

# Design and Evaluation of a Decision Aid for Inviting Parents to Participate in a Fragile X Newborn Screening Pilot Study

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**Abstract** The major objectives of this project were to develop and evaluate a brochure to help parents make an informed decision about participation in a fragile X newborn screening study. We used an iterative development process that drew on principles of Informed Decision Making (IDM), stakeholder input, design expertise, and expert evaluation. A simulation study with 118 women examined response to the brochure. An independent review rated the brochure high on informational content, guidance, and values. Mothers took an average of

6.5 min to read it and scored an average of 91.1 % correct on a knowledge test. Most women rated the brochure as high quality and trustworthy. When asked to make a hypothetical decision about study participation, 61.9 % would agree to screening. Structural equation modeling showed that agreement to screening and decisional confidence were associated with perceived quality and trust in the brochure. Minority and white mothers did not differ in perceptions of quality or trust. We demonstrate the application of IDM in developing a study brochure. The brochure was highly rated by experts and consumers, met high standards for IDM, and achieved stated goals in a simulation study. The IDM provides a model for consent in research disclosing complicated genetic information of uncertain value.

**Keywords** Newborn screening · Informed decision making · Decision aids

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## Introduction

Newborn screening (NBS) historically has been a mandatory public health program, on the assumption that some health conditions are so serious and require such urgent treatment as to warrant screening without consent (Grosse et al. 2006). The U.S. Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children currently recommends that states screen all newborns for 31 core conditions, such as phenylketonuria or galactosemia, and report out 26 "secondary conditions" that are also detected when screening for the core conditions. A current list of recommended conditions may be obtained at [www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders](http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders). But many parents are not aware of or do not remember

receiving information about NBS (Davis et al. 2006; DeLuca et al. 2011; Hasegawa et al. 2011), often because information is provided in the hospital shortly before or after birth. Parents prefer to receive this information prenatally (Detmar et al. 2007), as recommended by professional associations (Faulkner et al. 2006; AAP American Academy of Pediatrics (AAP) Newborn Screening Taskforce 2000). However, in practice this rarely happens (Kim et al. 2003), especially for low-income mothers (Tluczek et al. 2009), or the information is included with other prenatal materials and is not noticed (Kemper et al. 2005). Brochures often have suboptimal readability and clarity, either about NBS (Arnold et al. 2006) or associated practices such as blood spot retention (Haga 2010). Parents need more complete, timely, and noticeable information (Cunningham-Burley 2006; Mann et al. 2006).

Experimental screening for conditions under consideration for NBS is more complicated. Although ethics guidelines argue that parental consent is necessary for pediatric research (Diekema 2006), informed consent is difficult and expensive to obtain in large pilot studies (Feuchtbaum et al. 2007). Some authors suggest that under certain circumstances (high potential benefit, minimal risk, impracticability) waiving consent may be appropriate (Tarini et al. 2008). However, this decision is likely to be nuanced and contingent on many factors, including whether the study is state authorized or the value of information disclosed.

Some studies, such as our fragile X pilot NBS study, clearly must be performed under a consent protocol. Fragile X syndrome (FXS) is the most common inherited form of intellectual disability, caused by a CGG repeat expansion within the *FMR1* gene at Xq27.3. Individuals with 200 or more CGG repeats are considered to have FXS, in which a protein necessary for normal brain development is reduced or absent. Individuals with 55–199 repeats are considered “fragile X(FX) carriers,” meaning that they have a gene expansion that increases their risk of having a child with FXS. In the absence of population screening, FXS typically is not diagnosed until 36 months or later (Bailey et al. 2009). Parents are frustrated by diagnostic delays, 25 % to 30 % have a second affected child before the first is diagnosed, and despite lack of medical treatment, identified children would be eligible to participate in early intervention programs (Bailey et al. 2005). Parents of affected children strongly support NBS (Bailey et al. 2012), but lack of data on early phenotype, the fact that a DNA-based screening test detects carriers, and absence of a proven FX-specific treatment mean that the evidence base is insufficient for FXS to meet current criteria for population screening (Calonge et al. 2010).

To provide a stronger evidence base, we are conducting a pilot study, designed as the social science equivalent of a Phase I clinical trial. In this case, the “treatment” is the

information families receive from screening. We are interested in whether and why parents agree to have their child screened and the “safety” of screening, as evidenced by any “adverse events” (e.g., postpartum depression, anxiety, disrupted parent-child relationships) (Bailey et al. 2008). Families are offered FXS NBS using a test that also detects carriers (Tassone et al. 2008). We recently reported a consent rate of 67.5 % for mothers and 63 % for couples (Skinner et al. 2011).

Given the genetic complexities of the *FMR1* gene (X-linked inheritance pattern with potential implications for many family members, triplet repeat expansion with “anticipation,” adult-onset conditions), we needed written materials to communicate this information. At the onset of the study, we created a pink and blue trifold brochure (no photographs) providing brief answers to 13 questions: (1) what is FXS? (2) how do children get FXS? (3) what is NBS? (4) what is the FX NBS study? (5) how is the study different from state NBS? (6) how will my baby be screened? (7) what will the study do? (8) why should I consider participating? (9) what risks are involved? (10) will I be contacted after the research screening is done? (11) will some babies need more testing? (12) what happens if my baby needs more testing? and (13) what else should I know about being in this study?

However, following an initial period of study implementation, four factors led us to decide to revise the original brochure. First, less than half of the parents reported reading it. Second, because carriers are more common than affected children, we wanted to emphasize the likelihood of carrier identification. Third, African American families were less likely than other families to agree to screening (Skinner et al. 2011), and we wanted to ensure that parents knew that FX affects all ethnicities. Finally, exposure to literature on Informed Decision-Making (IDM) led us to question whether parents were making truly informed decisions. IDM in health care is generally defined as the process by which patients are supported and involved in decisions about treatments or tests, weighing various considerations, examining values and preferences, and making a decision in partnership with a health professional (Briss et al. 2004; Charles et al. 1999; Mullen et al. 2006). IDM is especially important in situations where there is no right or wrong decision, because insufficient evidence exists to advise one option over another, or because the options all have risks and benefits that an individual must consider in order to be comfortable with the final decision (Elwyn et al. 2010). Often, printed materials or “decision aids” are developed to support IDM, using words, pictures, and figures to convey information, suggest reasons to consider or reject a course of action, and emphasize making a choice consistent with individual values and preferences (Bekker et al. 2003).

Decision aids work by providing information directly relevant to decision making and placing decisions in the context of personal values (Mullen et al. 2006). A systematic review of decision aids for prostate cancer screening (Volk et al. 2007) found that they generally result in improved knowledge and greater decisional confidence. A Cochrane review of 55 randomized clinical trials (O'Connor et al. 2009) concluded that decision aids increase knowledge and reduce decisional conflict.

The literature on IDM primarily focuses on helping patients make decisions about medical tests or treatments. With a few exceptions (e.g., Sorenson et al. 2004), less attention has been given to using decision aids to help individuals decide whether or not to participate in a research study. Nonetheless, the assumptions underlying IDM, namely that people ought to be supported in making health care decisions in a way that is consistent with their values and preferences, are directly applicable to decisions about study participation. Drawing on the IDM literature, we designed a new brochure to move beyond meeting Institutional Review Board requirements for informed consent, to meeting well-accepted standards for informed decision making. We wanted printed information that would: (1) be more visually appealing, hopefully increasing the chances that parents would read it; (2) clearly convey the likelihood of carrier identification; (3) use pictures to show that FX affects all races/ethnicities; and (4) support informed decision making. As the first step in a two-stage evaluation process, this article describes the process by which the brochure was developed; summarizes findings from an independent evaluation relative to decision aid standards; and reports the results of a simulation study with pregnant women or recent mothers. A subsequent paper will examine the effect of the brochure when implemented in a hospital recruitment environment. We conclude with a discussion of the growing need for informational aids to help parents understand complicated genetic information well enough to make an informed choice about participation in genetic research.

## Materials and Methods

### Brochure Design

Brochure development was guided by four IDM principles. It needed to (1) promote understanding of the study, risks, and uncertainties; (2) foster consideration of preferences; (3) support participation in decision making at a level that is desirable and personally comfortable; and (4) lead to a decision consistent with personal values (Mullen et al. 2006). A draft was developed by the first author and colleagues in health communications, then shared with the FX research team, including a certified genetic counselor, a

medical geneticist, an anthropologist, an attorney, an early childhood special educator, and an experienced bilingual (Spanish) research assistant. Multiple drafts were exchanged between the research team and the brochure development team. A pilot study was conducted with six pregnant women, and their suggestions were incorporated.

The text underwent several editorial reviews. We followed the tools/tips from PlainLanguage.gov, including the *Document Checklist for Plain Language* ([www.plainlanguage.gov/howto/quickreference/checklist.cfm](http://www.plainlanguage.gov/howto/quickreference/checklist.cfm)), for content and layout (e.g., useful headings, organized to serve readers' needs, active voice). Photographs, white space, and other design elements were used to enhance clarity and appeal. The SMOG readability formula (McLaughlin 1969) indicated that the brochure is written at a 9th grade level (+/- 1.5 grades), primarily due to 3-syllable words such as "family," "carrier," "development," and "genetics." Because these words were essential to understanding the study, by retaining them we were unable to further reduce reading grade level.

This iterative process resulted in a full-color, 8-page brochure with numerous photographs depicting infants and parents of multiple ethnicities. The first two pages differentiate fragile X syndrome from fragile X carrier, describe the incidence rate of each, and makes the point that carriers are much more likely to be identified than affected children. The brochure states that although there is no cure for FXS, children can receive help from early intervention programs and doctors can treat some symptoms. Following a description of what will happen in the study, two pages are devoted to "things to consider when making your decision." One page lists reasons to participate and another lists reasons not to participate. Also included are two quotes from parents who had decided to participate (e.g., "I'm the type of person who just wants to know") and two from parents who had declined (e.g., "I don't want to know if my child is a carrier; I think I would worry unnecessarily"). The final page contains five "questions to help you decide": (a) would you want to know if your infant has FXS? (b) would you want to know if your newborn is a FX carrier? (c) are you OK knowing that right now there is no cure for FXS? (d) do you have the information you need to make a decision? and (e) do you feel prepared to learn the answer of the screening test? The brochure concludes: "If you answered 'yes' to most of these questions, maybe you are ready to have your newborn screened. If you answered 'no' to most, maybe this is not the right decision for you."

### IPDAS Review

A near-final version of the brochure was submitted to an independent review group (the Cardiff University Decision Laboratory) to assess adherence to standards established by the International Patient Decision Aid Standards Collaboration

(IPDAS) ([http://www.ipdas.ohri.ca/IPDAS\\_checklist.pdf](http://www.ipdas.ohri.ca/IPDAS_checklist.pdf)) (Elwyn et al. 2006). The Decision Laboratory provided a detailed formative and summative assessment, including recommendations for improvement. The result was near-perfect scores on *informational content* (93 %—the brochure describes the problem, the decision to be made, and the options available. Positive and negative features are presented using equal detail in a format that allows fair comparison); *guidance* (100 %—the brochure provides structured guidance toward making a decision); and *values* (95 %—the brochure facilitates the expression and clarification of user values and attitudes). The brochure received low ratings on other items because it (1) did not include data on chances of false positive or false negative results and did not present probabilities in multiple ways; (2) did not provide details about the development process; (3) did not provide evidence supporting brochure content; and (4) lacked evidence for efficacy. These lower ratings were expected, given that the brochure was still in development; the primary purpose of the review was to determine whether the content of the brochure was consistent with recommended practices for developing patient decision aid.

#### Participants

The study was reviewed and approved by the Institutional Review Board at RTI International. A local firm recruited 118 pregnant women (59 %) or recent (within the past 6 months) mothers (41 %) for a simulation study. The women had a mean age of 30.4 years, ranging from 18–43. The group was relatively well educated: only 13 % had a high school degree or less, 24 % had some college or technical school, and the remainder had at least a college degree. Nine (7.6 %) were Hispanic/Latino (one also self-identified as African American), 47 (39.8 %) African American, and 62 (52.5 %) white. Most (74 %) were married and 58 % were employed. Their median household income was approximately \$50,000; 11 % had a household income of less than \$20,000 and 16 % over 100,000. Twenty (17 %) had heard of FXS but only 3 (2.5 %) knew someone with FXS. Participants received \$50 upon activity completion.

#### Procedures and Instrumentation

Each woman participated in one of nine 1-hour group sessions facilitated by a member of the research team. They were told that the goal was to understand their opinions and reactions to a brochure about a research study. They were not given any other information about FXS, the study, or NBS. The women needed 2–26 min to read the brochure, an average of 6.5 min. Each then responded to the following statement: “Based on the information I read in the brochure about the FX NBS study, if I was approached by someone to participate in this study, I would agree/not agree to have my

baby screened” and wrote reasons for their decision. Women then took a knowledge test containing 12 true-false statements designed by the authors to assess factual recall. They completed a survey containing 31 statements rated on a 5-point scale from Strongly Agree to Strongly Disagree. The statements addressed reactions to the brochure (e.g., I like the way this brochure looks; I trust the information) and included selected items adapted from the Decisional Conflict Scale (DCS) (O’Connor 1995).

#### Results

Descriptive statistics and summary scores were used to characterize performance on the knowledge test, reactions to the brochure, and hypothetical screening decisions. We used structural equation modeling to examine whether selected demographic variables were associated with test performance, perceptions of the brochure, decisional uncertainty, or screening decisions.

#### Knowledge

Mean percentage correct on the knowledge test was 91.1, ranging from 50–100 % (Table 1). All but three items were answered correctly by >91 % of the participants. The terms “small gene change” and “large gene change” used to differentiate carriers from affected children resulted in some confusion. About 20 % of the women incorrectly thought that an extra prick of the baby’s heel was needed for the study.

#### Perceptions of the Brochure and Decisional Support

Combining ratings of agree and strongly agree, most women reported that the brochure was easy to read (95.8 %) and understand (89.9 %); they liked the way it looked (91.6 %); and it provided helpful information (90.8 %). The majority agreed or strongly agreed that it would help them make an informed decision about participating in the study (78.2 %) and they trusted the information (69.8 %). Some (26.9 %) said that the brochure left them with many unanswered questions about FX and 21 % reported that they were still unsure about study participation. The most common suggestions for improvement were more information about FXS and the study itself.

#### Decision to Participate

When asked to make a hypothetical decision, 61.9 % indicated that they would agree to have their child screened. Some non-significant variation was seen across ethnic

**Table 1** Percent correct on knowledge test items for participants in the simulation study ( $N=118$ )

Item	Answer	% Correct
1. Having fragile X syndrome and being a fragile X carrier are the same thing.	False	97.5
2. Children with fragile X syndrome can have delays in development, learning problems, signs of autism or anxiety.	True	98.3
3. There is a cure for fragile X syndrome.	False	98.3
4. Fragile X is only found in certain ethnic or racial groups.	False	91.5
5. Being a fragile X carrier means there is <i>small</i> change in the fragile X gene.	True	89.9
6. During the Fragile X Newborn Screening Study an extra prick of the baby's heel is needed so that a blood spot can be taken for the study.	False	79.8
7. Children who have fragile X syndrome cannot receive help from early intervention programs.	False	97.5
8. A newborn that tests positive as a fragile X carrier has a parent who also is a fragile X carrier.	True	94.2
9. Fragile X syndrome is caused by a <i>large</i> gene change.	True	63.6
10. The Fragile X Newborn Screening Study hopes to learn about the early development of children with fragile X syndrome and children who are fragile X carriers.	True	99.2
11. Most people who are carriers of fragile X already know it.	False	100
12. There are many more people who have fragile X syndrome than people who are fragile X carriers.	False	95.8

groups, with 63.9 % of non-Hispanic whites and 56.8 % of African Americans agreeing. Six (75 %) of the eight Hispanic (non-African American) women would agree to have their child screened.

An open-ended question asked women to explain their choice. Their reasons are summarized in Tables 2 and 3 and compared with the reasons reported in our larger hospital study using the original brochure (Skinner et al. 2011). Most women (91.7 %) who would agree to participate reported benefit to knowing earlier: "I would want to know if my baby had FX or was a carrier so that I could prepare for any challenges down the road"; "I would have the available resources that are out there to help my baby as well as our family to cope with this genetic disease." The next most common reasons (25 %) reflected a belief that participation posed minimal risk: "As long as the child is not undergoing any additional unnecessary pain, I only see good in testing, whether it is curable or not"; "it's a non-invasive test that can give an enormous amount of information." These two reasons were also commonly mentioned with the original brochure (Skinner et al. 2011); however, parents responding to the new brochure were less likely to mention "contribute to research" as a reason to participate.

The reasons for not participating were more diverse and exemplified a different pattern than seen with the old brochure. The most common was lack of a cure or treatment, mentioned by 51 % of the women in this study but only 5 % in the hospital study. These women made comments such as "since there is not a cure at this moment, I would prefer not to test my child"; "if there is no cure, it's just knowledge without purpose."

A substantial portion (44.4 %) also indicated that they did not want to worry, compared with 21.4 % with the original brochure. These women made comments such as "I am one of those people that would worry myself about it"; "being pregnant with my first there are a lot of things that I can worry about, most of them I must choose not to." Also, 28.9 % of women who read the new brochure (compared with only 9.4 % in the hospital) reported that they would rather wait for symptoms to appear: "I will continue to monitor my child to see if any developmental issues appear over time." Other responses (28.9 %) referenced issues regarding test accuracy: "the brochure mentions that the test results could be wrong"; "if there was a wrong diagnosis, that would upset me as well." Understandably, concerns about logistics (e.g., the context or timing is not good) were more common in the hospital group (21.4 %) compared with the simulation group (4.4 %).

#### Decisional Uncertainty

The three items in the Decisional Uncertainty subscale of the Decisional Conflict Scale (O'Connor 1995) were adapted for this study, displayed in Table 4. About 75 % of the mothers agreed or strongly agreed that the brochure made it easier to decide about study participation; 62.2 % disagreed or strongly disagreed that they were still uncertain about study participation, and 66.4 % agreed or strongly agreed that the brochure made it clear "what the best choice is for me."

**Table 2** Reasons for accepting screening: percentages across studies and ethnic groups

Reason	Skinner et al. (2011) (n=1288)	Simulation Study Total (n=72)	African American (n=28)	White (n=38)	Hispanic (n=6)
Knowing is good; benefit to knowing; knowing earlier is better	71.6	91.7	92.9	89.5	100
To contribute to research	32.0	6.9	3.6	7.9	16.7
Test is minimal risk; non-invasive; just an additional test	27.5	25.0	21.4	23.7	50
Participating can't hurt; nothing to lose	8.4	1.4	–	2.6	–
Family has history of problems	5.9	2.8	3.6	2.6	–
Screening is free	4.7	9.7	3.6	15.8	–
Just curious	2.1	–	–	–	–
Spouse/partner convinced me	2.1	–	–	–	–
Because the screen was offered	1.4	–	–	–	–
To provide reproductive risk information	0.6	1.4	3.6	5.3	–

Percentages sum to greater than 100 % because participants reported more than one reason

### Factors Associated with Outcomes

The path diagram in Fig. 1 outlines our hypothesized model of the decision-making process. In this model, demographics and having heard of FXS were predicted to influence perceptions of brochure quality; quality, in turn, was predicted to affect the screening decision and decisional confidence, both directly and indirectly through trust in the information. We hypothesized that non-white respondents would be less likely to trust the information, given prior research showing ethnic differences in trust in research more broadly and elevated concerns about research and the consequences of research findings for members of ethnic minority groups (Bussey-Jones et al. 2010; Nwulia et al. 2011). We also hypothesized that individuals who were somewhat familiar with FXS would be more likely to trust the information in the brochure and thus more likely to accept

screening, since they would be more aware of the consequences of FXS for children and families. We conducted a path analysis to test the model using the Mplus software program for structural equation modeling (Muthén and Muthén 1998–2010). Various model fit indices were used to assess goodness of fit; values of 0.95 or higher for the comparative fit index (CFI) and Tucker-Lewis Index (TLI) and values of 0.06 or less for the root mean square error of approximation (RMSEA) indicate good fit (Bentler 1990; Browne and Cudeck 1990; Hu and Bentler 1999).

The path model fit very well (Fig. 1; CFI=0.98, TLI=0.96, and RMSEA=0.05). Women with a college education rated the quality of the brochure less positively than those with less education (coefficient=-0.25,  $p<0.05$ ); and perceived quality of the brochure was not significantly related to age, ethnicity, or familiarity with FX. Ethnicity was not associated with trust in the information; however, those who

**Table 3** Reasons for declining screening: percentages across studies and ethnic groups

Reason	Skinner et al. (2011) (n=565)	Simulation Study Total (n=45)	African American (n=21)	White (n=22)	Hispanic (n=2)
Logistics (the context, timing is not good)	21.4	4.4	0	9.1	0
Don't want to worry	21.4	44.4	52.4	36.4	50.0
Issues regarding testing or test accuracy	19.3	28.9	38.1	22.3	0
Don't want to know	17.7	20.0	19.1	22.8	0
Don't want to be in a study, not interested	14.9	2.2	0	4.6	0
It's not necessary	13.8	2.2	4.8	0	0
Little chance of having it; no family history	12.4	11.1	19.1	0	50
Spouse/partner declined or disagreed	11.5	NA	NA	NA	NA
My baby is fine/ healthy	11.0	NA	NA	NA	NA
Rather wait for symptoms to appear	9.4	28.9	19.1	59.1	0
No cure or treatment	5.3	51.1	42.9	59.1	50

Percentages sum to greater than 100 % because participants reported more than one reason

**Table 4** Percentage of respondents endorsing different levels of agreement with decision uncertainty items ( $N=118$ )

Item	Strongly agree	Agree	Neither agree nor disagree	Disagree	Strongly disagree
The brochure made it easier to make a decision about participating in the fragile X newborn screening study	26.1 <sup>a</sup>	48.74	16.8	7.6	0.8
After reading the brochure, I'm still unsure about participating in the fragile X newborn screening study	6.8	14.3	16.8	36.13	26.1
The brochure made it clear what the best choice is for me in terms of participating in the fragile X newborn screening study	20.2	46.2	18.5	12.6	2.5

<sup>a</sup> Percentage of respondents

had heard of FX were significantly less likely to trust the information (coefficient=-0.22,  $p<0.05$ ). Greater perceived quality was associated with greater trust in the information (coefficient=0.62,  $p<0.001$ ). Women who gave high quality ratings and those who trusted the information more were significantly more likely to agree to screening and reported greater decisional confidence.

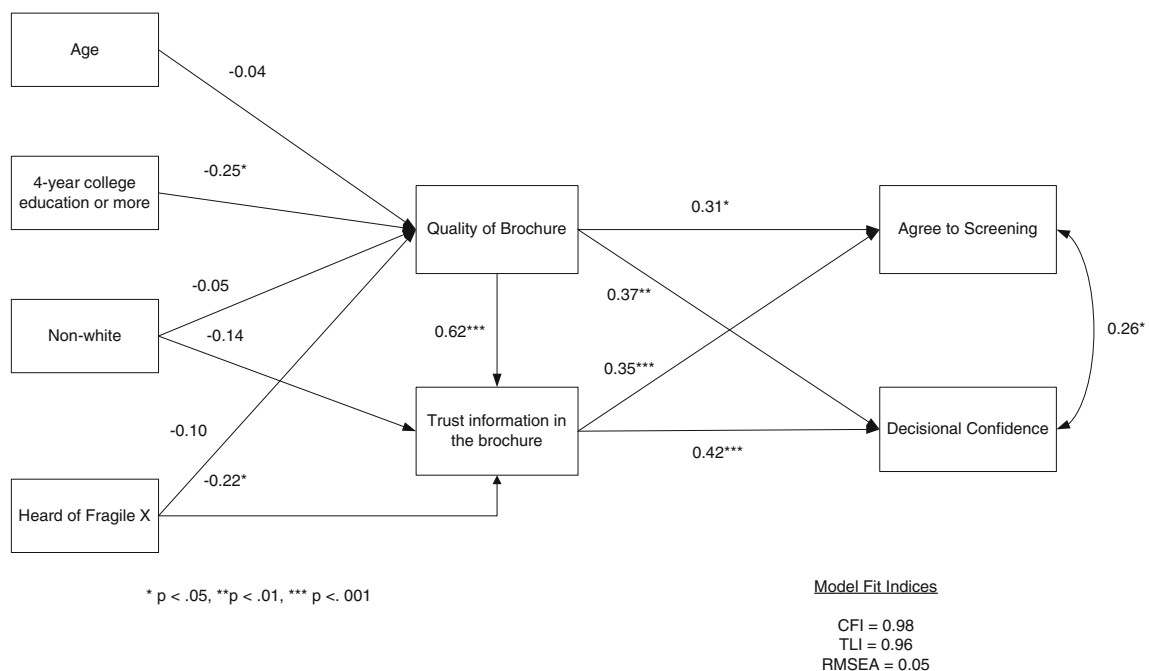
**Discussion and Conclusions**

**Discussion**

Study recruitment materials typically are designed to meet Institutional Review Board (IRB) requirements for informed consent. When research involves complicated decisions with direct ramifications for study participants, as in the case of a study disclosing genetic information of uncertain value about newborns, researchers have an obligation to

provide information that supports informed decisions. Newborn screening for FXS and the disclosure of infant carrier status clearly exemplify this obligation. No urgent medical treatment is currently available for FXS, and some parents may not want to know infant carrier status. In developing new written materials about the study, our goal was not to increase study participation rates, but to develop print materials that, if read, would maximize awareness of all facets of the study and enable parents to make an important decision in a relatively short period of time.

IDM provides a theoretical framework for fulfilling this obligation, because its underlying premise is to help people make decisions consistent with their values and preferences. IDM is well established in the design and evaluation of patient decision aids, but with few exceptions (Sorenson et al. 2004), relatively little attention has been given to its application in research recruitment. This article demonstrates that using IDM as a guiding framework can result in recruitment materials that are informative, balanced, and



**Fig. 1** Path analysis of agreement to newborn screening and decisional confidence

supportive of decision-making. Both IDM experts and women who could potentially be invited to participate in a hospital study rated the brochure high in quality and trustworthiness. The brochure required most women only about 6 min to read and resulted in high recall of facts. Perceptions of quality and trustworthiness were directly associated with the decision to participate and decisional confidence. African American and Latino mothers were no less likely to trust the brochure than white mothers and did not differ in terms of perceived quality of the brochure, suggesting that we were able to make some inroads into offsetting mistrust in research among minority families.

Interestingly, although the new brochure did not result in substantial difference in hypothetical rates of study participation, it did result in a shift in reasoning. For example, in this study few women reported “to contribute to research” as a reason for deciding to participate in the study. This difference may be due to the fact that the original report of consent rates was conducted in a teaching hospital where research is more common, or it may be due to the fact that the simulation study was focused on reactions to the brochure whereas in the hospital the focus was on making an actual decision to have your baby screened. Women who said they would decline after reading the new brochure were much more likely to mention lack of a cure and not wanting to worry as reasons for not participating in the study. The original brochure did not mention “no cure,” and so it is not surprising that few parents mentioned it in our original study. The fact that more than 50 % of decliners in the simulation study mentioned lack of a cure and 44 % did not want to worry suggests that for these women, a clear treatment option is a salient factor in their decisions about whether they would want to know information about their child’s fragile X status.

#### Study Limitations

The study has several limitations. It is possible that the study participants, by virtue of the fact that they agreed to be in the simulation study, were more inclined to be in and trust research, and thus their perspectives on research might be more favorable than the general population. We did not directly compare the old and new brochure nor did we do a knowledge pre-test, so we cannot say that the new brochure was better than the original. We were unable to reduce the reading level below 9th grade without eliminating essential 3-syllable words such as *family* or *carrier*. Most women performed well on the knowledge test, but we do not know the literacy threshold below which this brochure would not be effective, given that study participants were relatively well educated. Alternative strategies are clearly needed for low-literacy parents. The finding that mothers who

had heard of FXS were less likely to trust the brochure is puzzling, and we have no data to suggest why that might be the case. The survey only asked mothers if they had heard of FXS, but we do not know how or what they knew about it. Finally, in the IDM literature, decision aids typically are used in conjunction with discussions with health care providers or family members. In our original hospital study, a research assistant was available to talk with families about the study, but the simulation study offered no such opportunity, so our findings are limited to hypothetical decisions made alone by women after a single reading.

#### Future Directions

Using IDM as a foundational framework, we developed a study recruitment brochure that was highly rated by experts and consumers, met high standards for IDM, and achieved some of our stated goals in a simulation study. But a brochure only has the potential to be useful if it is read. We are conducting a companion implementation study in a hospital to test its ultimate utility, assessing whether the brochure was more likely to be read and the extent to which the new brochure changes rates of study participation.

#### Practice Implications

With the advent of DNA-based and other next-generation sequencing technologies, research will be needed to determine how families understand and use genetic information, and, more fundamentally, whether they want that information at all. For example, if these technologies became standard for newborn screening, the public health screening program as we know it could change fundamentally (Goldenberg and Sharp 2012). State health departments will obtain information about a wide range of genetic variants and decisions will have to be made about what information to disclose, when to disclose it, and how. Systematic practice-based research will be needed and ultimately newborn screening may need to include a voluntary component for disclosing information that does not meet the “public health emergency” standard. Parents will expect to have a say in the information disclosed, but their decision must be informed and supported. IDM provides a set of relevant guiding principles, because this context mimics prior applications of IDM to health care decisions where there is no right or wrong answer. But making the information understandable and finding realistic opportunities for parents to weigh alternatives and make an informed decision will be an enormous public health challenge.



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