

From Invasive to Non-invasive and Back Again: Exploring the Changing Conceptions of Disability and NIPT

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Abstract

We often hear that genomics and biotechnology drive humanity forward; unlocking the molecular secrets of life and gifting us with the tools to know more of who we are and what we want future generations to look like. Yet, genetic technologies are mired by sharp ethical and political debates. Individuals, families, and epistemic communities have reservations about their potential to overdetermine identities of disability and difference. Non-invasive prenatal testing (NIPT) detects with greater clinical accuracy certain genetic conditions during pregnancy, but this too begins to further nuance identities of disability. Indeed, as discoveries are made and boundaries change, these technologies demand scholarly analysis.

This paper investigates how NIPT shapes the changing notions of disability identities. Using reflections of risk theory and the authors' preliminary observations of NIPT in Iceland, it is argued that this method of prenatal screening exhibits invasive concepts of personhood and newfound ideas about disability.

Introduction

The social sciences provide us with a range of tools to understand the world and what it means to be human. This is extremely valuable given the monumental changes taking place in modern society, with technoscientific innovations visibly shaping how we as a species contemplate their effects. Genomics and biotechnology in particular are ongoing reminders that science and society invariably transform highly politicised concepts such as kinship (Franklin, 2003), the body (Lock, 2015, 2017), and even how we define our own history as a species (Palsson, 2015; Zwart, 2009).

NIPT is a new kind of prenatal genetic testing, said to have the potential to revolutionise prenatal care, foetal diagnostics and enhance autonomous reproductive decision-making. Whereas until recently prenatal genetic testing was only possible from invasive tests such as amniocentesis and chorionic villus sampling, NIPT stands to offer earlier and more accurate genetic testing with no risk of miscarriage and results available in a couple of weeks. While initially developed to detect extra foetal chromosomes in maternal blood using next-generation sequencing (NGS), other clinical applications have been developed and researchers speculate on its potential to screen beyond this function. However, as an 'innovative technology in transition' (Thomas et al., 2020), we need to match the speed of NIPT's rollout with research that critically investigates the manifold sociological implications and how it shapes cultural constructions of disability. To date, such a focus is lacking in the literature which tends to falsely ascribe disability as a fixed unchanging category.

This paper is a conceptual analysis of how NIPT triggers changing cultural conceptions of disability articulated through elevated discourses of genetic risk. After defining NIPT, I then reflect on the epistemic inroads afforded by risk theory to analyse changing disability identities. Finally, I draw on these insights to explicate my preliminary observations of NIPT and disability in Iceland.

Bringing NIPT into view

NIPT is “a [non-invasive] strategy of prenatal testing for analyses [of] cell-free DNA” (Vanstone et al., 2015, p.54), used primarily to detect trisomy 13 (Patau syndrome), 18 (Edwards syndrome), and 21 (Down syndrome). The test derives from a 1997 discovery that fragments of cell-free foetal DNA (cffDNA) circulates within a pregnant woman’s blood around 4 weeks (Lo et al., 1997). However, for accurate analysis a small maternal blood sample is taken between 8 to 10 weeks (de Wert et al., 2015).

Compared with other techniques – which are more readily available, widely used, and arguably regarded as a ‘normalised’ part of pregnancy – NIPT is still relatively novel, and the limits of its screening and diagnostic capabilities remain to be seen. Yet, many worldwide continue urging for its incorporation into antenatal healthcare (Brady et al., 2016; Tamminga et al., 2015; Verweij et al., 2013), with studies citing earlier and safer testing, the ability to yield significantly higher sensitivity and false positive rates for trisomy 13, 18 and 21 (Gadsbøll et al., 2020), and the potential to acquire infinite amounts of bio-information as an indication of the perceived self-evident benefits of NIPT. However, these studies are mainly conducted in fields such as medicine and bioethics, and their cultural ramifications are underwritten.

Meanwhile, the number of targeted diseases that are seemingly possible to detect with NIPT is expanding; researchers and rare genetic disease advocacy groups push to include more chromosomal conditions, monogenic disorders and microdeletions (Butler, 2017). Already, NIPT has been used to test for genetic diseases caused by single gene mutations such as cystic fibrosis (Jeppesen et al., 2021), and in the UK, NIPT recently became available on the NHS to be used alongside other tests for sex-linked conditions like Duchenne muscular dystrophy (‘Genomics Education Programme’, 2017). Some studies are also testing whether NIPT provides additional or discrepant diagnostic information for patients who conceived with PGT-A (pre-implantation genetic testing for aneuploidy) screened embryos (Harjee et al., 2020, p.156). Yet, as Navon cautions, while individually these conditions are extremely rare, cumulatively they are not and are hitherto forcing researchers to re-evaluate existing categories of disease and disability (Navon, 2019).

Many private companies have also begun to offer additional results based on sex chromosome abnormalities, while a handful of smaller companies are offering “whole genome sequencing (WGS)-based NIPT tests, promising to analyse every chromosome of a baby” (Thomas et al., 2020, p.89). In the absence of clear-cut regulatory governance, consumer companies give the illusion of full control, downplay the gap between screening and diagnosis, and can exploit pre-existing cultural anxieties of disability. This confronts scientists, prospective parents and inevitably biosocial stakeholders, with having to make sense of potentially overwhelming amounts of bio-information and how this could classify different ‘kinds’ of persons (Hacking, 2007).

Understandably, this question of governance has sparked debates about who will be responsible for overseeing the construction of appropriate regulations and what might be considered responsible innovation (Shakespeare & Hull, 2018). Currently, NIPT is only offered to all women in Belgium and the Netherlands for detecting trisomy 13, 18 and 21 (Gadsbøll et al., 2020). Nevertheless, the abilities of NIPT to screen more efficiently, have potential to enhance other biotechnologies including IVF, and the economic incentives to reduce the need for expensive invasive procedures, mean that barriers are likely to diminish, notions of disability increasingly blur, and we are left to see NIPT become further incorporated into the 'moral economies of prenatal testing' (Zeng et al., 2016)

As researchers, we should be attentive to the ways that NIPT shifts boundaries and shape identities. Even the ability to discover familiar conditions like down syndrome in utero (Löwy, 2014), complicates how technology and reproductive decision-making dialectically determine distinct embodiments. By bringing these embodiments forward, prospective parents and medical professionals will be confronted with having to make difficult decisions. While genetic counselling is designed to help facilitate people's reproductive decision-making, the presence of the medical practitioner is always a temporary one (Atkinson et al., 2013, p.1223), and the cultivation of disability identities accelerates outside the clinic. Indeed, they emerge through what Ginsburg and Rapp call 'kinship imaginaries', which refers to complex acts of cultural imaginations that occur in families trying to encompass the 'fact of disability' in the family (Ginsburg & Rapp, 2013).

Risk theory

NIPT clearly presents new and familiar theoretical challenges for developing scholarship on prenatal screening and disability identities. There are opportunities to analyse NIPT and changing disability identities by drawing influence from risk theory. While risk has a particular affinity with Foucauldian theory, this is by no means the only framework available.

For example, Landsman uses feminist theory to show how risk and prenatal screening dialectically intertwine with notions of stigma and responsible motherhood, appearing most egregious for those families with children whose conditions were undetected (Landsman, 2009). Furthermore, because screening results have an interpretive label (i.e., either negative or positive) rather than a numerical score, this leads to changes in risk perceptions and what prospective parents can expect from screening (Krimsky & Gruber, 2013, p.206).

Alternatively, Kelly found that prenatal screening technologies themselves became inherently risky for some parents with experience of raising a child with disabilities (Kelly, 2009). As Felicity Boardman has illustrated in various works, families of children with genetic disabilities contain 'experiential knowledge' about screening for different genetic conditions (Boardman, 2014, 2017), which may redirect internalised embodiments of risk towards different identities forming. Taking their contributions seriously, we learn that notions of risk and how that affects people's identities are particularly complex for those with embodied familial experiences of genetic conditions. However, this complexity does not feature in current studies of NIPT and parents of children with disabilities.

For example, a study with some Dutch parents of children with down syndrome found them expressing difficulties reconciling their gratefulness for obtaining an earlier diagnosis with a sense that NIPT also represents predetermined pathways to termination (How et al., 2019). Similarly, some Dutch parents of children with down syndrome were in favour of expanding the conditions screened for to 'divert the spotlight away from down syndrome' (van Schendel et al., 2017, p.529). In Germany, Reinsch et al learned that for some women who previously declined prenatal screening in earlier pregnancies, they struggle to justify their decisions not to test now that NIPT is presented as completely risk free (Reinsch et al., 2020). While together insightful for risk theory, these studies are few and they tend to treat disability identities in the NIPT literature as fixed and unchanging.

Before concluding, the final section attempts to synthesise these insights into the author's preliminary ethnographic fieldwork. It should be noted therefore, that these are not foregone conclusions, but are instead guiding principles that are being used to investigate changing notions of disability.

Iceland

While NIPT is strictly reserved for 'high risk' pregnancies, all women have the option to have the combined test between 11 and 14 weeks in addition to routine ultrasound (Halle & Fjose, 2016). Despite incurring a small fee and not regarded as a standard part of antenatal care (Halle et al., 2018), this has not dissuaded individuals from requesting the combined test (ibid). Rather, prenatal screening appears regarded by prospective parents and healthcare professionals as a 'normal' and 'expected' part of pregnancy (Gottfreðsdóttir & Björnsdóttir, 2010).

NIPT is however, likely to become implemented as part of the targeted screening approaches in all of Scandinavia in the near future (Juvet et al., 2016), and since 2019, a private IVF clinic began offering NIPT for 79,500 ISK (Livio Reykjavík, n.d). A survey conducted among those who received false-positive and true-negative first trimester screening results demonstrated that 77% of an 101 sample wanted NIPT in their next pregnancy if it were an option, despite only 21% knowing what NIPT was (Thorolfssdóttir et al., 2020). Similarly, for many healthcare professionals and pregnant women, NIPT and the broader objective of prenatal screening appears valued, even for those who decline testing (Ingvarsdóttir et al., 2016). However, there is no existing research which has taken to question the truisms of these conclusions, nor studies attempting to understanding how NIPT or prenatal screening has implications for changing disability identities. This is somewhat understandable, given how invisible NIPT is at this stage. However, given the pace at which these developments are unfolding, NIPT may become a focal point in national debates.

Iceland has a turbulent history of genomics, mainly associated with deCODE genetics which triggered many biopolitical debates in recent years about biodata and ownership (Palsson, 2008). Despite only being in existence for less than 30 years, deCODE has been key to shaping the public image and perception of Icelandic genes, linked with constructing ideas of citizenship and the 'Nordic body' (Winickoff, 2006). However, this raises further questions about how these developments are driving changing conceptions of disability, and how do

they take shape for those deemed genetically ‘at risk’? Understanding disability in Iceland goes back to the development of the Nordic welfare states and the principles of normalisation in the 1970s (Björnsdóttir & Jóhannesson, 2009, p.436). Given that transformations in genomics and biotechnology have taken place in such a short space of time, research can be conducted with different generations and social actors to capture how notions of genetic risk impact societal concepts of disability and how this effects the category of disability itself.

Conclusion

This paper has demonstrated how NIPT shapes the changing conceptions of disability through the augmentation of what it means to be ‘at risk’. The most obvious takeaway is a timely need for further research. It is likewise a limitation of this paper that the authors observations have only just begun. However, the hope is that the conceptual discussion developed here helps to encourage readers and future researchers to respond in kind.

References

- Atkinson, P., Featherstone, K., & Gregory, M. (2013). Kinscapes, timescapes and genescapes: Families living with genetic risk. *Sociology of Health & Illness*, 35(8), 1227–1241. <https://doi.org/10.1111/1467-9566.12034>
- Björnsdóttir, K., & Jóhannesson, I. (2009). People With Intellectual Disabilities in Iceland: A Bourdieuean Interpretation of Self-Advocacy. *Intellectual and Developmental Disabilities*, 47(6), 436–446. <https://doi.org/10.1352/1934-9556-47.6.436>
- Boardman, F. K. (2014). Knowledge is power? : The role of experiential knowledge in genetically ‘risky’ reproductive decisions. *Sociology of Health & Illness*, 36(1), 137–150. <https://doi.org/10.1111/1467-9566.12048>
- Boardman, F. K. (2017). Experience as knowledge: Disability, distillation and (reprogenetic) decision-making. *Social Science & Medicine*, 191, 186–193. <https://doi.org/10.1016/j.socscimed.2017.09.013>
- Brady, P., Brison, N., Van Den Bogaert, K., de Ravel, T., Peeters, H., Van Esch, H., Devriendt, K., Legius, E., & Vermeesch, J. R. (2016). Clinical implementation of NIPT - technical and biological challenges: Clinical implementation of NIPT. *Clinical Genetics*, 89(5), 523–530. <https://doi.org/10.1111/cge.12598>
- Butler, M. G. (2017). Benefits and limitations of prenatal screening for Prader-Willi syndrome: Prenatal screening for Prader-Willi syndrome. *Prenatal Diagnosis*, 37(1), 81–94. <https://doi.org/10.1002/pd.4914>
- de Wert, G., Bombard, Y., Bianchi, D. W., Bergmann, C., Borry, P., Chitty, L. S., Fellmann, F., Forzano, F., Hall, A., Henneman, L., Howard, H. C., Lucassen, A., Ormond, K., Peterlin, B., Radojkovic, D., Rogowski, W., Soller, M., Tibben, A., Tranebjærg, L., ... Cornel, M. C. (2015). Non-invasive prenatal testing for aneuploidy and beyond: Challenges of responsible innovation in prenatal screening. *European Journal of Human Genetics*, 23(11), 1438–1450. <https://doi.org/10.1038/ejhg.2015.57>
-

-
- Franklin, S. (2003). *Re-thinking nature—Culture: Anthropology and the new genetics*.
<https://journals.sagepub.com/doi/10.1177/1463499603003001752>
- Gadsbøll, K., Petersen, O. B., Gatinois, V., Strange, H., Jacobsson, B., Wapner, R., Vermeesch, J. R., The NIPT-map Study Group, & Vogel, I. (2020). Current use of noninvasive prenatal testing in Europe, Australia and the USA: A graphical presentation. *Acta Obstetrica et Gynecologica Scandinavica*, 99(6), 722–730. <https://doi.org/10.1111/aogs.13841>
- Genomics Education Programme. (2017, December 18). *What Is NIPT?*
- Ginsburg, F., & Rapp, R. (2013). *Disability Worlds*. 53–68.
<https://doi.org/10.1146/annurev-anthro-092412-155502>
- Gottfreðsdóttir, H., & Björnsdóttir, K. (2010). 'Have you had the test?' A discourse analysis of media presentation of prenatal screening in Iceland: Fetal screening and discourse analysis. *Scandinavian Journal of Caring Sciences*, 24(2), 414–421.
<https://doi.org/10.1111/j.1471-6712.2009.00727.x>
- Hacking, I. (2007). *Proceedings of the British Academy, Volume 151, 2006 Lectures*. British Academy.
- Halle, K. F., & Fjose, M. (2016). *Early prenatal screening in Iceland and Norway*.
- Halle, K. F., Fjose, M., Kristjansdóttir, H., Björnsdóttir, A., Getz, L., Tomasdóttir, M. O., & Sigurdsson, J. A. (2018). Use of pregnancy ultrasound before the 19th week scan: An analytical study based on the Icelandic Childbirth and Health Cohort. *BMC Pregnancy and Childbirth*, 18(1), 512. <https://doi.org/10.1186/s12884-018-2134-1>
- Harjee, R., Nakhuda, G. S., & Jing, C. (2020). NIPT RESULTS IN PREGNANCIES RESULTING FROM EUPLOID EMBRYO TRANSFERS. *Fertility and Sterility*, 114(3), 156–157.
<https://doi.org/10.1016/j.fertnstert.2020.08.450>
- How, B., Smidt, A., Wilson, N. J., Barton, R., & Valentin, C. (2019). 'We would have missed out so much had we terminated': What fathers of a child with Down syndrome think about current non-invasive prenatal testing for Down syndrome. *Journal of Intellectual Disabilities*, 23(3), 290–309. <https://doi.org/10.1177/1744629518787606>
- Ingvarsdóttir, S., Stefansdóttir, V., & Gottfredsdóttir, H. (2016). Women's preferences for prenatal tests A discrete choice experiment to contrast noninvasive prenatal testing with current invasive tests. *Laeknabladid*, 277-282. <https://doi.org/10.17992/lbl.2016.06.85>
- Jeppesen, L. D., Hatt, L., Singh, R., Ravn, K., Kølvrå, M., Schelde, P., Uldbjerg, N., Vogel, I., & Lildballe, D. L. (2021). Cell-based non-invasive prenatal diagnosis in a pregnancy at risk of cystic fibrosis. *Prenatal Diagnosis*, 41(2), 234–240. <https://doi.org/10.1002/pd.5861>
- Juvet, L. K., Ormstad, S. S., Schneider, A. S., Solberg, B., Arentz-Hansen, H., & Kvamme, M. K. (2016). Ikke invasiv prenatal testing (NIPT) for påvisning av trisomi 21, 18 og 13. Oslo: Folkehelseinstituttet.
-

-
- Kelly, S. E. (2009). Choosing not to choose: Reproductive responses of parents of children with genetic conditions or impairments. *Sociology of Health & Illness*, 81–97.
<https://onlinelibrary.wiley.com/doi/full/10.1111/j.1467-9566.2008.01110.x>
- Krimsky, S., & Gruber, J. (Eds.). (2013). *Genetic explanations: Sense and nonsense*. Harvard University Press.
- Landsman, G. H. (2009). *Reconstructing motherhood and disability in the age of 'perfect' babies*. Routledge.
- Livio Reykjavík. (n.d). *Verðskrá*. <https://livio.is/livio-reykjavik/verdskra/>
- Lo, Y. M. D., Corbetta, N., Chamberlain, P. F., Rai, V., Sargent, I. L., Redman, C. W. G., & Wainscoat, J. S. (1997). Presence of fetal DNA in maternal plasma and serum. *THE LANCET*, 350, 3.
- Lock, M. (2015). Comprehending the Body in the Era of the Epigenome. *Current Anthropology*, 56(2), 151–177. <https://doi.org/10.1086/680350>
- Lock, M. (2017). Recovering the Body. *Annual Review of Anthropology*, 46(1), 1–14.
<https://doi.org/10.1146/annurev-anthro-102116-041253>
- Löwy, I. (2014). Prenatal diagnosis: The irresistible rise of the 'visible fetus'. *Studies in History and Philosophy of Biological and Biomedical Sciences*, 10.
- Navon, D. (2019). *Mobilizing Mutations: Human Genetics in the age of Patient Advocacy*. University of Chicago Press.
- Palsson, G. (2008). The rise and fall of a biobank. The case of Iceland. In *Biobanks: Governance in Comparative Perspective*. (pp. 41–55). Routledge.
- Palsson, G. (2015). Anthropology, Genomics, and Human Variation: National Roots. In *International Encyclopedia of the Social & Behavioral Sciences* (pp. 758–764). Elsevier.
<https://doi.org/10.1016/B978-0-08-097086-8.82040-1>
- Reinsch, S., König, A., & Rehmann-Sutter, C. (2020). Decision-making about non-invasive prenatal testing: Women's moral reasoning in the absence of a risk of miscarriage in Germany. *New Genetics and Society*, 1–17.
<https://www.tandfonline.com/doi/full/10.1080/14636778.2020.1805305>
- Shakespeare, T., & Hull, R. (2018). Termination of Pregnancy After Non-Invasive Prenatal Testing (NIPT): Ethical Considerations. (Preprint not peer reviewed). *Journal of Practical Ethics*, 6(2), 23.
- Tamminga, S., van Schendel, R. V., Rommers, W., Bilardo, C. M., Pajkrt, E., Dondorp, W. J., van Maarle, M., Cornel, M. C., & Henneman, L. (2015). Changing to NIPT as a first-tier screening test and future perspectives: Opinions of health professionals: Changing to NIPT as a first-tier screening test and future perspectives. *Prenatal Diagnosis*, 35(13), 1316–1323.
<https://doi.org/10.1002/pd.4697>
-

-
- Thomas, G., Katz Rothman, B., Strange, H., & Latimer, J. E. (2020). *Testing Times: The Social Life of Non-invasive Prenatal Testing*.
<https://journals.sagepub.com/doi/10.1177/0971721820960262>
- Thorolfsson, E., Lunde, Å., Stefansson, V., Hjartardottir, H., & Haraldsson, K. R. (2020). *Comparing prenatal screening experiences of Icelandic women who received false-positive and true-negative first-trimester combined screening results in Iceland in 2012–2016*. <https://onlinelibrary.wiley.com/doi/abs/10.1002/jgc4.1269>
- van Schendel, R. V., Kater-Kuipers, A., van Vliet-Lachotzki, E. H., Dondorp, W. J., Cornel, M. C., & Henneman, L. (2017). What Do Parents of Children with Down Syndrome Think about Non-Invasive Prenatal Testing (NIPT)? *Journal of Genetic Counseling*, 26(3), 522–531.
<https://doi.org/10.1007/s10897-016-0012-4>
- Vanstone, M., Yacoub, K., Winsor, S., Giacomini, M., & Nisker, J. (2015). What Is “NIPT”? Divergent Characterizations of Noninvasive Prenatal Testing Strategies. *AJOB Empirical Bioethics*, 6(1), 54–67. <https://doi.org/10.1080/23294515.2014.993102>
- Verweij, E. J. (Joanne), Oepkes, D., de Vries, M., van den Akker, M. E. (Elske), van den Akker, E. S., & de Boer, M. A. (2013). Non-invasive prenatal screening for trisomy 21: What women want and are willing to pay. *Patient Education and Counseling*, 93(3), 641–645.
<https://doi.org/10.1016/j.pec.2013.08.006>
- Winickoff, D. E. (2006). Genome and Nation: Iceland’s Health Sector Database and its Legacy. *Innovations: Technology, Governance, Globalization*, 1(2), 80–105.
<https://doi.org/10.1162/itgg.2006.1.2.80>
- Zeng, X., Zannoni, L., Löwy, I., & Camporesi, S. (2016). Localizing NIPT: Practices and meanings of non-invasive prenatal testing in China, Italy, Brazil and the UK. *Ethics, Medicine and Public Health*, 2(3), 392–401. <https://doi.org/10.1016/j.jemep.2016.06.004>
- Zwart, H. (2009). Genomics and identity: The bioinformatisation of human life. *Medical Health Care and Philosophy*, 12, 125–136. <https://doi.org/10.1007/s11019-009-9187-x>
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