

Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing

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Abstract Noninvasive prenatal testing (NIPT) allows for highly sensitive detection of Down syndrome early in pregnancy with no risk of miscarriage, therefore potentially increasing the number of pregnancies identified with Down syndrome. This study assesses how mothers of children with Down syndrome perceive NIPT, especially the impact they think it will have on their families and other families with children who have Down syndrome. Seventy-three self-reported mothers of children with Down syndrome responded to an anonymous online survey emailed to, and posted on, message boards of various Down syndrome support groups and networks. Data analysis included chi-square tests and thematic analysis. Fifty-nine percent of respondents indicated they would use NIPT in the future; respondents who had not used prenatal testing in the past were significantly less likely to report interest in using NIPT in the future than those who had prenatal testing previously ($p < .001$). Many respondents felt NIPT could lead to increased terminations (88 %), increased social stigma (57 %), and decreased availability of services for individuals with Down syndrome (64 %). However, only 16 % believed availability of new noninvasive tests would be the most important factor in determining the number of pregnancies with Down syndrome terminated in the future. Additionally, 48 % believed health care providers give biased or incorrect information about Down syndrome at the time of

diagnosis, and 24 % felt this incorrect information leads to terminations of pregnancies affected with Down syndrome. Results suggest although mothers of children with Down syndrome believe new noninvasive testing will lead to an increase in termination of pregnancies with Down syndrome, they do not think it is the MOST important factor. They also highlight the need to provide a diagnosis of Down syndrome in a balanced and objective manner.

Keywords Noninvasive prenatal testing (NIPT) · Down syndrome · Stakeholder views · Parent perceptions · Information provision

Although prenatal testing for fetal aneuploidies has been standard of care for decades, the recent availability of cell-free fetal DNA in maternal serum, first discovered in 1997 (Lo et al. 1997), has allowed for the screening of fetal aneuploidies (Chiu et al. 2008; Fan et al. 2008) with significantly higher levels of sensitivity than in the past (Bianchi et al. 2012; Palomaki et al. 2011, 2012) and without the risk of miscarriage associated with invasive, diagnostic prenatal tests. Current evidence supports the use of noninvasive prenatal testing (NIPT) only in select, high-risk populations (ACOG 2012; Benn et al. 2012a, 2012b; Devers et al. 2013), and only with appropriate pre- and post-test counseling (Gregg et al. 2013). However, given that NIPT provides highly sensitive results with no risk to the fetus, its use as a first pass screening measure is expected to grow dramatically in coming years, including in low risk women (Greely 2011). Since NIPT confers no risk of miscarriage, which is proposed as one of the key decision making factors for women who decline invasive testing, it may be more appealing to people who would otherwise not have undergone prenatal testing (Nakata et al. 2010; Tischler et al. 2011).

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There is some concern that an increase in prenatal testing could lead to the routinization and trivialization of termination (De Jong et al. 2010). This could lead to decreased births of children with Down syndrome (Skotko 2009), and ultimately a decrease in social acceptance of the condition, with a corresponding decrease in social support and services for those who have Down syndrome and their families (Skotko 2009). Therefore, it is important to understand how members of the Down syndrome community view NIPT, as it has the potential to significantly impact their families.

Public attitudes towards non-invasive prenatal diagnosis have been assessed. A prior study of pregnant women ($N=114$) at Stanford found that 71.9 % expressed interest in using NIPT (Tischler et al. 2011); a study utilizing a diverse sample population ($N=71$) in United Kingdom found that 63 % of respondents had a positive first response to hearing about NIPT (Kelly and Farrimond 2011); and a study in Japan ($N=252$) found that 97.8 % of pregnant respondents felt that a woman should be allowed to undergo noninvasive prenatal testing if she so desired (Yotsumoto et al. 2012). Tischler et al. (2011) also found that 1 in 5 women ($N=114$) would pursue whichever prenatal test their doctor recommended, raising ethical questions about the inconsistent informed consent process involving prenatal screening. The attitudes of relatives of individuals with Down syndrome towards serum screening and invasive diagnostic testing have been assessed as well, with the majority of respondents ($N=78$, $N=101$) typically responding favorably towards these tests (Bryant et al. 2005; Inglis et al. 2012, respectively). However, to our knowledge, no study has explicitly examined the attitudes of mothers of children with Down syndrome towards NIPT.

Since NIPT has the potential to impact the Down syndrome community in such a dramatic way, it is important to assess the attitudes of this valuable group of stakeholders in order to implement NIPT such that it respects the rights, interests, and autonomy of all involved. This study will assess both current attitudes towards NIPT and what impact mothers of children with Down syndrome think increased use of NIPT in the future will have on their families and other families with children who have Down syndrome.

Method

Sample

Respondents are self-reported mothers of children with Down syndrome. A link to the online survey was disseminated via email to, and posted on the message boards of, regional and local Down syndrome support groups and networks between October and December 2012.

Instrumentation

The survey (Appendix) began with a short description of NIPT developed by the investigators. NIPT was described as a test that “can diagnose some chromosome differences, like Down syndrome, earlier in pregnancy” and as a test that does “NOT have a risk of miscarriage and is very accurate.” NIPT (Noninvasive prenatal testing) is the term that was used in the materials that were provided to the study participants and is used in this manuscript for consistency, although the recent recommendation from ACMG is to use the term NIPS (non-invasive prenatal screening) for this testing. The listed limitations of NIPT included having a more limited scope of prenatally detectable conditions as compared to invasive tests, and the possibility for ambiguous results. The description included options available to a woman if a chromosome abnormality was diagnosed prenatally, including termination of pregnancy and expectant management.

The 17 question survey took between 10 and 20 min to complete, and included both demographic questions (6 questions, see Table 1) and Likert-scale format attitudinal questions about NIPT (11 questions, see Table 2) that assessed current attitudes towards NIPT, and also what impact the respondent felt increased use of NIPT would have on her child’s future. Genetic counselors, including a counselor who practices in a clinic specifically for individuals with Down syndrome, and a medical geneticist developed attitudinal questions. Questions were designed to be similar to those asked in previous studies of the attitudes of pregnant women towards NIPT (Tischler et al. 2011) and of the attitudes of parents of children with Down syndrome towards traditional prenatal testing methods (Inglis et al. 2012). Respondents were asked to rank order the factors that they felt would most impact how many pregnancies affected with Down syndrome would be terminated in the future, and to answer multiple choice questions regarding to whom and for what reasons NIPT should be available. Finally, respondents were able to write in any additional thoughts they had about NIPT in a free-response section at the end of the survey.

Data Analysis

SPSS Statistical Software (Version 21) and Qualtrics Survey Software were used to perform chi-square analysis to determine if demographic differences correlated to respondents’ responses to questions regarding NIPT (Table 3). Demographic groupings included ethnicity, religiosity, education level, and age; these groups have shown differences in attitudes towards both NIPT and other forms of prenatal screening and testing in previous studies (Press and Browner 1998; Tischler et al. 2011). Free text comments were evaluated qualitatively by two investigators (GK, KO). Potential biases in coding data were discussed by authors prior to beginning

coding. Comments were read by both investigators, who generated a coding list for discussion, and then applied the codes to comments. All codes were developed from responses. Codes were first applied by GK, and then audited by KO. Discrepant coding was discussed until agreement was reached on the final coding assignment. Key themes from the free comment section are presented with quotes to illustrate the typical response (Table 4).

The survey and research methodology were approved by the Stanford Internal Review Board.

Results

Sample Demographics

Given the method of survey distribution, the total number of surveys distributed is unknown and a response rate cannot be calculated. Eighty-two respondents clicked the survey link, 73 at least partially completed the survey, and 67 surveys were completed fully (82 %). Table 1 lists the demographic features of the sample, which was predominantly Caucasian (78 %) and well educated (78 % had a college degree or higher). Twenty-nine percent of respondents reported having received no prenatal screening or testing for Down syndrome in their pregnancy, while 22 % ($n=16$) reported having had an invasive diagnostic test. Of these 16 respondents who had an invasive diagnostic test, 7 reported having screening tests with a high risk for Down syndrome, while 5 reported that they had had screening with low or average risks (4 did not report screening risks).

Participant Attitudes

Table 2 lists the attitudes reported by study participants. Fifty-nine percent of respondents agreed or strongly agreed that they would consider using NIPT if they become pregnant in the future, and slightly fewer (44 %) would recommend NIPT to a pregnant friend. The likelihood of considering using NIPT in a future pregnancy was not significantly related to current age, $\chi^2(1)=1.25$; $p=0.26$, age at birth of child with Down syndrome, $\chi^2(1)=0.76$; $p=0.38$, religiosity, $\chi^2(1)=3.1$; $p=0.08$, or education level, $\chi^2(1)=0.09$; $p=0.77$ (Table 3). However, respondents who did not use any prenatal testing in their previous pregnancy were significantly less likely to report considering NIPT in a future pregnancy than those who had used prenatal testing, $\chi^2(1)=11.37$; $p<.001$ (32 % versus 76 %, respectively).

The majority of respondents agreed or strongly agreed NIPT would lead to more prenatal tests for Down syndrome (80 %), and that NIPT would lead to the termination of more pregnancies with Down syndrome (88 %). When asked to indicate the factors that would most influence the number of

Table 1 Demographics

	<i>n</i> *	(%) Total
Current age		
20–25	1	1
26–30	5	7
31–35	10	14
36–40	22	30
41–45	18	25
Over 45	17	23
Age at birth of child with down syndrome		
20–25	1	1
26–30	15	21
31–35	25	34
36–40	19	26
41–45	11	15
Over 45	2	3
Religiosity		
Very	20	27
Moderately	17	23
Somewhat	19	26
Not at all	17	23
Ethnicity ^a		
African-American	0	0
Asian/Pacific Islander	8	11
Caucasian (Non-Hispanic)	57	78
Hispanic	13	18
Educational level		
High school degree or less	2	3
Some college	14	19
College degree	33	45
Graduate/Post-graduate degree	24	33
Previous prenatal testing ^a		
Screening blood test and/or ultrasounds with average or low risk results for DS	30	41
Screening blood test and/or ultrasounds with increased or high risk results for DS	16	22
Screening blood test and/or ultrasounds but I am unsure what the results were	1	1
CVS or amniocentesis that confirmed Down syndrome	16	22
No prenatal screening or diagnostic tests for Down syndrome	21	29
I don't know	2	3

^a Participants could endorse multiple responses

*n's vary as not all participants responded to every question

pregnancies affected by Down syndrome in the future that would be terminated in the future, respondents most often chose a person's moral or religious beliefs (36 %), and the information provided to families at the time of a prenatal diagnosis of Down syndrome (30 %), while only 16 % selected the availability of new noninvasive tests. When asked to

Table 2 Interest in and attitudes towards NIPT

	<i>n</i> *	(%) Total
I think that noninvasive prenatal testing should be available to		
All women	49	67
Women who are at increased risk to have a baby with Down syndrome, either based on their age or screening results	14	19
No one	10	14
In my next pregnancy, or if I were going to have another pregnancy, which of the following best describes the plan I would choose for prenatal screening and/or testing?		
Screening through blood tests and/or ultrasounds only	9	12
Invasive diagnostic tests (amniocentesis, CVS) only	3	4
Screening followed by diagnostic tests if the pregnancy were at increased risk	6	8
Noninvasive prenatal testing followed by invasive diagnostic tests for confirmation	8	11
Noninvasive prenatal testing only	24	33
No prenatal testing for Down syndrome	23	32
If I become pregnant again, I would think about using noninvasive prenatal testing		
Strongly agree	26	36
Agree	17	23
Neither agree nor disagree	5	7
Disagree	8	11
Strongly disagree	17	23
I would recommend noninvasive prenatal testing to a pregnant friend		
Strongly agree	17	23
Agree	15	21
Neither agree nor disagree	16	22
Disagree	9	12
Strongly disagree	16	22
I believe that noninvasive prenatal testing will lead to more prenatal diagnostic tests for Down syndrome		
Strongly agree	32	44
Agree	26	36
Neither agree nor disagree	12	17
Disagree	0	0
Strongly disagree	2	3
I think that noninvasive prenatal testing will lead to the termination of more pregnancies affected with Down syndrome.		
Strongly agree	41	57
Agree	22	31
Neither agree nor disagree	7	10
Disagree	2	3
Strongly disagree	0	0
I think that new noninvasive tests will cause women to feel they have to have diagnostic prenatal testing for Down syndrome		
Strongly agree	31	43
Agree	23	32
Neither agree nor disagree	9	13
Disagree	7	10
Strongly disagree	2	3

Table 2 (continued)

	<i>n</i> *	(%) Total
I think new noninvasive tests will cause an INCREASE in social stigma for having a child with Down syndrome		
Strongly agree	27	38
Agree	14	19
Neither agree nor disagree	14	19
Disagree	14	19
Strongly disagree	3	4
I think that if fewer children are born with Down syndrome services, including medical care, physical therapy, occupation therapy, speech therapy, and school programs, available to people with Down syndrome will DECREASE		
Strongly agree	27	38
Agree	19	26
Neither agree nor disagree	11	15
Disagree	10	14
Strongly disagree	5	7
Which of the following best describes how I feel about noninvasive prenatal testing?		
It is a <i>good</i> thing that noninvasive prenatal tests for Down syndrome are available. They help people to find out for sure whether or not their pregnancy is affected with Down syndrome early and without the risk of miscarriage that current invasive tests have, and <i>allow people to make decisions about whether or not to continue with the pregnancy based on that information.</i>	10	16
It is a <i>good</i> thing that noninvasive prenatal tests for Down syndrome are available. They help people to find out for sure whether or not their pregnancy is affected with Down syndrome early and without the risk of miscarriage that current invasive tests have, and <i>can allow people more time to prepare themselves to care for a child with Down syndrome.</i>	28	44
It is a <i>bad</i> thing that noninvasive prenatal tests for Down syndrome are available. I feel their only purpose is to enable people to terminate pregnancies that are affected with Down syndrome.	18	28
I am not sure what I think about noninvasive prenatal tests for Down syndrome.	8	13
What is the MOST important factor that will determine the number of pregnancies with Down syndrome that are terminated?		
The availability of new noninvasive prenatal tests	11	16
The information provided to families about Down syndrome when they receive a prenatal diagnosis	21	30
A person's personal beliefs (ethical, moral, religious) about terminating a pregnancy	25	36
A person's past experience with people with disabilities	9	13
A person's socioeconomic status or life circumstances	3	4

*n's vary as not all participants responded to every question

choose one statement that best reflected their feelings about NIPT (see Table 2), 44 % indicated that NIPT was a “*good* thing ...[because NIPT] help[s] people to find out for sure whether or not their pregnancy is affected with Down syndrome early and without the risk of miscarriage that current

Table 3 Interest in using NIPT in a future pregnancy compared with demographic and other factors^a

	Strongly agree/Agree	Strongly disagree/Disagree	χ^2 -Value	<i>p</i> -value
Education level	<i>n</i> (%)	<i>n</i> (%)		
College degree or higher	34 (64 %)	19 (36 %)	0.09	0.77
Less than a college degree	9 (60 %)	6 (40 %)		
Religiosity				
Very/Moderately	18 (53 %)	16 (47 %)	3.1	0.08
Somewhat/Not at all	25 (74 %)	9 (26 %)		
Current age				
>35 years	31 (46 %)	21 (47 %)	1.25	0.26
<35 years	12 (75 %)	4 (25 %)		
Age at birth of child with Down syndrome				
Above 35	16 (57 %)	12 (43 %)	0.76	0.38
Below 35	27 (68 %)	13 (33 %)		
Prenatal testing in pregnancy with Down syndrome				
Had prenatal testing	37 (76 %)	12 (24 %)	11.37	0.00075*
No prenatal testing	6 (32 %)	13 (68 %)		
Ethnicity				
Asian/Pacific Islander	5 (71 %)	2 (29 %)	1.16	N/Ab
Caucasian	35 (65 %)	19 (35 %)		
Hispanic	6 (50 %)	6 (50 %)		

^a Categories collapsed to avoid small cell size; ^b N/A due to insufficient number of responses

*significant at *p*-value significance level < .001

invasive tests have, and *can allow people more time to prepare themselves to care for a child with Down syndrome,*” while a significant minority (29 %) stated that NIPT was a “*bad thing [whose] only purpose is to enable people to terminate pregnancies that are affected with Down syndrome.*” Sixteen percent selected the option stating “*It is a good thing that noninvasive prenatal tests for Down syndrome are available. They help people to find out for sure whether or not their pregnancy is affected with Down syndrome early and without the risk of miscarriage that current invasive tests have, and allow people to make decisions about whether or not to continue with the pregnancy based on that information.*” The remaining percentage (13 %) selected the option stating “*I am not sure what I think about noninvasive prenatal tests for Down syndrome.*”

Qualitative Results

Thirty-six open-ended responses were recorded; three were omitted from the qualitative analysis since they were exclusively comments about the survey wording. The remaining 33 were coded thematically, and yielded 10 major themes. The themes are listed in Table 4, along with illustrative quotes. The most frequent themes included: health care providers have biased or incorrect information about Down syndrome (48 %); a personal story or anecdote (39 %); the test (NIPT)

leads to termination [of affected pregnancies] (30 %); a description of how prenatal testing is related to the autonomy of the patient (30 %); a statement regarding the social implications of disability (27 %); and how wrong information leads to termination [of affected pregnancies] (24 %).

Discussion

This study is one of the first to assess the attitudes of mothers of children with Down syndrome towards noninvasive prenatal testing. In the few studies that have been performed assessing the attitudes of relatives of children with Down syndrome towards traditional prenatal testing, the majority of respondents (typically 55–65 %) have responded positively towards prenatal testing for themselves (Bryant et al. 2005; Inglis et al. 2012) or others (Bryant et al. 2005). These numbers are consistent with our findings that 50 % of respondents would personally consider NIPT in a future pregnancy, and that 67 % felt it should be available to all women, citing that testing increases the autonomy of the patient (30 %), and that it can help families prepare for the birth of an affected child (21 %). Respondents also felt that the availability of NIPT would lead to an increase in prenatal diagnosis uptake (80 %). Despite this overall support for the availability of NIPT, the

Table 4 Parents of children with down syndrome free response themes

Theme	Typical Quote	n* (%)
Health care providers have biased or incorrect information about Down syndrome	I think prenatal tests allow medical professionals to tell parents all of the negatives about their baby. [A mother] is told all of the scary parts about DS and none of the good parts.. [Parents should be] presented with balanced information about the positives AND negatives of raising a child with special needs. Only if they are exposed to both sides of the story can they TRULY make an informed decision about whether to terminate.	16 (48 %)
Personal story or experience about the prenatal or postnatal diagnosis of child with Down syndrome	We knew ahead of time. It gave us time to prepare and learn more about people with Down Syndrome.	13 (39 %)
Test leads to termination	I fear that finding out earlier and without risk will make it easier for other parents to disengage from their child and make a decision to terminate their pregnancy without having fully considered the child they are giving up.	10 (30 %)
Autonomy	I think everyone has a right to information that can affect their future.	10 (30 %)
Social implications of disability	[Having a child with Down syndrome entails] such a complicated web of issues because without social support, government assistance, affordable healthcare, etc., the burdens can become overwhelming and propitiate the stigmas associated with developmental delays. However, those same stigmas cause some/many people to terminate their pregnancies, which, along with more prenatal screening, results in few babies and therefore fewer services and increase stigma.	9 (27 %)
Wrong information leads to termination	I worry that the misinformation that medical professionals are currently providing paints a bleak picture of the child's future that will result in increased terminations.	8 (24 %)
Test can help families prepare	I would like to think that earlier and more reliable testing will give people more time to prepare for their life changes.	7 (21 %)
Health care providers encourage termination after a diagnosis of Down syndrome	I think the medical community as a whole actively pushes for termination if a positive result is returned.	6 (18 %)
Test not bad in and of itself	...Like the saying that guns don't kill people, people kill people, the test itself does not cause termination, but the stigmas, misinformation, and pressures put on by trusted doctors is what is bad.	4 (12 %)
Test gives information	I think the tests are good in that people born with [Down syndrome] can often have heart problems, etc. that need to be looked out for and addressed immediately. These tests could possibly be advantageous in addressing these health issues.	4 (12 %)

*Thirty-three participants provided written comments that were classified into multiple themes

overwhelming majority of respondents felt NIPT would ultimately lead to an increase in termination of pregnancies with Down syndrome (88 %), and a decrease in social services for children with Down syndrome (64 %). However, only a minority of respondents felt that the mere availability of NIPT would be the primary factor leading to termination of pregnancies diagnosed as having Down syndrome (16 %). This perceived contradiction in our data can be explained in part by examining the qualitative responses of participants. A moderate percentage of the 33 respondents who expressed free comments suggested that prenatal testing of all types supported a patient's autonomy and provided families with valuable time to prepare for the birth of a child with Down syndrome, but also that these advances in prenatal testing were only useful if the testing was accompanied by accurate, complete, and unbiased information about Down syndrome.

Receiving information they retrospectively considered to be biased or overly negative about Down syndrome (including incorrect life expectancy information, and only information related to medical problems and birth defects), was a theme

commonly expressed by participants in this study, and is consistent with reports by parents in other studies (Hodgson et al. 2010; Roberts et al. 2002). This issue raises interesting questions about how and when parents make decisions in the face of abnormal prenatal diagnosis, and the role of pre-test and post-results genetic counseling, education and resource provision in facilitating those decisions. A study of women who had continued a pregnancy after a diagnosis of Down syndrome (Hurford et al. 2013) found that only 33 % went into their pregnancy knowing that they would not terminate a pregnancy for any reason. Of the remaining 67 %, who assumedly had not yet made up their minds about pregnancy termination, only 8 % of respondents in Hurford et al. (2013) identified the post-test genetic counseling they received (from unspecified healthcare providers) as the primary factor in determining whether or not they would continue the pregnancy. Instead, the majority of Hurford et al.'s sample of women who continued their pregnancy after a diagnosis of Down syndrome chose "moral beliefs," as having the "greatest impact" on their decision. We understand that studies are currently underway assessing the

needs and experiences of families who have both continued and terminated affected pregnancies (Sheets and Crissman, personal communication).

The assertion that medical professionals provide biased information about Down syndrome prenatally is not new, but there are little data objectively assessing what information is communicated in a prenatal setting after a diagnosis of Down syndrome is made. Respondents in this study did not indicate what types of healthcare providers had provided them with information during their prenatal diagnostic process, nor did we review any medical records or transcripts of these information sessions as they occurred in the past and this was beyond the scope of the current project. An analysis of standardized patient pre-test genetic counseling session transcripts by Farrelly et al. (2010) found 95 % of counselors described physical aspects of disability, and only 27 % discussed social aspects. A series of workshops conducted by Hodgson and Weil (2012) investigated the obstacles that genetic counselors reported when discussing the nature of Down syndrome in a prenatal setting. Counselors reported lack of confidence in their own skills, a lack of knowledge regarding parenting a child with a disability, and fear of overwhelming the patient as impacting the nature of discussion about Down syndrome throughout the prenatal testing process.

The National Society of Genetic Counselors (NSGC) acknowledged the challenging role genetic counselors have in attempting to balance support for clients' autonomy and reproductive freedoms while still advocating for patients with disabilities and their families by correcting misconceptions and educating patients about what life might be like living with a child who has Down syndrome. The organization identified a range of position statements, educational opportunities (e.g., NSGC Annual Education Conference), publications and scholarship, public policy efforts, and member activities and recognition towards that end (Dent et al. 2011). Some groups of genetic counselors support further efforts in this area. For example, Madeo et al. (2011) suggested that in order to respect the rights of individuals with disabilities, genetic counselors must actively pursue engagement with disability groups, training programs should integrate exposure to individuals with disabilities into their curricula. Programs currently exist that allow medical professions, including genetic counselors, to better understand the lived experience of individuals with Down syndrome. These programs include *Operation House Call*, in which medical professionals spend time at the home of an individual with Down syndrome.

In addition to seeking out experiences to further their knowledge of what is like to parent a child with Down syndrome, counselors must also acknowledge their own limitations, and they should be knowledgeable of appropriate

resources to which to refer their patients. Some of these include, but are not limited to, Downsyndromepregnancy.org, and "Understanding a Down Syndrome Diagnosis," available as a free e-booklet via lettercase.org, and the "First Call" program provided by many local and state Down syndrome groups. Counselors should also be familiar with the "NSGC Practice Guidelines for Communicating a Prenatal and Postnatal Diagnosis of Down Syndrome" (Sheets et al. 2011).

Further research should examine what portion of prenatal genetic counseling sessions is spent discussing medical/procedural aspects of prenatal testing for Down syndrome, and how much time is spent exploring family values and the impact the uptake of such testing would have on an individual patient. Health care professionals of all types have a responsibility to present testing in a balanced manner, being respectful of patient views about whether or not they want prenatal testing and what options they might select if faced with a prenatal diagnosis of Down syndrome.

Study Limitations

There are potential biases in using support groups for recruitment, including the possibility these groups do not represent the views of other families with children who have Down syndrome and that the individuals who join such groups are self-selecting and may represent those with the strongest views. A considerable minority (28 %) of respondents in our study felt that the only purpose of noninvasive tests for Down syndrome was to enable people to terminate pregnancies affected with Down syndrome. These individuals are likely a select population, as they were significantly more likely to report not having had prenatal testing for Down syndrome in the past, and that they would not pursue prenatal testing in a future pregnancy. The sample size was small and consisted only of mothers, thus further limiting generalizability of the findings. The recruitment methods did not allow for the accurate ascertainment of response rate, and the authors were not in control of to whom individuals sent the survey link once it had been posted on the message boards, or received as an e-mail. The description of NIPT given to participants defined NIPT as nearly diagnostic, and not as highly sensitive screening, which could also have influenced participant responses. Additionally, the sample was largely Caucasian, and very well educated. Further studies should be done addressing a more ethnically and socio-economically diverse sample of mothers and fathers of children with Down syndrome. Finally, given that many of our participants discussed the perception that post-diagnosis information is biased, and that little information exists to quantify these conversations, we encourage empirical research in this area to the extent that is possible.

Conclusions

This study shows that while many mothers of children with Down syndrome would consider using or recommending NIPT in a pregnancy, the vast majority perceived this new technology will lead to increased terminations of affected pregnancies. This assessment is based at least in part on the assumption that the tone and content of information provided to individuals at the time of prenatal diagnosis influences termination of pregnancy. In recognition of this, and consistent with the goal of supporting patient autonomy around any and all prenatal decisions, this necessitates that all healthcare providers who discuss prenatal diagnosis results, should strive to provide a balanced view, recognizing both the positive and challenging aspects of raising a child with Down syndrome or any genetic condition. This type of service provision should include a balanced, up-to-date and accurate discussion describing both the medical and social aspects of Down syndrome, recognizing one's own biases, and offering resources to patients that can help them.

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References

- American College of Obstetricians and Gynecologists (ACOG). (2012). Committee Opinion No. 545. Noninvasive prenatal testing for fetal aneuploidy. *Obstetrics and Gynecology*, *120*(6), 1532–1534.
- Benn, P., Borrell, A., Cuckle, H., Dugoff, L., Gross, S., Johnson, J., et al. (2012a). Prenatal detection of Down Syndrome using massively parallel sequencing (MPS): a rapid response statement from a committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenatal Diagnosis*, *32*, 1–2.
- Benn, P., Cuckle, H., & Pergament, E. (2012b). Non-invasive prenatal diagnosis for Down syndrome: the paradigm will shift, but slowly. *Ultrasound in Obstetrics & Gynecology*, *39*(2), 127–130.
- Bianchi, D. W., Platt, L. D., Goldberg, J. D., Abuhamad, A. Z., Sehnert, A. J., & Rava, R. P. (2012). Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. *Obstetrics and Gynecology*, *119*(5), 1–13.
- Bryant, L., Hewison, J., & Green, J. (2005). Attitudes towards prenatal diagnosis and termination in women who have a sibling with Down's syndrome. *Journal of Reproductive and Infant Psychology*, *23*, 181–198.
- Chiu R. W., Chan K. C., Gao Y., Lau V. Y., Zheng W., Leung T. Y., et al. (2008). Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. *Proceedings of the National Academy of Sciences of the United States of America* *105*, 20458–20463. pubmed id: 24327983.
- De Jong, A., Dondorp, W. J., de Die-Smulders, C. E. M., et al. (2010). Non-invasive prenatal testing: ethical issues explored. *European Journal of Human Genetics*, *18*, 272–277.
- Dent, K. M., Harper, C., Kearney, L., Lieber, C., & Finucane, B. (2011). Embracing the unique role of genetic counselors: response to the commentary by Madeo et al. *American Journal of Medical Genetics Part A*, *155*, 1791–1793.
- Devers, P. L., Cronister, A., Ormond, K. E., Facio, F., Brasington, C. K., & Flodman, P. (2013). Noninvasive prenatal testing/noninvasive prenatal diagnosis: the position of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, *22*, 291–295.
- Fan, H. C., Blumenfeld, Y. J., Chitkara, U., Hudgins, L., & Quake, S. R. (2008). Noninvasive diagnosis of fetal aneuploidy by shotgun sequencing DNA from maternal blood. *Proceedings of the National Academy of Sciences of the United States of America*, *105*(42), 16266–16271.
- Farrelly, E., Cho, M. K., Erby, L., Roter, D., Stenzel, A., & Ormond, K. E. (2010). Genetic counselors and prenatal testing: where is the discussion about disability? *Journal of Genetic Counseling*, *19*, 671.
- Greely, H. T. (2011). Get ready for the flood of fetal gene screening. *Nature*, *469*, 289–291.
- Gregg, A., Gross, S., Best, R., Monaghan, K., Bajaj, K., Skotko, B., et al. (2013). ACMG statement on noninvasive prenatal screening for fetal aneuploidy. *Genetics in Medicine*, *15*, 395–398.
- Hodgson, J., & Weil, J. (2012). Talking about disability in prenatal genetic counseling. *Journal of Genetic Counseling*, *21*(1), 24–26.
- Hodgson, J., Gillam, L., Sahhar, M., & Metcalfe, S. (2010). “Testing times, challenging choices”: an Australian study of prenatal genetic counseling. *Journal of Genetic Counseling*, *19*, 22–37.
- Hurford, E., Hawkins, A., Hudgins, L., & Taylor, J. (2013). The decision to continue a pregnancy affected by Down syndrome: timing of decision and satisfaction of receiving a prenatal diagnosis. *Journal of Genetic Counseling*. doi:10.1007/s10897-013-9590-6.
- Inglis, A., Hippman, C., & Austin, J. (2012). Prenatal testing for down syndrome: the perspectives of parents of individuals with down syndrome. *American Journal of Medical Genetics Part A*, *158A*, 743–750.
- Kelly, S.E., Farrimond, H.R. (2011). Non-invasive prenatal diagnosis: A study of public attitudes. *Public Health Genomics*, in press.
- Lo, Y. M., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W., et al. (1997). Presence of fetal DNA in maternal plasma and serum. *Lancet*, *350*(9076), 485–487.
- Madeo, A. C., Biesecker, B. B., Brasington, C., Erby, L. H., & Peters, K. F. (2011). The relationship between the genetic counseling profession and the disability community: a commentary. *American Journal of Medical Genetics*, *155*, 1777–1785.
- Nakata, N., Wang, Y., & Bhatt, S. (2010). Trends in prenatal screening and diagnostic testing among women referred for advanced maternal age. *Prenatal Diagnosis*, *30*, 198–206.
- Palomaki, G. E., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., Ehrich, M., et al. (2011). DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine*, *13*(11), 913–920.
- Palomaki, G. E., Decui, C., Kloza, E. M., Lambert-Messerlian, G. M., Haddow, J. E., Neveux, L. M., et al. (2012). DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. *Genetics in Medicine*, *14*(3), 296–305.
- Press, N., & Browner, C. H. (1998). Characteristics of women who refuse an offer of prenatal diagnosis: data from the California maternal serum alpha-fetoprotein experience. *American Journal of Medical Genetics*, *78*, 433–445.

- Roberts, C. D., Stough, L. D., & Parrish, L. H. (2002). The role of genetic counseling in the elective termination of pregnancies involving fetuses with disabilities. *Journal of Special Education, 36*(1), 48–55.
- Sheets, K. B., Crissman, B. G., Feist, C. D., Sell, S. L., Johnson, L. R., Donahue, K. C., et al. (2011). Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: Recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling, 20*, 432–441.
- Skotko, B. G. (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? *Archives of Disease in Childhood, 94*(11), 823–826.
- Tischler, R., Hudgins, L., Blumenfeld, Y. J., Greely, H. T., & Ormond, K. E. (2011). *Prenatal Diagnosis, 31*(13), 1292–1299.
- Yotsumoto, J., Sekizawa, A., Koide, K., Purwosunu, Y., Ichizuka, K., Matsuoka, R., et al. (2012). Attitudes toward non-invasive prenatal diagnosis among pregnant women and health professionals in Japan. *Prenatal Diagnosis, 32*, 674–679.