



Public Awareness of and Attitudes toward Newborn Screening

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Abstract

Objectives: To understand public awareness of newborn screening and public views on criteria needed for a disorder to be included in universal newborn screening.

Methods: Adults in waiting rooms of internal medicine and pediatric clinics completed a survey.

Results: 55% of participants (n=213) were unaware that all newborns are screened at birth for certain disorders. Nearly 60% supported screening only for disorders occurring more frequently than 1 in 10,000 births. The risk of death or long-term impairment, accuracy of screening test, frequency of disorder, availability of appropriate treatment and help with future reproductive decisions were considered "important" or "very important" while cost of the screening test, associated parental anxiety, and parental burden were not.

Conclusion: Public awareness of newborn screening program remains limited. The results of this survey suggest that public support for the addition of rare disorders to the list of newborn screening may be limited.

Keywords: Newborn screening; Neonate; Public awareness; Attitudes

Introduction

Newborn screening (NBS) is one of the most successful public health programs of the 20th-century. Since the first NS test for phenylketonuria was introduced in the 1960's, NBS has expanded significantly and now includes a wide range of conditions, including some with limited treatment options and uncertain prognoses. Inclusion of some of these recently added disorders to NBS panel does not meet some of the longstanding guiding principles defined by Wilson and Jungner in their seminal paper [1]. Wilson and Jungner emphasized that a disorder should be considered for NBS only if further diagnostic testing and management of screen positive individuals was available and if an effective treatment for confirmed cases existed. They also highlighted the importance of a well-understood natural history of the disorder when considering a disorder for NBS. However, the expansion of NBS by tandem mass spectrometry is allowing for the detection of conditions so rare that information about natural history and effective treatments is sometimes lacking. Although early diagnosis of these disorders may have certain benefits such as avoidance of a long diagnostic quest, it may also have adverse psychosocial and financial implications for the families as well as other ethical, social, and legal issues [2]. These concerns have sparked a debate in the medical community. In a commentary on the future of NBS, authors recommended the need for major efforts to educate and prepare health care professionals and the public [3].

Since parents are directly affected by NBS policies, it is important that their views are researched and considered in decision making. It is even more important to do so in view of the reports that parental opinions about NBS differ significantly from those of professionals [4-6]. In a recent study, investigators reported that parent attitudes differ from those of many professional communities with regard to timing of NBS education, informed consent, NBS for disorders that lack an effective treatment, and predictive testing of children for late-onset disorders [6].

However, to the best of our knowledge, no studies have been done to understand the preferences of US adults with regards to important thresholds, such as the frequency of included disorders and the cost of identifying one case, which are important factors when selecting disorders appropriate for universal NBS. Therefore, the purpose of this study was to understand public awareness of NBS and public views on inclusion criteria for a disorder to be considered for universal NBS.

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Table 1: Participant characteristics.

Participant Characteristics	Number (%)
Gender	
Female	163 (76.5)
Age (years)	
<25	20 (9.4)
25 – 35	95 (44.6)
>35	90 (46.0)
Education Level	
High school or less	37 (17.4)
College	123 (57.8)
Graduate school	53 (24.9)
Healthcare Professional	
Yes	55 (26.1)
Marital Status	
Married	118 (55.7)
Do you have children	
Yes	169 (79.3)

Methods

With approval from the Institutional Review Board, we recruited a convenience sample of 213 adults in the waiting rooms of pediatric and internal medicine clinics. After informed consent, participants answered questions on demographics and their understanding and attitudes toward NBS. Responses were tabulated and analyzed using a Fisher's exact test (Stata13, Stata Corp LP, 2011, College Station, TX) to evaluate significant differences ($p < 0.05$) in responses between gender, marital status, age group, level of education, occupation (health professional or non-health professional), and parental status. Logistic regression was used to estimate odds ratios (OR) for adjusted associations using Generalized Estimating Equations (GEE) for change models to account for repeated measures.

Results

Of 213 participants, 77% were female, 54% were under 35 years of age, 56% were married, and 79% had children. Eighty three percent had a college or graduate education and 26% worked in healthcare (Table 1). Forty-six percent of all participants and 40% of those not working in healthcare knew that all newborns in Illinois receive NBS and 9% of these participants answered correctly that each newborn is screened for 31 or more disorders.

Participants' views on the factors that should be considered when adding a new NBS test are summarized in Table 2. An overwhelming majority (over 85%) considered the risk of death or long-term impairment, accuracy of screening test, and the frequency of the disorder as either "important" or "very important". The availability of

appropriate treatment and the screening test being done to help with future reproductive decisions were considered as either "important" or "very important" by nearly 70% of all participants. In contrast, less than 50% of participants considered cost of the screening test, associated parental anxiety, parental burden, and potential need for transfer to another hospital as "important" or "very important" in deciding if a new screening test was appropriate for universal NBS or not (Table 2). Married participants and participants with children were more likely to support addition of a new screening test in absence of availability of treatment as compared to unmarried participants and participants with no children.

When asked "how often would the disease have to occur to support its inclusion in the NBS panel," 61% supported screening for disorders only if more than 400 affected infants are born per year in US which will be consistent with an incidence of 1:10,000 or higher; 13% supported screening for disorders with incidence of 1 in 50,000 or less (i.e. less than 80 affected infants per year in US) and 26% of participants supported screening regardless of the frequency of the disorder. Nearly 80% of participants supported screening if there was any or even a low risk of death or long term impairment from the disorder. Females were more likely to support screening if there was any or even a low risk of long-term impairment from the disorder ($p = 0.005$).

Forty five percent of participants believed that cost should not be a consideration for a screening test, while 41% reported that an acceptable total cost of identifying one newborn by universal screening should be \$10,000 or less. For nearly a third of all participants (32%), extended length of hospital stay did not matter but only 16% considered that a delay in discharge of more than 3 days would be acceptable. Thirty three percent of females responded that a delay in discharge did not matter versus 24% of males ($p=0.001$). Nearly 80% of participants responded that factors such as the need for transfer, additional blood draws, and additional imaging studies would not affect their decision to participate in the NBS program.

Discussion

A large majority of previous reports on parental attitudes toward NBS have focused on a single disorder, such as cystic fibrosis or fragile X syndrome, and most of these reports have evaluated the acceptability of a particular screening test and/or the impact of screening for a specific disorder on parents of children with that disorder. In contrast, this study sought to evaluate which factors are

Table 2: Participant views on general newborn screening.

Which of the following factors should be taken into consideration before adding a disorder to those screened for by the newborn screening program?	Neutral/not important		
	Important/very important	Neutral/not important	Not answered
Frequency of disorder	86.4%	10.3%	3.3%
Risk of death from disorder	96.2%	1.5%	2.3%
Risk of long term impairment from disorder	95.2%	2.0%	2.8%
Availability of treatment for disorder	77.2%	19.5%	3.3%
Cost of screening test	43.6%	53.1%	3.3%
Associated parental anxiety due to screening	44.4%	52.8%	2.8%
Burden on parents (longer hospital stay or more office visits)	44.9%	53.3%	2.8%
Potential need for transfer to another hospital	45.7%	52.0%	2.3%
Accuracy of screening test	93.7%	3.5%	2.8%
Potential to aid with reproductive choices for parents in the future	70.4%	26.3%	3.3%

Table 3: Public views on general newborn screening.

Authors	Country	study period	Type/setting	No. of Participants	Participant Characteristics	Primary Objective	Results
Hasegawa et al. [6]	USA	2003-2004	Focus groups	17 focus groups, 114 participants	mothers of children 10 years old or younger; 7 focus groups included only women from underrepresented populations, including non-English speaking Hispanics, African-Americans, and Asians and Pacific Islanders from rural communities	To assess parent knowledge of NBS and parent attitudes toward NBS for untreatable conditions, NBS for late-onset disorders and informed consent in NBS	Most participants did not recall receiving information about NBS, and all wanted this information prenatally. All women supported NBS for conditions that occur in infancy without a proven treatment. However, they disagreed about NBS for disorders that manifest in late childhood or adulthood.
Detmar et al. [13]	Netherlands	2005	Focus groups	7 groups, 36 participants	parents-to-be, parents of healthy children, and parents of children with disorders; 29 females and 7 males; 34 of 36 with intermediate or higher education	To investigate the preferences and views of parents and future parents with respect to information about, and consent to, neonatal screening and the possible expansion of the program	Parents were not well informed about newborn screening, only received a result if something was wrong and did not know about the possibility of refusing it. Participants wanted more information if screening program were to be expanded, and preferred an opt-out consent approach.
Quinlivan JA et al. [14]	Australia	2006	Questionnaire	232	Postpartum mothers within 24 hours of blood being taken from their baby by heel prick for the newborn screen	To evaluate new mothers' opinions of genetics and newborn screening	Supported newborn screening programs where outcomes could be used to prevent or reduce the severity of a disease but were less supportive of screening for other benefits such as future family planning. The majority felt that parental consent should be mandatory. The majority expressed concern that a child with a genetic illness would face discrimination and difficulty obtaining insurance or employment.
Plass et al. [11]	Netherlands	2007	Web based questionnaire	1392	96.5% females; 64% parents; 57% pregnant; Nearly half were urban and highly educated	To explore the opinion of parents/ future parents on addition of treatable, less treatable and untreatable disorders to the newborn screening program	The majority favored addition of a disorder regardless of the treatability of the disease; less educated were more in favor of including untreatable diseases than highly educated
Hayeems et al. [15]	Canada	2009	Focus groups	Eight focus groups; 60 participants	60% female, 87% had at least some college or university education and 43% had children.	To understand public expectations and values regarding the types of conditions that should be included in NBS and whether parents should provide consent	82% supported NBS for disorders with no treatment. 88% endorsed screening without explicit consent for treatable disorders, but 62% supported parental choice for untreatable disorders. Concern for anxiety, stigma and unwanted knowledge depended upon disease context and strength of benefits.
Lipstein et al. [12]	USA	Before 2010	Focus groups and interviews with parents of young children at pediatric primary care practices or a hospital-based genetics clinic	6 primary care focus groups, with a total of 40 participants, and 4 interviews	The majority of participants were mothers with at least a college education, and 69% of participants self-reported their race as white	To elucidate the factors parents would consider when making a decision about newborn screening, as well as the risks and benefits they perceive for such screening.	The majority favored universal screening for disorders with specific treatments, accurate tests, early onset, and well-understood natural histories. Opinions were less consistent for disorders with a later onset, and less-definitive treatments. For less-treatable conditions, parents perceived increased risk with screening, such as psychosocial impacts, including stress and worry.
Etchegary H et al. [16]	Canada	2010	Pen-and-paper survey; The majority of surveys were completed on site, whether at survey tables or in prenatal classes	648	General public and prospective parents; 74% female, 69% married, 53% had no children; 33% with university degree and 33% with trade school or college.	To measure attitudes toward newborn genetic testing	High interest in NBS for genetic disorders regardless of availability of an effective treatment. 93% of respondents wanted that informed consent should be obtained before NBS is undertaken.

Mak et al. [7]	Hong Kong	2010	Phone interview	172	Parents of newborn infants before discharge from hospital	To examine the parental knowledge and attitudes towards expanded newborn screening	87.8% had never heard of NBS; 99.4% wanted more parental education; 97.7% supported screening for incurable diseases; 93.9% accepted the possibility of false positive and false negative results; 70.4% preferred a voluntary basis; 83.2% want it to be fully government funded as basic primary care; 98.2% required pre-test counseling; and 96.4% required an explicit parental consent before blood sampling.
Miller et al. [17]	Canada	2013	Internet survey	1213	66% had children; 67% had college or higher degree. Sample was reflective of the Canadian population but was better educated and had a more narrowly distributed income.	To understand public expectations of newborn population screening	Respondents favored screening. Clinical benefits were the most important outcome; reproductive risk information and early diagnosis were also valued, and reproductive risk information was least important. All respondents preferred to avoid FP results and over-diagnosis but were willing to accept these to achieve moderate clinical benefit.

important to parent and non-parent adults for adding a disorder to the NBS program from a public policy perspective. To the best of our knowledge, this is the first study to evaluate public views on this issue in the United States.

The results of our study show that a majority of both parent and non-parent adults are not well aware of the existing NBS program which confirms the findings of previous studies that have shown that the public, and parents in particular, have very limited knowledge and awareness of NBS programs [7,8]. Fifty five percent of all participants in our study did not know that all newborns receive screening for certain metabolic and endocrine disorders before being discharged from the hospital and only very few participants knew how many disorders are included in the NBS program. It is concerning that this lack of knowledge has not improved over time and suggests that public education campaigns and other efforts may be necessary. A better public understanding of NBS is a prerequisite for more meaningful public participation in the development of new policies.

Nearly 60% of all participants in our study thought that, from a public policy perspective, screening should only be done if a disorder occurs more frequently than 1 in 10,000 births, and 42% supported screening for disorders with an incidence of more than 1 in 1,000. Currently only two disorders, newborn hearing loss and pulse-oximeter screening for critical congenital heart disease, meet a threshold of incidence of 1:1000 or more; and only three disorders, phenylketonuria, cystic fibrosis and congenital hypothyroidism, have an incidence of 1:10,000 or more. All other currently screened disorders have much lower incidences ranging from 1:50,000 to 1:500,000 [9]. We are not aware of other studies which have evaluated public views on this issue. However, the results of this study do suggest that public support for the addition of rare disorders to universal newborn screening panel may be limited.

Participants in this study were fairly evenly split on the issue of the cost of identifying one infant. Although 45% of the participants reported that cost should not be a consideration, nearly 41% considered a cost of \$10,000 or less as acceptable to identify one infant which is significantly less than the current cost of diagnosing one infant for most disorders in the NBS program. In a recent review, it was estimated that the cumulative cost of diagnosing one infant with a metabolic disorder in California was \$68,750 [10]. Although the overall cost and benefit analysis rather than the basic

cost of identifying a case is more informative and helpful for making policy decisions, these analyses are challenging to perform and vary from disease to disease and from country to country. The factors influencing these analyses include frequency of disorder, cost and accuracy of screening test, morbidities associated with the disorder, and availability and cost of treatment among others. In addition, it is also important to recognize that there is only limited consensus on how to interpret this information. The acceptable cost of early diagnosis not only depends on available resources but also from whose perspective it is being looked at such as from the perspective of a parent, healthcare provider, payer or the society at large.

Several studies from different countries have reported parents' and prospective parents' views on expansion of NBS to include untreatable disorders and other genetic testing [6,7,11-17]. Table 3 summarizes the findings of these studies. Similar to other studies, participants in our study considered the risk of death and/or long-term impairment as the most important factor for inclusion of a disorder in the NBS panel. Furthermore, a large majority of participants in our study considered the availability of treatment and the accuracy of the screening test as very important. In contrast, a cross-sectional survey of Chinese parents from Hong Kong reported 98% support for screening of even incurable disorders [7]. Another survey from the Netherlands reported that 88% of respondents supported screening for less treatable disorders and 73% supported screening for untreatable disorders [11]. Respondents who already had children at the time of completing the questionnaire were even more in favor of screening for especially untreatable disorders. However, the fact that 96.5% of all respondents in this Dutch study were female and that more than half were pregnant at the time of survey may have influenced these findings [11]. Some of these seemingly cultural differences in support for NBS for less treatable and incurable disorders could be related to the perceived financial burden on parents in different societies. This speculation is supported by the results of another US study with findings very similar to ours. Based on interviews with focus groups and parents of young children, this study reported less parental support for screening of disorders with less-definitive treatments and less-accurate screening tests [12]. These findings may suggest limited public support for adding rare metabolic disorders to the universal newborn screening panel in the United States. Our findings and those of other researchers suggest that factors which parents consider important in decision-making

can differ from those that are important to clinicians and policy makers and can vary from country to country.

Increase in parental stress and anxiety has been cited as an important barrier in other NBS programs and the greatest periods of stress and anxiety are reported to be between the positive screening test and the confirmatory test [18-20]. However, several investigators have reported that the concerns and anxiety related to false-positive tests persist long after a negative confirmatory test [21-24]. One study found that children with false-positive NBS results compared with children with normal results were twice as likely to experience hospitalization in the first 6 months of life and mothers of children in the false-positive group compared with mothers of children with normal screening results attained significantly higher scores on the Parental Stress Index and the Parent-Child Dysfunction subscale [24]. Potential negative psychosocial and financial effects of pre-symptomatic diagnoses due to mandated screening have been reported by others. Lipstein et al. [12] reported that although parents strongly supported population-wide screening for disorders even if the treatments had significant morbidity, the most emphasized risks were psychosocial impacts, including stress and worry. The concerns about anxiety and parental burden were important to less than half of all participants in our study. It is also encouraging to note that studies have reported that the distress and anxiety felt by most parents of children with false-positive results usually resolve quickly after diagnostic testing is completed [7,25-27] and parental stress can be reduced further by providing better information and communication [24].

The strengths of our study include a reasonable sample size and diversity of participants in terms of gender, age, parental status, and profession (healthcare versus non-healthcare). The limitations of our study include the use of a convenience sample, the inherent limitations of data collected by a structured questionnaire and the limitations of any qualitative study. Eighty three percent of all participants had a college or graduate education and 26% worked in healthcare. Due to these limitations, we may have missed important views of the participants on issues not included in the questionnaire, and it may be inappropriate to use these findings while making decisions for a different population. It can also be speculated that given the complexity of these issues, survey participants did not have enough background information to make correct choices. This survey was only offered in English and may not represent the views of non-English speaking communities.

In conclusion, the results of this survey suggest that public awareness of NBS program remains limited and it may be important to obtain public views before future expansions of the universal NBS panel. Future studies with larger and more diverse sample sizes are necessary to fully understand public views on this very important public program.

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