

The Future of Prenatal Genetic Testing: Bioethical Reflections



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The benefit of discarding the dilemma

- Access to desired information without risking a desired pregnancy
- Discarding a dilemma that has been haunting women since invasive prenatal diagnosis was first offered
- The dilemma: weighing different types of risks - ‘comparing apples and oranges’:
 - risk of bringing an affected fetus to term, against
 - risk of losing a healthy fetus

The benefit of discarding the dilemma

- Speaking as a patient – the lived experience of this dilemma
- The agony surrounding the decision to test invasively
- The excruciating fear of losing a desired (sometimes long awaited) pregnancy

The benefit of testing earlier

- The increasing emotional and moral difficulty of terminating a pregnancy as time goes by
 - “Now I have seen this baby twice already. I have known it for 18 weeks and I don’t want to part with it. Oh, I think this test [amnio] is a boon, a real boon for older women. I’m all for it. But I wish it had come earlier.” (quoted in Rapp)
- Diagnostic information early in the first trimester could make the decision to terminate an affected pregnancy less traumatic for some

NIPD: An 'ethical imperative'

- NIPD promotes reproductive autonomy by allowing access to desired information without associated undesired risk
- NIPD will prevent devastating miscarriages
- NIPD will allow earlier testing, possibly attenuating the implications of a decision to terminate the pregnancy
- The benefits of NIPD make its integration into clinical practice ethically justified
- Moreover, ethical concerns should be addressed but they do not justify *delaying* this integration

Ethical concerns surrounding consent

- Current prenatal testing involves a two-step approach
 - non-invasive screening
 - if screening results raise concerns, invasive diagnostic testing is offered including counselling and consent
- This two-step approach provides women with some built-in protection from exposure to unwanted diagnostic information
- Currently, NIPD is presented as an additional screening tool (e.g. MaterniT21)
- However, once the need for screening is eliminated, clinical practice will probably adopt a one-step approach of going straight for NIPD

Ethical concerns surrounding consent

- Even within today's two-step approach, consent procedures for screening are not always appropriate and need enhancing
- The transition to a one-step approach might threaten consent procedures for diagnostic testing
- The risk-free nature of NIPD and its future accessibility and affordability will probably promote its integration into routine prenatal care
- ...particularly in light of the growing tendency toward the medicalization of pregnancy and pressures created by a medical system that favors testing
- The challenge: ensuring appropriate counselling and consent mechanisms and securing funding for their implementation alongside the implementation of NIPD
 - NIPD should not become 'just another blood test' offered during routine prenatal care

Painting a richer moral picture

- Practical concerns regarding appropriate consent and counseling do not cover the entire ethical landscape of reproductive autonomy
- Autonomy as expressed through the notion of informed consent has merit...
- ...however, “naïve reliance on individual-focused bioethics practices like informed consent may — by themselves, and without attention to the broader context in which individual decisions are made — be inadequate” (Marshall and Koenig)

Painting a richer moral picture

- The notion that reproductive autonomy is promoted as long as informed consent is appropriately obtained reflects a very **narrow conception of choice**
- It limits the analysis of women's choice to "their ability to accept or decline a particular option that is offered to them... [but]
- ...allows no room for reflection on the *practice* that is making those particular choices available, or...
- on other contextual influences outside the clinic that may not qualify as coercive, but may have a significant impact on women's decision-making" (Seavillekline)

Painting a richer moral picture

- The bioethical debate surrounding prenatal diagnosis has evolved beyond seeing individual consent to a specific procedure as -
 - being able to tell the full story of women's moral experience
 - being able to capture what is at stake from a social standpoint
- The simplistic view of reproductive autonomy as unequivocally served by risk-free access to information should be contextualized

Prenatal diagnosis and women's moral experience

- Barbara Katz Rothman (medical sociologist);

The Tentative Pregnancy: How Amniocentesis Changes the Experience of Motherhood (1986, 1993)

- Once a reproductive technology is widely available, it shapes not only the experience of those who choose to use it, but also the experience of those who choose not to
- The widespread use of a technology - to the point of becoming an integral routine part of prenatal care - reshapes the social construction of pregnancy and motherhood for everyone

Prenatal diagnosis and women's moral experience

- From a social perspective, sometimes new options quietly foreclose old possibilities by narrowing and structuring choices
- “As choices become available, they all too rapidly become compulsions to ‘choose’ the socially endorsed alternative”
- The example of contraception and family size

Prenatal diagnosis and women's moral experience

- Rayna Rapp (medical anthropologist);

Testing Women, Testing the Fetus: The Social Impact of Amniocentesis in America (2000)

Prenatal diagnosis and women's moral experience

- Women's moral experience surrounding prenatal testing is not prevalently focused on the 'moral status of the fetus', which has been the focus of the ethical-political abortion debate
- Rather, their moral experience is focused on the implications of their choices for others
- What are the implications of having an affected child, a child with special needs, a child that will demand more time and resources, for:
 - their existing children
 - their relationship
 - their own identity

Prenatal diagnosis and women's moral experience

- So what can we learn from past social science research on prenatal testing for the future implementation of NIPD?
- The 'choice' of whether or not to use invasive testing and whether or not to terminate a pregnancy should be understood in a wider social context
- If we want consent to be not only 'informed' but also 'voluntary' or 'free' in a meaningful sense, refusal to test must be a meaningful / socially viable alternative

Prenatal diagnosis and women's moral experience

- Currently, women and their partners already have to cope with social and medical pressures to screen and to test
- The social framing of testing is such that many women believe “testing somehow promotes the birth of a healthy child, such that a caring woman is not doing her motherly duty if she forgoes testing” (Seavilleklein)
- This framing makes refusal seem “irresponsible and irrational” and creates social pressures that actually limit the options open to some women
- The result: the possibility of testing shapes our society in a way that threatens – rather than promotes – reproductive autonomy

Prenatal diagnosis and women's moral experience

- The social construction of testing as 'responsible motherhood' exists even now when invasive testing *does* involve risk to a healthy fetus
- Will NIPD - which poses no risk - further exacerbate these current social trends?
- The challenge:
 - How can we prevent exacerbating current social pressures to test, while introducing a new technology that eliminates the risk to healthy fetuses and allows earlier termination of affected pregnancies?

What is at stake from a social standpoint?

- ‘Social pressure’ is a phenomenon that can be hard to measure and quantify
- However, there are other measurable social consequences
- The case of Down syndrome

The screenshot shows the New York Times Magazine website. The main article is titled "A Drug for Down Syndrome" by Dan Hurley, published on July 23, 2011. The article text begins: "Early in the evening of June 25, 1995, hours after the birth of his first and only child, the course of Dr. Alberto Costa's life and work took an abrupt turn. Still recovering from a traumatic delivery that required an emergency Caesarean section, Costa's wife, Daisy, lay in bed, groggy from sedation. Into their dimly lighted room at Methodist Hospital in Houston walked the clinical geneticist. He took Costa aside to deliver some unfortunate news. The baby girl, he said, appeared to have Down syndrome, the most common genetic cause..."

The article is accompanied by a photograph of Alberto Costa and his daughter, Tyche, sitting on the grass. The photo is credited to Lydia Fanos for The New York Times.

On the right side of the page, there are several widgets: a Facebook login prompt, a "What's Popular Now" section featuring "The Path Not Taken" and "Gov. Andrew M. Cuomo Calls for Same-Sex Marriage in All States", a Rolex advertisement, and a "TicketWatch: Theater Offers by E-Mail" sign-up form.

What is at stake from a social standpoint?

- Data shows that with current diagnostic testing, most pregnancies of fetuses with Down syndrome are terminated
- NIPD will probably lead to an increase in diagnosis and therefore an increase in the volume of termination of such pregnancies
- If over time NIPD will nearly decrease the population of individuals with Down syndrome –
 - will funding for research and treatment be affected?
 - will funding of social services and support systems be affected?
 - will stigmatization of the condition increase as its prevalence decreases?
- The answer to all three can too easily be ‘yes’

What is at stake from a social standpoint?

- NIPD will force *more* mothers to “judge the quality of their own fetuses” and to make “concrete and embodied decisions about the standards for entry into the human community” (Rapp)
- This underlying notion of ‘standards of entry’ creates an enormous ethical challenge regarding the social acceptance of individuals already living among us with the same conditions we are testing for
- It links reproductive autonomy with disability rights – both politically charged issues

What is at stake from a social standpoint?

- “The intersection of disability rights and reproductive rights is a busy crossroad”
- It involves two related and tension-fraught issues:
 - “the reproductive rights of women to carry or refuse to carry to term a pregnancy that would result in a baby with a serious disability”
 - “the need to support adequate, nonstigmatizing, integrative services for all the children, including disabled children, that women bear” (Rapp)
- Without such services, women’s reproductive autonomy is restricted, since their choice not to test or not to terminate might entail unbearable costs and burdens

What is at stake from a social standpoint?

- However, the ethical challenge is even deeper
- It touches not only on social services and support systems.... but also on social attitudes
- The ‘expressivist’ argument:
 - “Prenatal tests to select against disabling traits express a hurtful attitude about and send a hurtful message to people who live with those same traits”

(Parens, *The Disability Rights Critique of Prenatal Genetic Testing*)

The ‘expressivist argument’

- “By developing and offering tests to detect some characteristics and not others, the professional community is expressing the view that some characteristics, but not all, warrant the attention of prospective parents”

(Press, Assessing the Expressive Character of Prenatal Testing: The Choices Made or the Choices Made Available?)

- “Do not disparage the lives of existing and future disabled people by trying to screen for and prevent the birth of babies with their characteristics”

(Asch, Reproductive Liberty)

The ‘expressivist argument’

“The message at the heart of widespread selective abortion on the basis of prenatal diagnosis is the greatest insult: some of us are ‘too flawed’ in our very DNA to exist; we are unworthy of being born....

Fighting for this issue, our right and worthiness to be born, is the fundamental challenge to disability oppression; it underpins our most basic claim to justice and equality - we are indeed worthy of being born, worth the help and expense, and we know it!”

(Marsha Saxton, *Disability Rights and Selective Abortion*)

The 'expressivist argument'

Some responses:

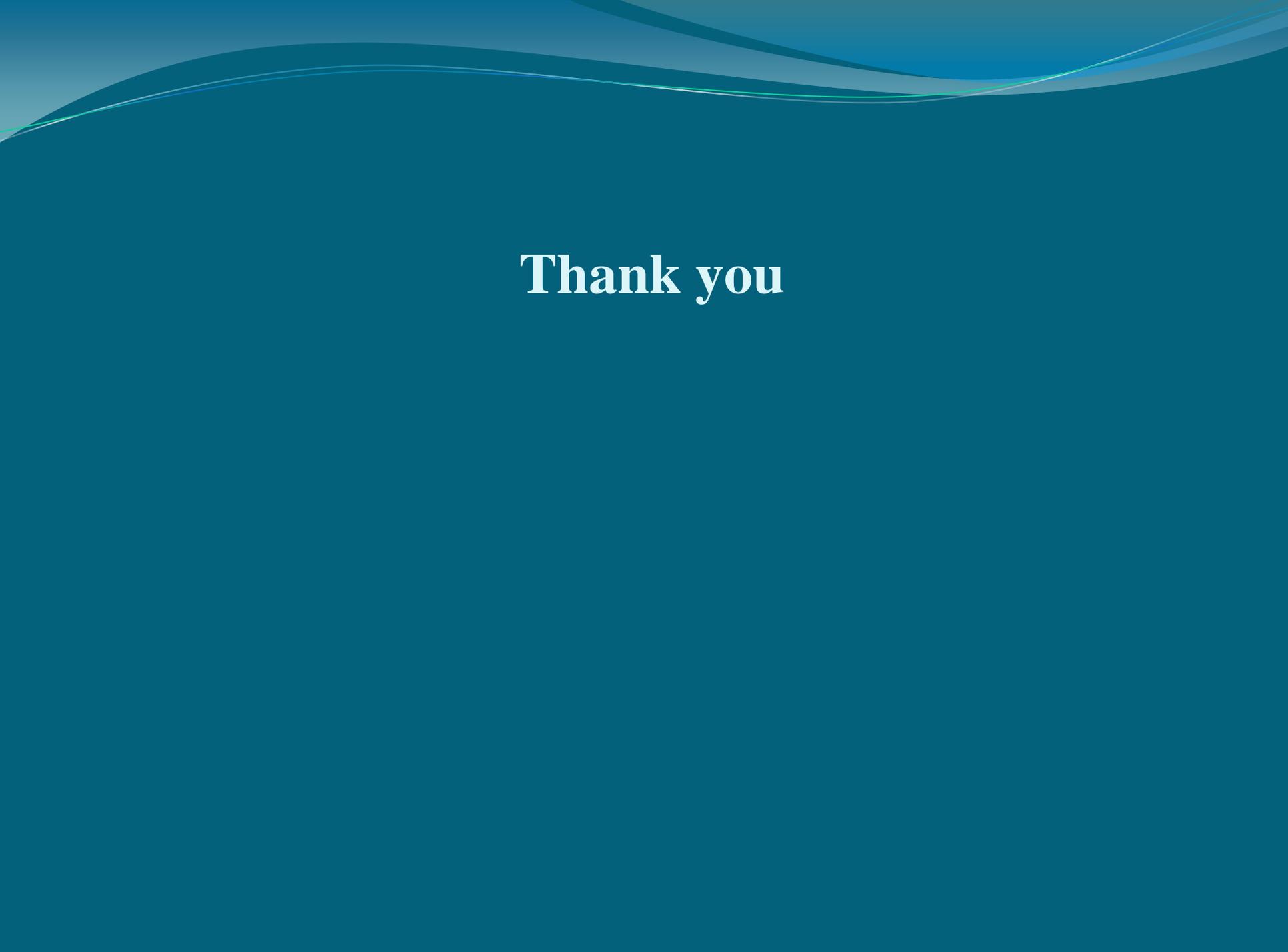
- There are numerous reasons for choosing against a disabling trait that do not entail hurtful attitude towards individuals living with that trait (e.g. parents feel they do not have the resources to cope with special needs)
- A fetus cannot be compared to a child: parents can ethically choose to terminate a pregnancy but have an ethical obligation to care and love a child born with a disabling trait

So what should we do?

- Make appropriate public policy decisions to ensure that individuals living with disabling conditions and their families are not negatively impacted by increased prenatal diagnosis
 - secure funding for research
 - secure funding for social services and support systems
 - support education to promote social acceptance and prevent stigmatization
- Provide strong arguments against the ‘expressivist’ argument
 - Demonstrate that our attitudes towards fetuses do *not* reflect our attitudes towards living individuals by promoting respect for human diversity

Conclusions

- NIPD promises tremendous benefits
- Its integration into clinical practice is ethically justified and even mandated
- At the same time, NIPD raises ethical challenges that require addressing the complex ethical landscape surrounding reproductive autonomy

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Thank you