) |
| Children with SCD have an increased risk of infections.<br>Individuals with CF get frequent respiratory (breathing or lung) infections. | True | 87 (83–90) | 91 (86–96) |
| Children with SCD take prophylactic (preventative) antibiotics to prevent infections.<br>Individuals with CF need to take special vitamins. | True | 87 (83–90) | 82 (75–89) |
| SCD affects different people in different ways, but almost always includes pain.<br>Individuals with CF usually have breathing problems. | True | 84 (80–87) | 80 (74–87) |
| Playing sports will worsen the symptoms of [SCD/CF].* | False | 72 (67–76) | 46 (37–55) |
| Men with [SCD/CF] often have fertility problems.* | SCD = False<br>CF = True | 68 (63–72) | 43 (34–52) |
| Women with [SCD/CF] often have fertility problems. | False | 61 (56–66) | 48 (40–57) |
| [SCD/CF] is most common in which ethnic group? (Options: African Americans, Caucasians and Equally in all races) | SCD = African American<br>CF = Caucasian | 60 (55–65) | 62 (53–70) |
| To inherit [SCD/CF], both parents must have at least one [SCD/CF] gene. | True | 60 (55–65) | 59 (50–67) |
| You can be a carrier for [SCD/CF] even if neither parent has [SCD/CF] or is a carrier. | False | 54 (49–59) | 51 (42–60) |
| People who are carriers of [SCD/CF] have a mild form of[SCD/CF]. | False | 51 (46–56) | 59 (51–68) |
| Individuals with SCD have an increased risk of stroke.<br>Individuals with CF usually have problems with hearing.* | SCD = True<br>CF = False | 47 (42–52) | 73 (66–81) |
| Individuals with [SCD/CF] often fail to grow at a normal rate. | True | 46 (41–51) | 52 (43–60) |
| Over time, people who are carriers for [SCD/CF] can develop[SCD/CF]. | False | 40 (35–45) | 48 (40–57) |
| [SCD/CF] can be inherited if one parent has [SCD/CF], even if the other parent is not a carrier and does not have the disease. | False | 29 (25–34) | 32 (24–40) |
| There is NO cure for [SCD/CF]. * | SCD = False<br>CF = True | 22 (18–26) | 80 (73–87) |
| MEAN SCORE | N/A | 66 (65–67) | 63 (60–65) |

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Abbreviations: CI=Confidence Intervals; CF=Cystic Fibrosis; SCD=Sickle Cell Disease

^N varies because questions were only asked of those participants who had heard of the condition.

*95% CI do not overlap between SCD and CF knowledge scores.