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"This Lifetime Commitment": Public Conceptions of Disability and Noninvasive Prenatal Genetic Screening

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Abstract

Recently, new noninvasive prenatal genetic screening technologies for Down syndrome and other genetic conditions have become commercially available. Unique characteristics of these screening tests have reignited long-standing concerns about prenatal testing for intellectual and developmental disabilities. We conducted a web-based survey of a sample of the US public to examine how attitudes towards disability inform views of prenatal testing in the context of these rapidly advancing prenatal genetic screening technologies. Regardless of opinion toward disability, the majority of respondents supported both the availability of screening and the decision to continue a pregnancy positive for aneuploidy. Individuals rationalized their support with various conceptions of disability; complications of the expressivist argument and other concerns from the disability literature were manifested in many responses analyzed.

Keywords

Prenatal testing; Cell-free DNA; Noninvasive prenatal screening; NIPS; Down syndrome; Trisomy; Intellectual disabilities; Disability rights; Public opinion

INTRODUCTION

In the decades since prenatal screening and diagnosis were first introduced to clinical practice, concerns have emerged about their implications for the disability community in the United States (US) and abroad [Chandler & Smith, 1998; Miller & Levine, 2013; Newson, 2008; Parens & Asch, 1999; Rapp, 1998; Scott, 2005]. Many of these concerns have been raised by parents [Kellogg et al., 2014] and advocates for individuals with trisomy 21 (Down syndrome). Down syndrome is the most common trisomy, and one for which genetic screening is broadly available in the US. Since the clinical introduction of prenatal genetic

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testing, a majority of women who receive a prenatal diagnosis of Down syndrome have chosen to terminate the pregnancy, resulting in a gap between affected pregnancies and actual live births of individuals with Down syndrome [de Graaf, Buckley & Skotko, 2015; Egan et al., 2004; Khoshnood et al., 2004; Lin et al., 2013; Lai et al., 2002; Méndez-Rosado et al., 2014; Natoli et al., 2012]. Responses to a self-perception survey by Skotko, Levine & Goldstein [2011a] indicate that some individuals with Down syndrome have an awareness of prenatal genetic screening, and possibly their own concerns about termination of affected pregnancies. When asked about their recommendations for new parents, some individuals with Down syndrome "directly encouraged parents to continue their pregnancies (e.g., 'Take the baby home'; 'They should keep their kid. Don't abort' …)." Disability rights advocates have warned in the past that encouraging screening for Down syndrome implies a lack of value for individuals with disabilities, and may obstruct the broader growth of social acceptance and material support for people with disabilities [Parens & Asch, 1999; Skotko, 2009]. This theme is in a complicated historical dialogue with social ideas of 'good' parenthood, as elucidated by sociocultural research on the experience of pregnancy and the 'genetic responsibility' parents feel to produce a 'healthy' child [Rapp, 1998; Raspberry & Skinner, 2011; Rapp, 1999; Charo & Rothenberg, 1994; Rothman, 1993].

Noninvasive prenatal genetic screening (NIPS) for Down syndrome and other aneuploidies, using cell-free DNA (cfDNA) found in maternal serum, are now available in the US and abroad [Agarwal et al., 2013; Chandrasekharan et al., 2014]. NIPS offers increased sensitivity and specificity over Down syndrome serum screening, offering near-diagnostic results without a test-related risk of miscarriage [Bianchi et al., 2012; Nicolaides et al., 2012; Norton et al., 2012; Norwitz & Levy, 2013; Palomaki et al., 2012; Zimmermann et al., 2012]. NIPS has been endorsed as a screening test for high-risk pregnancies by multiple professional societies [ACOG Committee on Genetics and SMFM Publications Committee, 2015; Benn et al., 2015; Devers et al., 2013]. Clinical studies conducted in 2012 and 2013 found that NIPS improved upon the accuracy of standard screening for trisomies 21 and 18 even in low-risk pregnancies [Bianchi et al., 2014; Norton et al., 2015], and the most recent position statement from the International Society for Prenatal Diagnosis endorses NIPS for low-risk pregnancies as well [Benn et al., 2015]. A majority of US obstetricians, surveyed in late 2012, expected to offer NIPS clinically within a year [Musci et al., 2013], and rapid patient uptake has already reduced the use of invasive testing at some prenatal clinics [Beamon et al., 2014; Chetty, Garabedian, & Norton, 2013; Larion et al., 2014; Pettit et al., 2014]. In the UK and The Netherlands [Hill et al., 2013; van Schendel et al., 2014], patients and clinicians have also reported positive perceptions of NIPS and, as of March 2014, it was marketed in 61 countries worldwide [Chandrasekharan et al., 2014]. In 2012, the whole fetal exome and genome were sequenced noninvasively [Kitzman et al., 2012; Fan et al., 2012; Snyder et al., 2013]. With further refinement, this screening, offering huge volumes of complex genetic information, may well become a routinized part of clinical prenatal care.

The uptake of NIPS, and the expanding range of genetic conditions that can be detected prenatally, is rekindling ethical challenges around the value and social integration of individuals with disabilities [Van Riper, 2012]. Existing concerns about prenatal screening are exacerbated by worries that the technical advantages of noninvasive screening will cause an increase in the volume of screening and an absolute increase in the detection of true

positives and the selective termination of more affected pregnancies [Tischler et al., 2011; Madeo et al., 2011; Kellogg et al., 2014]. Previous research has addressed the attitudes of the general public toward prenatal genetic testing and screening [Farrimond & Kelly, 2013; Kelly & Farrimond, 2012; Sayres et al., 2014; Allyse et al., 2014] and their possible implications for the disability community [Steinbock, 2000; Saxton, 2000; Asch, 1999]. Researchers have found correlations between uptake of prenatal screening or testing and the views pregnant women have of disability [Bryant, Hewison, & Green, 2005; García, Timmermans, & van Leeuwen, 2011; Reid et al., 2009]. However, there is little available information about the relationship between public perceptions of disability and attitudes towards NIPS.

This qualitative analysis examines how attitudes towards disability interact with views of prenatal screening in the context of rapidly advancing prenatal genetic screening technologies. Our results highlight the nuanced ways in which individuals rationalize and qualify recommendations to accept or refuse screening, and provide a platform for discussion of how the practical and ethical concerns of the disability community are reflected in public attitudes. We compare our results to previous studies of prenatal technologies, such as serum screens or amniocentesis, and how they appeared to be related to views on disability and the offering of prenatal screening. We close by addressing our results in light of three disability critiques of prenatal genetic testing and screening: the *expressivist* argument that prenatal testing denigrates those with disabilities [Parens & Asch, 1999; Asch, 2000]; the *synecdoche* argument that prenatal testing allows one characteristic to stand in for a whole person [Parens & Asch, 1999; Asch & Wasserman, 2005]; and the argument that prenatal testing may result in apathy and hostility toward those with disabilities [Parens & Asch, 1999]. The views expressed by our study participants offer a glimpse into the ways that opinions about prenatal screening are entangled with beliefs about the inherent worth and potential burdens of a child with disabilities, and add nuance and complexity to concerns about prenatal screening and testing voiced by the disability community. After examining these findings in detail, we discuss implications for future research.

MATERIALS AND METHODS

We developed an online survey platform, called INtegrating Values and Ethics and Science and Technology (InVEST), to encourage public engagement with ethical issues surrounding emergent technologies. The InVEST platform is based on the practice of constructive technology assessment (CTA), which “aims to produce better technology in a better society, and emphasizes the early involvement of a broad array of actors to facilitate social learning about technology and potential impacts” [Genus, 2006]. The goal of CTA studies is to assess the factors that might impede or facilitate the successful, ethical translation of emerging technologies by involving broader samples of stakeholders in the technology assessment process. Scenario development is an established method of enrolling stakeholders in CTA studies [Rip & Kulve, 2008]. This pilot study represents the first research and public engagement project using the InVEST platform.

A scenario [Appendix 1] was designed to mimic the clinical experience of a 40-year-old pregnant patient (named “Cindy”) who was presented with multiple prenatal screening options for trisomies 13, 18 and 21. Participants were asked to read the scenario and respond to open-ended questions about their recommendations for the patient’s best course of action. Participants then saw a diverse series of alternative recommendations from previous surveys and interviews with prenatal screening stakeholders. After reading the statements, participants were asked if any of the opinions presented caused them to change their previous recommendation. This study was reviewed by the Stanford University Institutional Review Board.

Data Collection and Analysis

We contracted a commercial online survey provider (Zoomerang, now SurveyMonkey) to procure a random sample of 200 adult (over 18) US residents from a nationwide panel of individuals who have internet access and have agreed to participate in online surveys in return for modest compensation. Participants in the panel are recruited to represent the overall US population by gender, income and education. The contractor validated prospective participants to ensure that demographic information was correct and that there were no duplicate responses; respondents were compensated by the contractor, and participation was anonymous. Responses were downloaded from a secure webpage and stored on a secured spreadsheet for analysis.

The InVEST platform yielded both closed- and open-ended responses; the majority of responses including some discussion of disability were open-ended answers to Part 2 of the survey, which asked (among other things) “What do you think Cindy should do? … What are the most important things for Cindy to think about when she is making this decision?” Participants were also asked to describe whether they were personally affected by NIPS and to provide their professional history or occupation (see Appendix I). These data were use to determine stakeholder status.

We analyzed the open-ended responses qualitatively, using an iterative approach to identify common themes. A team of four coders independently derived a list of prominent themes and then compared results to establish the codebook. The final codebook included 9 broad codes indicating a respondent’s personal experience, screening recommendations, justification for that recommendation, and other common elements of the survey responses. Each of these codes was divided into a set of sub-codes for finer-grained classification. For example, the code “Decision-Making Factors” encompassed 13 subcodes to track respondents’ rationales in making a recommendation. Each individual’s response was coded to one or multiple relevant codes and/or subcodes, and each respondent’s answers were coded in duplicate by a two-person team and reviewed by all four coders until consensus was achieved. The results described here were taken from a set of six subcodes that together provide an inclusive picture of attitudes toward disability in the sample (Table I). The InVEST scenario describes only trisomy 13, 18 and 21, so a participant’s definition of “disability” during the study may have been narrowed to these or similar genetic conditions. During coding, we broadly defined “disability” to include any participant’s use of the word “disability” or a clear alternative description (e.g. a set of “differences,” “challenges” or

"special needs"), or any explicit mention of or reference to the genetic disorders described in the InVEST scenario (e.g. "these conditions"). In many cases, a response included multiple value statements about disability and was given more than one code. Some responses given the subcode "Life Has Value" were left out of this analysis because they did not include any explicit mention of disability or people with disabilities.

RESULTS

Participants

Table II shows participant demographics. In total, 217 participants completed the questionnaire, and 84 participants (38.7%) invoked a conception of disability in their response. The analysis below examines the views of those 84 respondents. Participants represented a variety of stakeholder relationships, including medical professionals, prospective parents and members of the disability community as well as individuals with no apparent connection to prenatal screening. Quotes from participant responses have been lightly edited for readability, but natural language is preserved where possible.

Support of NIPS and Prenatal Testing

A majority of respondents supported the use of NIPS, either alone or in combination with another prenatal genetic test (65.0% of the 217 InVEST participants, and 71.4% of the subset of 84 participants who invoked disability in their rationale, analyzed in more detail below). NIPS appears to have been understood as a separate technology, evidenced by the rationale that participants gave in their responses (e.g. increased accuracy), by the separation of NIPS from amniocentesis, and by the greater than fivefold frequency of recommendations for NIPS alone compared to either serum screening or amniocentesis alone. However, many respondents grouped prenatal tests together in their recommendations: 32.2% of InVEST participants and 36.9% of the 84-participant disability subset recommended either all tests or no tests, suggesting that beliefs about prenatal genetic testing and screening are polarized around the acquisition of information, not around the specific tests involved.

Conceptions of disability

Although disability is inextricably linked to prenatal genetic screening, the majority of InVEST participants (61.3%) did not include any general opinion of disability or people with disabilities in their response. When disability was not invoked, respondents justifying a decision to test often cited the importance of the patient's autonomy, or the belief that more health information was inherently valuable ("I believe in having as much information as possible," Female, 73). Participants who opposed the availability of screening most commonly gave religious justification ("I am a god fearing person. This kind of testing opens up the door for the murder of babies just because they are different," Male, 25). Responses incorporating a view of disability followed two central themes: one of disability or individuals with disabilities as *burdensome*, and one of individuals with disabilities as *valuable*. These views were not mutually exclusive; a few participants expressed both themes in their responses.

The Burden of Disability—A majority of respondents who chose to discuss disability either presented it as exclusively burdensome, or gave greater weight to the theme of burden while affirming the value or potential of individuals with disability. Participants in this theme expressed concern that quality of life would be negatively affected by the birth of a child with disabilities. Three distinct subjects were identified as the primary bearers of this perceived hardship: the *individual*, the *family*, and *society*.

Overwhelmingly, individuals who described disability as burdensome supported the availability and/or use of prenatal genetic screening in some form; whether and in what ways this support was conditional depended primarily upon the perceived bearers of hardship.

On the Individual: Respondents who presented disability as a burden on the affected child expressed concerns that a child born with disabilities may be disadvantaged by biological differences, by the shortcomings of their caregivers, or by social reactions to or perceptions of their condition.

> Someone born with down syndrome is going to be at a disadvantage in living a full life as compared to a person without. (Male, 30)

> Children grow up & move on to support themselves ---- a child with a disability most often cannot. So if a parent is not ready, willing or able to make this lifetime commitment, the person most likely to be hurt later is that innocent child. (Female, 48)

The belief was often expressed, both implicitly and explicitly, that a child born with certain disabilities might have a life so disadvantaged or difficult that it would not be worth living.

> I do not believe that it is right to bring a child into the world knowing it is going to have a lifetime of difficulties ahead. (Male, 24)

> What sense does it make to keep the baby if it's going to be outcast for its entire (most likely short) life? (Male, 22)

Related themes involved the idea that the preventable birth or existence of a child with disabilities may itself be a wrong or injury done onto that child by a parent choosing either to abstain from screening or to continue an affected pregnancy. The language of life with congenital disability as something "brought upon" a child by his or her parents is indicative of a perceived moral justification or imperative for prenatal screening and, in some cases, termination.

> 9 times out of 10 I see [people with disabilities] being exploited and treated poorly. I don't understand why people would want to bring this upon their children. (Male, 20)

> I have been disabled all my life and just remember how cruel the other children were with their comments. I would not want to put another child through that. (Female, 72)

Themes including burden on the child suggested a distinction between conditions "severe" enough to warrant these kind of moral judgments or imperatives and those seen as relatively

"benign." Features of a disorder that might definitively include or exclude it from these moral criteria were generally not delineated, and opinions of "acceptable" difference or disadvantage varied between respondents. Most frequently, respondents like those above, who suggested there was an imperative to test, did not differentiate between Down syndrome and the less common genetic disorders (trisomies 13 and 18) discussed in the scenario, implicitly suggesting that Down syndrome was "severe" enough to warrant these moral judgments. However, there were exceptions that reflected variations in the perceived severity of Down syndrome.

> *Cindy should take both tests* [serum screening and NIPS] *to find out all conditions of her child (if any)…If it happened that the test results came back positive for Down Syndrome, I wouldn't suggest to stop the pregnancy because the child can still live a fairly long life. If test results were positive for trisomy 13 or 18, I would suggest she and her husband discuss all the possible outcomes and make a decision based on their beliefs. (Male, 19)*

As observed here, respondents who saw disability as a burden on the affected child overwhelmingly recommended prenatal screening, and they were generally likely to do so in order to inform decisions about termination.

On the Family/Parent: The most common theme among the subset of analyzed responses was that of the child with disabilities as a negative impact on the family's quality of life. Participants expressing this theme emphasized the potential financial and/or emotional strain on the family as the foremost consequences of disability and the primary concerns in decisions about whether to opt for prenatal screening or continue an affected pregnancy.

> …I could not go through with a pregnancy knowing that the baby would have a disability that I could not financially or emotionally handle (nor could I burden my family). If someone is up to that kind of a life long challenge, I support their decision to have a child with a disability 100%. (Female, 45)

A first set of responses focused on cautionary recommendations for the decision-making individuals and primary caretakers of the child. Implicit and explicit in this theme is the view that the birth of a child with disabilities would necessitate a level of self-sacrifice on the part of the parents that cannot be morally demanded.

> Could I both financially and mentally take care of a special needs child for the rest of my (and my partners) life? For me, the decision would be no. (Female, 43)

> This is an age old decision and hard for anyone to make unless they are the mother…[Participant's daughter and step-son] had another son who has apraxia and … financially they have had no life at all as there are so many medical bills to be paid. Do they love their son? Oh yes, they do but in order for him to have a life worth living they had to give up theirs. (Female, 78)

These concerns are distinguished from those of a second group of respondents who expressed worry that a child with disabilities might unfairly burden individuals who are

affected but have more limited decision-making power, including extended family members and other children.

> It needs to be a family decision because bottom line, everyone will be impacted by the care needed for a child with such conditions. It is not something every family can handle, and in the end, the child will suffer for it. (Female, 48)

This larger theme of burden on the family included the implication that negative factors, such as potential financial and emotional strain, should be given considerable weight in decisions about whether to test for genetic abnormalities or continue an affected pregnancy. As in the theme of burden on the child, potential burden on the family was seen as moral justification for a decision to test or to terminate an affected pregnancy; however, in contrast to the theme of burden on the child, here there was little indication of a moral imperative for either. Additionally, individuals discussing this theme of disability frequently endorsed the possibility of using prenatal genetic screening to prepare for the birth of a valued child with disabilities.

On Society: A third theme, which was uncommon but nonetheless present, expressed the belief that individuals with disabilities draw disproportionately and deleteriously on public resources.

> When I plan to have kids I will want to make sure I'm bringing healthy children into this world that can grow up to be productive members to this society. I have nothing against the disabled, but I think with over population as rampant as it is and the current economic situation we're in, it's not right to bring in such a drain of resources if preventable. (Male, 20)

> Have the most reliable test. If it's positive, she and her husband should seriously consider abortion…Kids are our most important resource, but a child who will not be self-sufficient is too much of a burden on society. (Male, 70)

Frequently included in this theme is the belief that expectant parents, if they are aware that they may be unable to afford the financial responsibility of a child with additional medical and therapeutic needs, have a moral imperative to opt for prenatal genetic screening and, in some cases, to terminate an affected pregnancy.

> I believe that one needs all the facts presented before them to make an adequate and informed decision. If the tests are positive one needs to know if they can handle what lies before them in their future…I believe they should end the pregnancy unless they know someone who wants to take the responsibility and raise that child to be happy as possible and not raise them to get money from the state…This may seem callous but why should the state pay for our mistakes as parents, and breeding without regard to health issues…I just feel that people need to be able to afford the care for their children and not rely on the state to take care of disabilities. (Female, 48)

Respondents emphasized the value of screening in making decisions about termination of an affected pregnancy while taking into consideration the monetary expense of disability, and

were unlikely to include the belief that screening might be used for other purposes (e.g. to prepare intellectually or emotionally for the affected child's birth).

The Value in Disability—The second set of themes coalesced around the idea of people with disabilities as valuable or having a positive impact on families or society. Whereas participants writing about burden generally pointed to a subject or bearer of this burden, respondents discussing value did not generally speak of individuals with disabilities as valuable to a specific subject. But most respondents did indicate a *reason* for their perceived value. These reasons ranged from the general humanity of individuals with disabilities to unique characteristics or contributions they offer. These reasons have been divided between two themes that capture incidences of this positive theme in the data: expressions of the value of *all life*, and expressions of the specific value of *individuals with disabilities*.

All Life Has Value: The most common incidence of a positive or value theme was associated with the opinion that all life is valuable, or that parents should unconditionally love any child. Frequently implicit in this theme was the belief that prenatal genetic screening is generally undergone for the purpose of terminating any pregnancies affected by a disabling condition, and generally explicit in this theme was the opinion that abortion of an affected pregnancy is morally impermissible.

> Knowing the results might give you some degree of peace. However, It also could force you into making a decision which you will regret…This baby is a person and has the right to life…[God] created them and they are beautiful in his eyes. (Female, 79)

> [It's] very offensive that anyone would think you must automatically terminate the pregnancy [if the screening result is positive]…Down syndrome people are people too…accept the child as is, unconditionally, and love him or her. (Male, 23)

Still, there was a divide among respondents regarding the use of prenatal genetic screening: some declared the tests unnecessary because they were either only valuable as tools for decisions about termination or because it is not possible to prepare for a child with disabilities. Others, however, saw value in prenatal genetic tests as tools to allow potential parents to prepare for the birth and life of a valued child with disabilities.

> It honestly is up to the mother to decide. My thoughts were that no matter what the test says I am going to keep this child and love this child no matter what, so I thought it was pointless to get checked. (Female, 23)

> *Cindy should have the test that gets results for everything…[*but*] I would never abort because of a baby's projected disabilities. We all deserve a chance. (Female, 73)*

> She should certainly have both tests done [serum screening and NIPS]…I believe that if there is life there is purpose. All children are special needs. No two are alike… The most important issue is how to best prepare to enable this child to have the best possible educational experience and life experience. (Female, 63)

It was at times difficult to distinguish responses describing people with disabilities as valuable for their humanity from those intended solely to reject abortion as morally impermissible (such opinions do not necessarily reflect any attitude about disability). However, the majority of individuals who indicated that people with disabilities have value because all people have value supported a decision to undergo prenatal genetic screening or testing.

Individuals with Disabilities Have Value: A second positive theme specifically described people with disabilities as valuable and/or having unique characteristics of value to others. Responses within this theme typically focused on Down syndrome, and generally portrayed individuals with Down syndrome as able to lead happy lives and contribute to the well-being of those around them.

> We have several friends with Down Syndrome and they are wonderful children who are happy and productive. (Male, 51)

> I have had friends with down syndrome kids. One of them was severe. For two years that I knew that kid he was happy and affectionate. Could not communicate at all but smiled and hugged everyone. (Male, 42)

> I don't agree with those that think she should have the test and then get an abortion…A baby with down syndrome can lead a great life. (Male, 25)

Although this theme most commonly emphasized the emotional value of individuals with disabilities, some individuals also expressed a belief that individuals with Down syndrome specifically are able to be independent and make material contributions to society.

> If she gets a positive then she can start preparing, as well as educating herself in how to care and what to expect having a child that has DS…DS children can grow into healthy functioning adults. (Female, 39)

Beliefs about screening were variable in this population. As in the theme of "all life as valuable," individuals who expressed that people with disabilities specifically had value largely supported screening, although they were less likely to do so than respondents with negative views of disability. Like those who felt "all life has value," these respondents frequently qualified recommendations for prenatal genetic screening with the stipulation that screening only be used to gather information and not to terminate a pregnancy; however, this view was not universal, and a number of participants expressed a belief in parental autonomy with regard to decisions about screening and termination.

Participants Reporting Experience with Disability—A small but intriguing subset of respondents reported having experience with disability, either first-hand or through family, friends or a profession.

> I am a retired teacher of special needs children…. If her child is a special needs child [Cindy] will need to know how she and her family must prepare for the homecoming of this tiny life. (Female, 63)

> It's not easy it's a lot of work. But [participant's son] has a good life that god gave him…I have two special needs children and want people to know that a test will help to prepare you for your journey. (Female, 41)

Few individuals in this small subset expressed a strictly negative view of disability; respondents generally discussed either a combination of positive and negative experiences or expressed only a positive view of disability and individuals with disabilities. However, these individuals almost unanimously supported prenatal screening and most explicitly recommend screening as a tool to prepare for the birth of a child with disabilities.

DISCUSSION

Regardless of their attitude toward disability, respondents supported the availability of both invasive tests and noninvasive prenatal screening for use by potential parents. However, individuals who expressed opposing themes of disability outlined different moral provisos for the use of information acquired from screening.

Noninvasive prenatal screening was portrayed in the survey scenario as more sensitive and specific than serum screening and without the test-related risk of miscarriage posed by amniocentesis. Many respondents, both those who reported that they would consider terminating an affected pregnancy and those who emphasized the value of such information for preparing for the birth of a wanted child with disabilities, recommended NIPS because of its perceived definitiveness and saw the value of increasingly reliable information in making health care decisions. However, when expressing views toward disability, participants frequently lumped NIPS and serum screening together, often failing to discuss the risk of miscarriage at all. In this context, it appears that *any* prenatal information is either seen as a valuable asset in preparing for the future or seen as threatening by those who oppose screening on the grounds that children with disabilities have worth and should be welcomed; it is the *desire* for prenatal information, as an aid to abortion decisions, which is seen as threatening — not merely the information itself, however sensitive and specific. Individual themes of disability seemed to correlate strongly with assessments of the value or role of prenatal screening.

The Expressivist Argument

Prenatal genetic screening was developed and introduced in the clinical setting as a preventative screen, and this is the primary manner in which it is described and marketed to patients [Browner & Press, 1995; Milunsky, 2004]. This history provides a context for the inherent weight of the *expressivist* argument as outlined by Parens and Asch [1999], which maintains that any prenatal screening for disabling conditions "[disparages] the lives of existing and future disabled people by trying to screen for and prevent the birth of babies with their characteristics" [Asch, 2000]. Previous studies of public attitudes towards prenatal testing have seemed to support this critique. Singer [1991], for instance, found that 39.1% of respondents felt that everyone should test for genetic defects in the fetus and 22.2% said that everyone should have an abortion if the test was positive. A decade later, Kalfoglou et al. [2004] found that half of survey respondents agreed with the statement, "Parents ought to do

everything technologically possible to prevent their child from suffering including using reproductive genetic technologies."

The results of our study suggest a more complex range of perceptions of disability and the role of prenatal screening among the general public. Among respondents included in this analysis, the only individuals to imply an imperative for screening or termination described disability as a burden falling either on an affected child or on society. These were also the only respondents who could be described as actively seeking to prevent the birth of children with disabilities. As discussed in the results, respondents only applied these moral imperatives to conditions perceived as relatively "severe," though individual opinions about the severity of genetic disorders varied. The relative absence of explicit bias in this sample does not rule out the presence of unstated or unconscious (implicit) bias in a larger segment of the public; in one example, a study of French adults and professional caregivers revealed implicit stigmatization of individuals with Down syndrome based on the degree of conformity of their facial features to stereotyped features of the disorder [Enea-Drapeau, Carlier & Huguet; 2012]. If implicit negative views are held by significantly more individuals than suggested by these responses, they appear at least to be joined by explicit supportive opinions of individuals with disabilities and their families.

A majority of responses and recommendations suggested that screening merely be made available for those families who might use it to consider termination. In these responses, it was not the general quality of life or characteristics of a child with disabilities that were described as undesirable. Rather, respondents were sensitive to the anticipated hardship that a specific family may experience in addressing additional care necessitated by a disability. Many felt unable or unwilling to assign greater value to either the birth of a child with disabilities or the preservation of a family's current emotional and financial well-being.

The Synecdoche Argument

In the case of respondents who described disability as a burden born by the affected child, it was frequently assumed that affliction with any of the trisomies described in the scenario decreased, or even precluded, the possibility of a life worth living. Such assumptions have been described in the "synecdoche" argument, which points out that stigmatization of disability causes potential parents to mistakenly allow one trait to stand in for the whole child in evaluating prenatal testing and termination decisions [Parens & Asch, 1999; Asch, 2000; Asch & Wasserman, 2005]. Synecdochal logic, which Parens and Asch [1999] argue entails "a preoccupation with what is trivial and an ignorance of what is profound," was evidenced in the wide range of responses that cited the severity of a disability as a decision-making point. Other studies have noted that anticipated suffering of a child with disabilities plays a key role in parental decision-making with regard to prenatal screening [Kelly & Farrimond, 2012; van den Berg et al., 2008]. In the InVEST scenario, respondents generally assumed the discussed disorders to be very severe, but there is deep disagreement regarding such inherently subjective assessments [Parens & Asch, 1999]. Disparities were seen in participants' perceptions of the severity of Down syndrome in particular, suggesting variations in depth of knowledge about genetic conditions that raise ethical concerns for health care providers charged with ensuring informed consent for prenatal testing [Williams,

Alderson, & Farsides, 2002]. The observed emphasis on disorder severity is especially concerning to issues of informed consent in light of research suggesting that parental feelings of love and pride for a child with Down syndrome are “not associated with the degree of medical problems in the child with DS and not related to his or her level of functional activities” [Skotko, Levine & Goldstein, 2011b].

Nonetheless, the majority of respondents who described having a positive view of disability or of individuals with disabilities—and who generally acknowledged the “profound” personal character of the child—still recommended the availability of prenatal genetic testing and screening.

Concerns of Apathy and Hostility

Conceptions of disability as a burden have a theoretical capacity to reinforce ‘ableism’ or discrimination against individuals with disabilities on the grounds that their existence represents a liability to society that could or should have been prevented with the aid of prenatal screening technologies [Parens & Asch, 1999]. However, responses echoing this view—that individuals with disabilities should not be born because they place a burden on others—were a small minority of all those analyzed. These statements were rooted in the idea that individuals are responsible for their reproductive choices and should not “burden” the larger community (e.g., by accepting Federal aid) with their decision to continue an affected pregnancy. This view resonates strongly with notions of neoliberal “genetic citizenship” that many theorists have observed, particularly in the realm of genetic testing, in which individuals—disproportionately women—become bearers of genetic risk and its concomitant responsibilities for management of self, kin, and future generations [Raspberry & Skinner, 2011; Hallowell, 1999; Novas & Rose, 2000]. From the perspective of disability rights, the logical underpinnings of this view seem to fall in line with concerns voiced by Andrews and Hibbert [2000], among many others, that a view of genetic disability as a reproductive choice “may reduce our communal commitment to people with genetic disabilities.” Given the appearance of the *burden on society* theme in our data, these concerns remain relevant to the discussion of prenatal genetic screening; however, these more extreme “individual responsibility” or “anti-disability” viewpoints were uncommon in this study.

Several characteristics of the more popular views described in the study may still be viewed as problematic from a disability rights perspective, including assumptions that a particular genetic disorder causes an objectively inferior quality of life. However, these opinions were not characterized by antagonistic sentiments toward individuals with disabilities or toward parents who choose to continue a pregnancy after receiving a positive genetic screen. Additionally, these responses did not convey the belief that society should not be accountable for the care and good treatment of these individuals. Many respondents highlighted shortfalls in the current infrastructure of support available to individuals with disabilities and their families, suggesting an active concern for the quality of life of families experiencing disability. It appears, then, that compared to previous studies of public attitudes towards prenatal testing in the context of disability, many of the concerns regarding the social implications of disability remain in the context of newer prenatal screening

technologies. However, respondents in this study overwhelmingly indicated respect or support for the use of prenatal screening and testing to prepare for a valued child, rather than focusing on screening as an avenue toward selective termination. This suggests that the offer of NIPS should be couched in informational, rather than preventative, terms.

Study Limitations & Research Recommendations

Respondents were not specifically invited to comment on disability in the scenario provided, so the subset analyzed here may reflect a population that is disproportionately aware of disability issues. The InVEST scenario describes only trisomy 13, 18 and 21, so participant's definitions of disability were likely focused on these or similar genetic disorders. "Disability" as discussed here applies most directly to trisomy 13, 18 and 21 – still, despite this likely narrowing of the definition of "disability," individual definitions vary and we were not able to precisely determine which conditions were included in each participant's definition of disability. Because responses pertaining to disability were volunteered and anonymous, it is likely that those received were in earnest and that participants may have felt less constrained by socially acceptable opinions about disability. However, participants' self-reported opinions toward disability and recommendations for prenatal genetic screening in this hypothetical scenario may not accurately predict their views or actions after experiencing a similar scenario first-hand.

Although notable differences in opinion between genders and various age groups were not observed in this study, it is not possible to rule out the existence of differences in opinion between individuals of different demographic groupings in the general US population. Because younger individuals will ultimately be tasked with the majority of decisions about uptake of NIPS (as they become parents), it would be particularly useful to survey a larger and/or younger population and evaluate possible generational differences in views toward disability and prenatal genetic screening.

The subset of individuals who indicated experience with disability presents an additional opportunity for further study. Because there was a small number of these individuals in the current dataset, a broader sample of the opinions of members of the public who have personal experience with disability would be valuable both for general consideration in ethical discourse and in recommendations for clinical practice and patient counseling. Previous research suggests that many individuals with disabilities and their families view prenatal genetic screening or testing favorably [Kellogg et al., 2014; Bryant, Hewison, & Green, 2005; Chen & Schiffman, 2000; Skinner, Sparkman, & Bailey, 2003]. However, this group has also expressed concerns about the implications of such screening for stigmatization of people with disabilities [Kellogg et al., 2014; Kelly & Farrimond, 2012; Boardman, 2014].

CONCLUSIONS

This analysis is intended as a preliminary investigation of how the views towards disability of the US population relate to or inform opinions of prenatal testing in the context of increasing uptake of noninvasive genetic screening options. Regardless of their opinion towards disability, the majority of respondents supported the availability of NIPS, and

prenatal testing more generally. Individuals rationalized their positions with varying conceptions of disability, which sometimes challenged and sometimes bolstered existing concerns about the effect of prenatal genetic screens on the disability community. Although previous studies have seemed to support the presence of *expressivist* beliefs in the United States [Kalfoglou et al., 2004; Singer, 1991], our data suggest that the beliefs and rationale behind support for prenatal genetic screening and testing are more complex. While the potential burden of disability was acknowledged, very few participants in this study suggested that NIPS and other forms of prenatal genetic tests should be used to prevent the birth of children with disabilities. On some level, though, synecdochal concerns—that a consideration of testing might cause individuals to ignore the sum of characteristics of those with disabilities, and instead to reduce them to their disability—were manifested in many of the themes analyzed in this dataset. In many cases, these sentiments might be portrayed as misguided or ill-informed; a small subset of apparently purposefully antagonistic sentiments did not reflect the general attitude of respondents. Although it is difficult to rule out the possibility that prenatal genetic screening might negatively affect the lives of individuals with disabilities in other ways, these results give little or no indication that the availability or use of prenatal genetic screening is likely to cause members of the US population to adopt hostile opinions towards individuals with disabilities or their caretakers.

The findings of this study, relating as they do to some of the disability community's most evident and recurrent ethical concerns, are pertinent to discussions of whether public health support for prenatal screening might come at the expense of individuals with disabilities. Additionally, this study further bolsters our understanding of the current context for discussions about the ethics of prenatal genetic screening and testing and best ethical practices for informed consent.

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Appendix 1: Text of the InVEST scenario and relevant questions

Scenario

The following scenario describes one person's encounter with a new technology. Please read it and respond to the questions that follow. PLEASE NOTE that you must answer all the questions on the following pages in order for your answers to be recorded.

Cindy is a 40 year-old mother of two who has just found out that she is six weeks pregnant. Due to her age, she is at higher-than-average risk of having a pregnancy affected by several genetic conditions, including Down syndrome and trisomy 13 and 18, and her doctor recommends that she receive prenatal tests that look for these conditions.

Down syndrome, or trisomy 21, is a genetic condition that leads to learning disabilities, an increased risk for heart and other medical problems. Some individuals with Down syndrome have severe symptoms, while some have mild symptoms. Individuals with Down syndrome live an average of 60 years and can lead fulfilling lives with good health care and other resources. However, these individuals can have special needs, often have trouble supporting themselves financially, and may face social discrimination. When Down syndrome is detected in a fetus, some families choose not to continue the pregnancy. Others choose to have a child with Down syndrome, and many say they are very happy. Trisomy 13 and 18 are also genetic conditions. Approximately 50% of pregnancies with trisomy 13 or 18 result in miscarriage or stillbirth, and of babies born with trisomy 13 or 18, approximately 90% to 95% die within the first year of life. Others may survive into their teenage years but often have severe cognitive impairment, medical problems, and physical abnormalities. Although there are support groups to help families and children with trisomy 13 or 18, there are no treatments that can cure either of these conditions. When trisomy 13 or 18 is detected in a fetus, most parents choose not to continue the pregnancy. However, some do continue the pregnancy and say that they made the right choice. Cindy's doctor tells her about two screening tests that can look for these conditions:

The first screening test detects not only Down syndrome and trisomy 13 and 18 but also other serious medical conditions in the pregnancy. This test often right: it misses 15 out of 100 cases of Down syndrome and trisomy 13 and 18. This test rarely, about 5 out of 100 times, misidentifies a fetus as having Down Syndrome or trisomy 13 or 18 when it really doesn't have it. To have this test, Cindy would have her blood drawn and analyzed so there would be no risk to her or the pregnancy. This test would cost Cindy about $20.

The second test is a new test that only looks for Down syndrome and trisomy 13 and 18. This test is almost always right: it misses 2 out of 100 of cases of Down syndrome and trisomy 13 and 18. This test almost never, less than 1 out of 100 times, misidentifies a fetus as having Down Syndrome or trisomy 13 or 18 when it really doesn't have it. To have this test, Cindy would have her blood drawn and analyzed so there would be no risk to her or the pregnancy. This test would cost Cindy about $250. Cindy's doctor also tells her that if one of

these screening tests were positive for Down syndrome or trisomy 13 or 18, she would have the option of receiving an amniocentesis, which is an invasive test during which a needle is inserted into the uterus. This test is always right. It finds 100 out of 100 of cases of Down syndrome and trisomy 13 and 18. Very rarely, about 1 out of 100 times, this test causes the fetus to miscarry.

Your Opinions

Now that you have read the scenario, what do you think Cindy should do? Should she have any tests? Which ones - the old test which tests for more conditions, the new one that is more accurate, or both? What should she do if she gets a “positive” result (meaning that the test indicates that the fetus has either Down Syndrome or trisomy 13 or 18)? Should she test for everything, or is it better to test only for some conditions and not others? What are the most important things for Cindy to think about when she is making this decision? To whom should she talk, if anyone? Please give as much explanation of your response as you can.

Other Opinions

[Respondents were asked to read various stakeholder opinions taken from earlier experiments.]

Response to Other Opinions

Did anything you read change your mind about what Cindy should do? With what did you agree in reading these opinions? With what did you disagree?

Follow-up

Does this technology affect you? How and why?

Demographics

1. What is your gender?
2. What is your age?
3. What is your professional background or current employment?
4. How did you hear about the INVEST forum?
5. Is there any other information you would like to share only with researchers and not publicly?

Table I

Subcodes of Responses Pertaining to Disability in the Dataset

Subcode	Example of Coded Response
Burden of Disability (*n* = 52)	"The care and cost of a child with challenges can be a burdensome against not only the mother, but the whole family as well." (Male, 37)
Value Healthy Child (*n* = 14)	"The most important thing Cindy should be thinking about now is insuring a healthy pregnancy and try to do whatever she has to to have a healthy baby." (Female, 32)
Life Has Value (*n* = 14)	"When a women decides to take the risk of getting pregnant, she assumes the risk that there may be complications. If there are, that does not discount the value of the life." (Female, 56)
Unconditional Love (*n* = 13)	"We believe if the lord wants us to have a baby with down syndrome then we will have the baby and love it like he first loved us." (Male, 28)
Value Disabled Child (*n* = 16)	"Many families have born Down Syndrome children successfully and greatly enriched their families." (Male, 81)
Experience with Disability (*n* = 18)	"I have worked with developmentally disabled children for 8 years and I know what the parents go through." (Female, 38)

Note. *n* = number of coded occurrences of each subcode among the original 217 responses

Table II

Participant Demographics

Variables	All Respondents (*n*=217)	Study Subset (*n*=84)
Mean age in years (SD)	46.1 (18.0)	44.0 (17.4)
Gender		
Male *n* (%)	109 (50.2)	39 (46.4)
Female *n* (%)	107 (49.3)	45 (53.6)
Decline to state *n* (%)	1 (0.5)	0 (0.0)
Stakeholder Group		
Offered/received NIPT		6 (7.1)
Prospective parent/grandparent		15 (17.9)
Healthcare		7 (8.3)
Payers		1 (1.2)
Disability community		11 (13.1)
None provided		44 (52.4)