Pediatricians’ Attitudes Toward Expanding Newborn Screening

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ABSTRACT. Objective. Traditional population screening focuses on conditions for which early treatment prevents severe morbidity and mortality. The classic example in pediatrics is newborn screening for phenylketonuria, which began in the 1960s. In 1968, Wilson and Jungner delineated 10 criteria that would justify population screening. These criteria have been reaffirmed by many newborn screening task forces as the standard for adding conditions to newborn screening programs. Today, however, some newborn screening programs are expanding to include conditions that may not meet all of the traditional screening criteria. Little is known about pediatricians’ attitudes toward expanding screening. We examine the attitudes of pediatricians and pediatric subspecialists toward screening for cystic fibrosis (CF), Duchenne muscular dystrophy (DMD), fragile X, and type 1 diabetes.

Methods. A cross-sectional survey was conducted of 600 pediatricians, including those who are members of the section of genetics, endocrinology, pulmonology, and neurology of the American Academy of Pediatrics. For each condition, pediatricians were queried about (1) testing high-risk infants, (2) newborn screening, and (3) population screening or testing beyond the newborn period. Demographic data were also collected.

Results. A total of 232 (43%) of 537 eligible pediatricians returned surveys. More than 75% support testing high-risk infants for all conditions except type 1 diabetes. CF was the only condition for which >50% supported newborn screening. Newborn screening was preferred over screening older infants for all conditions except fragile X. Subspecialty affiliation did not have a significant impact with respect to attitudes about testing high-risk children, newborn screening, or screening beyond infancy. We analyzed the data by the number of patients with the queried condition under the physician’s care and by the number of affected family members. Neither aspect was significant. We also analyzed the data by gender, by year of residency graduation, and by geographic location. None of these factors revealed significant differences in responses. For each condition, 8% to 41% of physicians would personally choose to test their own infant. We found that physicians’ opinion about what they would want for their own children correlated with their attitude about population newborn screening. Those who would personally choose testing of their own infants were highly likely to support newborn screening for CF (98%), DMD (94%), and fragile X (98%), but only 78% of those who would personally opt for newborn screening of type 1 diabetes would also endorse population-based screening. This was statistically significant for each condition. Those who would choose not to test their own infants were significantly less likely to support newborn screening of the general population. One third of those who did not want to test their own newborns for CF supported population screening, whereas only one fifth supported DMD and fragile X population screening. For type 1 diabetes, 98% of those who would not personally choose newborn testing did not want it offered as a population screening program.

Conclusions. Most physicians support diagnostic genetic testing of high-risk children but are less supportive of expanding newborn screening, particularly for conditions that do not meet the Wilson and Jungner criteria. Willingness to expand newborn screening does not correlate with professional characteristics but rather with personal interest in testing of their own children. Pediatrics 2005;116:e476-e484. URL: www.pediatrics.org/cgi/doi/10.1542/peds.2005-0453; newborn screening, cystic fibrosis, Duchenne muscular dystrophy, type 1 diabetes, fragile X, attitude, beliefs, genetic testing.

ABBREVIATIONS. PKU, phenylketonuria; DMD, Duchenne muscular dystrophy; CF, cystic fibrosis; CDC, Centers for Disease Control and Prevention; AAP, American Academy of Pediatrics.

Traditional population screening focuses on conditions for which early treatment prevents severe morbidity and mortality. The classic example in pediatrics is newborn screening for phenylketonuria (PKU), which began in the 1960s. Quickly, PKU screening became mandatory in many US states. Today, newborn screening programs are judged by whether they fulfill the screening criteria described by Wilson and Jungner in 1968 and reaffirmed by many newborn screening task forces. However, there are those who argue to expand newborn screening in the United States even if the programs do not meet all of the traditional screening criteria. Numerous states have also added pilot projects or experimental research projects to their newborn screening programs. Most pilot projects in the United States and other countries currently focus on conditions that can be detected using tandem mass spectrometry. There have been numerous pilot studies in the United States and other countries for screening for conditions for which early treatment does not exist (eg,
Duchenne muscular dystrophy [DMD] and type 1 diabetes.

The decision to piggy-back pilot projects and experimental research projects onto newborn screening programs is controversial.23,24 Because newborn screening programs are mandatory and often performed without parental consent, it is not clear that parents understand either that the additional tests are voluntary or that there is a difference in purpose between the 2 types of tests. Few programs, however, have attempted to procure a separate blood sample at a separate time because of cost, time, and lower sample yield.25,26

Although some research has been done to understand the attitudes of parents who have been asked to consent to these newborn pilot and research projects,27–33 little is known about the attitudes of pediatricians.34,35 This research examines pediatricians’ attitudes about testing and screening for 4 conditions (cystic fibrosis [CF], DMD, type 1 diabetes, and fragile X) in 3 different scenarios: testing a high-risk child as defined by a positive family history, newborn screening, and population screening beyond the newborn period. We chose to examine attitudes about screening in the newborn period and beyond because of ethical concerns raised about research in the newborn period, which may be a particularly vulnerable time for parents and children.24,36,37 We chose these 4 conditions because they are relatively common for genetic conditions and they present a diverse set of issues regarding availability and feasibility of screening methods, disease severity, and availability of treatment. All have been considered or are being studied for possible inclusion in a newborn screening program at some time. CF is currently screened for in 11 states and numerous Western countries.38 The Centers for Disease Control (CDC) held a conference in November 2003 in which the conclusion was to support newborn screening as justified but not to “suggest that states not doing it are below the standard of care; the evidence is not sufficiently overwhelming to suggest that.”39,40 After completion of our survey, CF was more vigorously recommended by the American College of Medical Genetics.41 We chose to include DMD because a CDC conference in March 2004 was convened to examine newborn screening for DMD.42 The CDC is currently funding 2 pilot screening studies, 1 in newborns and 1 at 6 months of age. We chose to include type 1 diabetes because 2 research pilot studies are on-going in the United States: Diabetes Autoimmunity Study in the Young (DAISY) in Colorado43,44 and Prospective Assessment in Newborns for Diabetes Autoantibodies (PANDA) in Florida.45 We chose to include fragile X because of interest within the fragile X community to develop newborn screening programs.46

We hypothesized that pediatricians would be supportive of predictive genetic testing of newborns for all of these conditions in high-risk families. We hypothesized that pediatricians who were members of the American Academy of Pediatrics (AAP) section of genetics would be most favorable to population screening for each condition. We also hypothesized that clinicians would be more supportive of population screening for diseases within their own subspecialty. That is, we hypothesized that pediatricians who were members of the AAP section of pulmonology would be more favorable to predictive genetic screening for CF than other pediatricians, that pediatricians in the AAP section of neurology would be more favorable to predictive genetic screening for DMD and fragile X than other pediatricians, and that pediatricians in the AAP section of endocrinology would be more favorable to predictive genetic screening for type 1 diabetes than other pediatricians. We also hypothesized that the subspecialists would be more accepting of a second blood spot in part because of their potential interest in early diagnosis and in part because they would not be burdened with the difficulties in its procurement.

METHODS

We randomly sampled 600 pediatricians listed in the 2004 edition of the AAP web-based directory. We excluded physicians who did not reside in the United States, were members of the section of retired physicians, or did not have an e-mail address listed in the AAP directory. Physicians were also excluded when they practiced in the states of Maryland, Wyoming, or the District of Columbia because these states require consent, and we wanted to focus our attention on the responses of physicians who practice in states in which newborn screening programs are mandatory. Pediatricians may electively belong to specialty pediatric subsections. We randomly chose 100 pediatricians from each of 4 sections (pulmonology, neurology, endocrinology, and genetics), as well as 200 physicians who are AAP members but are not affiliated with any of the aforementioned subsections.

We were interested in the attitudes of physicians regarding newborn testing and screening for CF, DMD, fragile X, and type 1 diabetes. In each of the 4 scenarios, we provided a description of the condition and an overview of the treatment alternatives that are available to children who test positive, regardless of whether the children are symptomatic. For CF, the physicians were told that the data clearly show a nutritional benefit for early diagnosis, although the pulmonary benefits remain controversial. For DMD, the physicians were told that steroids may prolong ambulation but do not prolong life expectancy. For fragile X, the physicians were told that intensive therapy begun early might be useful but it is still unproved. For type 1 diabetes, the physicians were told that the genetic information is not 100% predictive but would be used to determine who was at higher risk (but <10% lifetime risk) and that they would be offered close follow-up for autoantibody development. They were told that no treatment is available at this time to retard the disease or prevent it from progressing. For each condition, the questions attempted to elicit the physicians’ attitude about the following: (1) testing of high-risk infants, (2) newborn screening done with the same blood spot collected for the state-mandated newborn screen, and (3) screening of infants at a later date (3–9 months of age) in combination with other blood tests that may be done (eg, lead screening). For screening at a later date, physicians could choose universal population screening or voluntary testing. For statistical analysis, we grouped all physicians in favor of screening together, whether voluntary or universal. Physicians were also offered the choice of screening only boys for the X-linked conditions of fragile X and DMD, although for the purpose of statistical analysis, we grouped together those who supported screening all children and only boys. For statistical analysis, we also excluded responses that were left blank or marked as not sure. We also queried the physicians about whether they would want newborn testing for their own children. Spaces were provided after each question for comments. All comments were coded for themes by both K.A. and L.F.R., and disagreements were resolved by discussion. Demographic data were collected. A copy of the survey can be found in Appendix A. Each physician was contacted a maximum of 3 times, by e-mail, fax, or postal mail. Approval from the University of Chicago Institutional Review Board for the project and for waived written consent were
RESULTS

A total of 600 surveys were distributed. Forty-eight physicians were excluded because they could not be located, and 15 excluded themselves because they either were not practicing or were surgeons. Of the remaining 537 respondents, 232 (43%) returned complete or partial responses, 43 (8%) refused, and 262 (49%) did not respond.

Almost two thirds of the respondents were men (Table 1). A majority had completed subspecialty training. Seventy percent of responses came from physicians who graduated from residency before 1989. The respondents were not distributed equally by specialty. More members of the section of genetics responded than other sections. Undifferentiated members and members of the section of pulmonology were least likely to respond (36% and 35%, respectively). Respondents who were not members of the 4 subsections were geographically diverse. Forty-two percent provided at least some subspecialty care as part of their clinical practice. For each condition, ~40% of respondents stated that they had 4 or more affected patients in their practice. In contrast, very few respondents (<2.5%) had any family members with any of these conditions except for type 1 diabetes, for which almost 10% stated that they had an affected family member.

In Table 2, we describe the responses of the physicians to the following questions: (1) Do you support testing high-risk children (based on family history) (columns 1–3)? (2) Do you support newborn screening (columns 4–6)? (3) Do you support screening infants at a later date (columns 7–9)? There was overwhelming support for testing high-risk children for all of the conditions except type 1 diabetes. In contrast to the near-universal support for testing children who are at risk for CF (94%), only 34% supported high-risk testing for type 1 diabetes. Overall, there was less support for newborn screening than for targeted screening of high-risk children. Nevertheless, more than half of respondents did support newborn screening for CF. Fewer than one third of respondents favored newborn screening for fragile X (32%), DMD (26%), and type 1 diabetes (9%). The majority of respondents did not favor screening for any of these conditions at a later date, although there was still greater support for CF screening compared with the other conditions. For each condition, >80% of physicians who supported screening at a later date believed that it should be voluntary.

DMD and fragile X are X-linked conditions. DMD is almost exclusively expressed in boys. Fragile X is expressed more frequently and often more severely in boys. Nevertheless, among those who favored testing for these conditions, the majority wanted testing of both boys and girls (Table 3). This was particularly acute in fragile X, for which >80% supported testing of both genders in each scenario. We analyzed the data from each scenario by comparing the responses of the geneticists with all other pediatricians. We excluded from data analysis the not sure/not answered (between 4% and 19% of all responses for any question). For CF, we also compared the responses of the members of the section of pulmonology versus all other pediatricians, and members of the sections of pulmonology and genetics combined versus all other pediatricians. For DMD and fragile X, we compared the responses of the members of the section of neurology versus all other pediatricians, and members of the sections of neurology and genetics combined versus all other pediatricians. For type 1 diabetes, we compared the responses of members of the section of endocrinology versus all other pediatricians, and members of the sections of endocrinology and genetics combined versus all other pediatricians. Subspecialty affiliation did not have a significant impact with respect to attitudes about testing high-risk children, newborn screening, or screening beyond infancy. We analyzed the data by the number of patients with the queried condition under the physician’s care and by the number of affected family members. Neither aspect was significant. We also analyzed the data by gender, by year of residency graduation, and by geographic location. None of these factors revealed significant differences in responses.

Physicians were also queried about their attitude for testing their own children (Table 4). When presented with the option of testing their own newborn, only 8% wanted to test for type 1 diabetes, 16% for DMD, 18% for fragile X, and 41% for CF. In Table 5, we examined whether physicians’ opinion about what they would want for their own children correlated with their attitude about population newborn screening. Those who would personally choose testing of their own infants were highly likely to support newborn screening for CF (98%), DMD (94%), and fragile X (98%), but only 78% of those who would personally opt for newborn screening of type 1 diabetes would also endorse population-based screen-

TABLE 1. Demographics

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>n (%) (N = 232)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>145 (63)</td>
</tr>
<tr>
<td>Female</td>
<td>87 (38)</td>
</tr>
<tr>
<td>Year residency completed</td>
<td></td>
</tr>
<tr>
<td>Before 1989</td>
<td>161 (70)</td>
</tr>
<tr>
<td>1990 to present</td>
<td>68 (30)</td>
</tr>
<tr>
<td>Fellowship training</td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>182 (81)</td>
</tr>
<tr>
<td>No</td>
<td>44 (19)</td>
</tr>
<tr>
<td>Region where practicing</td>
<td></td>
</tr>
<tr>
<td>Northeast</td>
<td>51 (23)</td>
</tr>
<tr>
<td>Midwest</td>
<td>55 (24)</td>
</tr>
<tr>
<td>South</td>
<td>63 (29)</td>
</tr>
<tr>
<td>West</td>
<td>52 (24)</td>
</tr>
<tr>
<td>AAP section response rate</td>
<td></td>
</tr>
<tr>
<td>Genetics (n = 92)</td>
<td>55 (60)</td>
</tr>
<tr>
<td>Endocrinology (n = 92)</td>
<td>38 (41)</td>
</tr>
<tr>
<td>Pulmonary (n = 99)</td>
<td>34 (35)</td>
</tr>
<tr>
<td>Neurology (n = 84)</td>
<td>43 (51)</td>
</tr>
<tr>
<td>Undifferentated (n = 171)</td>
<td>62 (36)</td>
</tr>
</tbody>
</table>

* Percentages do not equal 100% because of rounding.
TABLE 2. Physician Attitude Regarding Testing and Screening Children for Each of 4 Conditions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Testing High-Risk Children, n (%)</th>
<th>Newborn Screening, n (%)</th>
<th>Testing/Screening Infants at a Later Date, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Yes (n)</td>
<td>No (n)</td>
<td>No Answer/Not Sure (n)</td>
</tr>
<tr>
<td>CF</td>
<td>218 (94)</td>
<td>5 (2)</td>
<td>9 (4)</td>
</tr>
<tr>
<td>DMD</td>
<td>179 (77)</td>
<td>28 (12)</td>
<td>25 (11)</td>
</tr>
<tr>
<td>Fragile X</td>
<td>178 (77)</td>
<td>39 (17)</td>
<td>15 (6)</td>
</tr>
<tr>
<td>Type 1 diabetes*</td>
<td>78 (34)</td>
<td>122 (53)</td>
<td>32 (14)</td>
</tr>
</tbody>
</table>

* Percentages do not equal 100% because of rounding.

TABLE 3. Whether Physicians Who Support Screening for X-Linked Conditions Would Limit Such Screening to Boys

<table>
<thead>
<tr>
<th>Do You Support…?</th>
<th>Testing High-Risk Infants, n (%)</th>
<th>Newborn Screening, n (%)</th>
<th>Testing/Screening Infants at a Later Date, n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Boys only (n = 179)</td>
<td>Boys and girls (n = 178)</td>
<td>Boys only (n = 60)</td>
</tr>
<tr>
<td></td>
<td>DMD (n = 60)</td>
<td>Fragile X (n = 73)</td>
<td>DMD (n = 60)</td>
</tr>
<tr>
<td>Yes</td>
<td>88 (49)</td>
<td>35 (20)</td>
<td>27 (45)</td>
</tr>
<tr>
<td>No</td>
<td>91 (51)</td>
<td>143 (80)</td>
<td>33 (55)</td>
</tr>
</tbody>
</table>

TABLE 4. Physicians’ Interest in Testing Their Own Children

<table>
<thead>
<tr>
<th>Condition</th>
<th>Would You Choose Testing for Your Own Child? n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Yes (n)</td>
</tr>
<tr>
<td>CF</td>
<td>96 (41)</td>
</tr>
<tr>
<td>DMD</td>
<td>38 (16)</td>
</tr>
<tr>
<td>Fragile X</td>
<td>42 (18)</td>
</tr>
<tr>
<td>Type 1 diabetes</td>
<td>18 (8)</td>
</tr>
</tbody>
</table>

DISCUSSION

When Guthrie sought state-wide screening in the early 1960s, many pediatricians did not support it.2,3 Some of the concerns focused on whether the bacterial inhibition assay was a sensitive and specific test, some were concerned whether the natural history of the disease was well understood, and some were concerned whether proper dietary management was known.2,3 It is also clear that opposition stemmed from pediatricians’ reluctance to be told how to practice medicine.2,3 Today, newborn screening for PKU is well established as a public health measure, and decisions about whom to test and for what to test are made at the state level, although there is growing interest in federal input.6,41,47 There is growing recognition of the important role that pediatricians should play in the newborn screening system as the providers in the medical home.6,48 This role may increase in importance if parents are given greater decision-making authority regarding expanded screening.

Our data show that pediatricians support testing of children for high-risk conditions when the test confirms a clinical diagnosis (CF, DMD, and fragile X). When testing determines only increased predispositional risk, as in type 1 diabetes, most physicians would not support newborn testing of children from high-risk families.

Although pediatricians are less likely to support universal newborn screening programs than testing of high-risk children for all 4 conditions, more than half supported newborn screening for CF. Support for screening for CF beyond infancy was lower because physicians believed that if one were to screen for CF, then it made economic sense and allowed for earlier treatment in the newborn period. Given that CF is already being screened for in newborns in >12 states, it would be useful to know whether experience with screening increases or decreases physician support of screening.

Physicians were equally or more willing to sup-
port screening infants at a later date for DMD, fragile X, and type 1 diabetes than they were for screening for these conditions in the newborn period. Nevertheless, most wanted screening in later infancy to be voluntary and to require informed consent. Support for screening at a later date has the advantage of distinguishing these screening programs from the state-mandated newborn screening programs, which may make it easier for parents to understand that these programs provide information about which reasonable people disagree as to their utility. It has logistic disadvantages because of disparate access to primary care.

Although DMD and fragile X are X-linked conditions and, therefore, boys are much more likely to be symptomatic, physicians often supported screening both boys and girls, especially for fragile X, for which >80% supported screening both boys and girls. Reasons given to support screening of both genders included (1) a not insignificant number of girls who carry the mutation or permutation may be symptomatic as a result of Lyonization and (2) early intervention could be helpful and was unlikely to be harmful even if the girls were not affected. The physicians were also supportive of testing girls to diagnose carriers. Interest in screening girls for DMD was lower (between 50% and 60%) because its main value is diagnosing carriers and no proven treatment is available. Although reproductive planning (for either the parents or the girl herself as an adult woman) has not traditionally been considered a valid reason for screening newborns,2,5 it was considered a valid and valuable reason for screening for both conditions by our respondents. This is also a justification provided by the larger community.19,49–52

In general, the majority of pediatricians did not want to test their own infants for any of these conditions, although they were more closely divided on screening for CF (41% in favor). Pediatricians were more likely to support screening in the general population than they were interested in testing their own children. This is consistent with other studies that have found that pediatricians are often willing to accede to parental requests to test their children for a myriad of genetic conditions,34,53,54 although they are less interested in testing their own children.54 Our question was slightly different in that we were asking whether they supported population screening, not whether they would respect a parental request for testing for a particular condition.

We were surprised that there was no correlation between support for screening and subspecialty affiliation, gender, or year of residency graduation. We thought that younger physicians would have greater knowledge of genetics and therefore would be more supportive of screening. We were also surprised that professional experience and familial exposure did not correlate with support for screening. However, the number of patients in practice is a single measure of a physician’s professional experience, and we may not have captured this.

The only significant correlation that we found was that physicians who expressed interest in testing their own child were more likely to support newborn population screening than were physicians who would not want to test their own child. The overwhelming willingness to provide families with testing when the physicians would test their own child could signify a justice concern (if I can have it, so should my patients) or a conflation between the professional and the personal (I want it and can get it; therefore, my patients should want it and have it). Other studies have also found that personal characteristics are more predictive of physician behavior than professional characteristics.55–59 Most pediatricians who did not want to test their own child were not supportive of population screening, although more than one third of pediatricians who did not want to test their own infant for CF would support newborn screening of their patients for CF. Approximately 15% supported screening children for DMD and fragile X, although they did not want their own children tested. Again, other studies have shown that physicians are able to distinguish between what they would want for themselves and how they would

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**TABLE 5.** Whether Physicians’ Interest in Testing Their Own Children Influences Their Attitude About Universal Newborn Screening

<table>
<thead>
<tr>
<th>Would You Test Your Newborn for…?</th>
<th>Do You Support Newborn Screening for…?</th>
</tr>
</thead>
<tbody>
<tr>
<td>CF, n (%)*</td>
<td>DMD, n (%)*</td>
</tr>
<tr>
<td>CF</td>
<td>Fragile X, n (%)*</td>
</tr>
<tr>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>88 (98)</td>
<td>39 (98)</td>
</tr>
<tr>
<td>No</td>
<td>2 (2)</td>
</tr>
<tr>
<td>DMD</td>
<td>No</td>
</tr>
<tr>
<td>Yes</td>
<td>30 (94)</td>
</tr>
<tr>
<td>20 (13)</td>
<td>1 (3)</td>
</tr>
<tr>
<td>No</td>
<td>62 (66)</td>
</tr>
<tr>
<td>Fragile X</td>
<td></td>
</tr>
<tr>
<td>Yes†</td>
<td></td>
</tr>
<tr>
<td>32 (34)</td>
<td></td>
</tr>
<tr>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* The total number of respondents who answered yes or no differed by disease. P < .001.
† Percentages do not equal 100% because of rounding.
treat their patients. Nevertheless, the respondents were dogmatic against screening other children for type 1 diabetes when they did not want to test their own children (98% against). This could mean that there was at least some tolerance for parental autonomy when the benefits and risks are ambiguous and entail mainly psychologic risks and benefits. Alternatively, it could suggest a conflation between the professional and the personal (I do not want it and will not get it; therefore, my patients should not want it and do not need it).

There are several limitations to our study. First, we had a 43% response rate, which is excellent for a survey of physicians, but it is impossible to know whether responders and nonresponders have similar attitudes. Second, we had a differential response between members of different groups of specialists, which is not surprising given the issues described on the survey. However, this raises the question of whether intergroup differences may be masked by differential response rates. Third, the undifferentiated AAP members were selected as a surrogate for general pediatricians. Because 42% of this group provided at least some subspecialty care, it did not truly represent a general pediatric sample. As such, this group is less affected by the procedural aspects of later testing and may have been more supportive than general pediatricians. Fourth, in the first version of the survey that was distributed, we asked respondents whether they supported newborn genetic screening for DMD. Although genetic testing is possible, it is much costlier than newborn screening for DMD using elevated creatinine kinase. Several respondents corrected this, but it is unclear whether some physicians were rejecting newborn screening for DMD because of the test method or because they were against newborn screening for DMD by any method.

CONCLUSIONS

Physicians support testing of high-risk children for CF, DMD, and fragile X but not type 1 diabetes. They are less supportive of expanding newborn screening to these conditions with particular reluctance when testing is for predispositions and not diagnosis (compare type 1 diabetes with CF, DMD, and fragile X). Although they are concerned about the nature of treatments and costs of screening, willingness to expand newborn screening does not correlate with professional characteristics but rather with personal interest in testing of their own children.

ACKNOWLEDGMENTS

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APPENDIX: SURVEY

Condition 1: CF is an autosomal recessive condition that presents with pulmonary disease and pancreatic insufficiency. It is more common in Caucasians with an incidence of 1 in 2500 births (boys and girls are equally likely to be affected). Most children are diagnosed symptomatically in the first 4 years of life. Vitamin supplementation is recommended after early diagnosis for pancreatic insufficiency, and it has been shown to improve height and weight. Early pulmonary treatment has more equivocal results. Whether early diagnosis will change long-term outcomes is still unknown.

1. Do you support testing for CF in an asymptomatic child from a “high-risk” family (parents are both known carriers) in the first 6 weeks of life? (mark your response)
   a. Yes
   b. No
   c. Not sure
   Feel free to explain:

2. Do you support including testing for CF in the mandatory universal newborn screening program? (mark your response)
   a. Yes
   b. No
   c. Not sure
   Feel free to explain:

3. Do you support population screening for CF at a later time (eg, at the 1-month or 6-month check-up) with a separate sample? (mark your response)
   a. Yes, voluntary
   b. Yes, universal and mandatory
   c. No
   d. Not sure
   Feel free to explain:

Condition 2: DMD is an X-linked condition with complete penetrance of neuromuscular degeneration with onset in early childhood; requiring a wheelchair by adolescence and death in early adulthood. The incidence is 1 in 3500 male births. Sometimes, carrier girls may be symptomatic. Some clinicians treat patients with DMD with steroids to retard the rate of muscular weakness.

4. Do you support testing for DMD of an asymptomatic child from a “high-risk” family (mother is a known carrier) in the first 6 weeks of life? (mark your response)
   a. Yes, only boys
   b. Yes, boys and girls
   c. No
   d. Not sure
   Feel free to explain:

5. Do you support including testing for DMD in the mandatory universal newborn screening program? (mark your response)
   a. Yes, only boys
   b. Yes, boys and girls
   c. No
   d. Not sure
   Feel free to explain:

6. Do you support population screening for DMD at a later time (eg, at the 1-month or 6-month check-up) with a separate sample? (mark your response)
   a. Yes, voluntary, only boys
   b. Yes, voluntary, boys and girls
   c. Yes, universal and mandatory, only boys
   d. Yes, universal and mandatory, boys and girls
   e. No
   f. Not sure
   Feel free to explain:

Condition 3: Fragile X (X-linked condition) is the most common hereditary cause of mental retardation. Boys are more frequently affected than girls. Its frequency has been estimated to be ~1 per 2000 males and 1 per 5000 females, and there is a wide range of degrees of mental retardation. Early diagnosis can lead to early enrollment in early intervention development programs.

7. Do you support testing for fragile X of an asymptomatic child from a “high-risk” family (mother is a known carrier) in the first 6 weeks of life? (mark your response)
   a. Yes, only boys
   b. Yes, boys and girls
   c. No
   d. Not sure
   Feel free to explain:

8. Do you support including testing for fragile X in the mandatory universal newborn screening program? (mark your response)
   a. Yes, only boys
   b. Yes, boys and girls
   c. No
   d. Not sure
   Feel free to explain:

9. Do you support population screening for fragile X at a later time (eg, at the 1-month or 6-month check-up) with a separate sample? (mark your response)
   a. Yes, voluntary, only boys
   b. Yes, voluntary, boys and girls

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Condition 4: Type 1 diabetes occurs in 1 in 300 children (boys and girls) before the age of 18. It can present in an acute life-threatening fashion (diabetic ketoacidosis). Type 1 diabetes is known to run in families, and there are certain genetic alleles that are known to place the child at increased risk and others that are protective. Still the majority of children with the high-risk genotypes do not develop type 1 diabetes, and some children with the protective alleles do develop type 1 diabetes. Currently, there are no preventive treatments. Researchers are interested in following children with high-risk genotypes for the development of autoantibodies and whether environmental factors may trigger the onset of disease.

10. Do you support genetic testing for type 1 diabetes of an asymptomatic child from a “high-risk” family (a first-degree relative has type 1 diabetes) in the first 6 weeks of life? (mark your response)
   a. Yes
   b. No
   c. Not sure
   Feel free to explain:

11. Do you support including genetic testing for type 1 diabetes in the mandatory universal newborn screening program? (mark your response)
   a. Yes
   b. No
   c. Not sure
   Feel free to explain:

12. Do you support population screening for type 1 diabetes at a later time (eg, at the 1-month or 6-month check-up) with a separate sample? (mark your response)
   a. Yes voluntary
   b. Yes, universal and mandatory
   c. No
   d. Not sure
   Feel free to explain:

Demographics
1. Gender M — F —
2. Year completed pediatric residency —
3. State(s) in which you currently practice —
4. Fellowship? no — yes —
   a. If yes, please elaborate (eg, pediatric neurology) —
5. Type of practice (mark all that apply)
   a. General pediatrics
   b. Pediatric neurology
   c. Pediatric pulmonology
   d. Developmental and behavioral pediatrics
   e. Pediatric genetics
   f. Pediatric endocrinology
   g. Other (please specify)
6. Does your practice include >4 patients with any of the conditions described in this survey? (circle all that apply)
   a. CF
   b. DMD
   c. Fragile X
   d. Type 1 diabetes
7. Do you have any family members (first- or second-degree relatives) who have any of the conditions described in this survey? (circle all that apply)
   a. CF
   b. DMD
   c. Fragile X
   d. Type 1 diabetes
8. If you (or partner) were going to give birth in the next 3 months, would you request newborn screening for CF?
   a. Yes, already part of newborn screening in my state
   b. Yes, not currently part of newborn screening in my state
   c. Yes, not sure if part of newborn screening in my state
   d. No
   e. Not sure
9. If you (or partner) were going to give birth in the next 3 months, would you request newborn screening for DMD?
   a. Yes
   b. No
   c. Not sure
10. If you (or partner) were going to give birth in the next 3 months, would you request newborn screening for Fragile X?
    a. Yes
    b. No
    c. Not sure
11. If you (or partner) were going to give birth in the next 3 months, would you request newborn screening for type 1 diabetes?
    a. Yes
    b. No
    c. Not sure
12. If you (or partner) were going to give birth in the next 3 months, would you request newborn screening for type 1 diabetes?
Pediatricians' Attitudes Toward Expanding Newborn Screening
Kruti Acharya, Paul D. Ackerman and Lainie Friedman Ross
Pediatrics 2005;116;e476
DOI: 10.1542/peds.2005-0453

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