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Patients and Professionals Perplexed: An Analysis of Non-Invasive Prenatal Testing Implementation in the United States and United Kingdom

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ABSTRACT

A comparative analysis of the United States and the United Kingdom reveals important cross-national differences in the implementation of non-invasive prenatal testing (NIPT). To establish these key differences I conducted interviews with healthcare professionals in the US, and I utilized qualitative studies conducted within the UK. I conclude that in the United Kingdom, health care professionals struggle to accurately describe and present testing to patients, and have a much higher subsequent Down Syndrome abortion rate. In addition, many health care professionals in the United Kingdom fail to provide informed consent when administering NIPT. Comparatively, health care professionals in the United States do a much better job gaining informed consent, but often encounter time constraints and cannot present all relevant information. In the United States health care professionals also struggle with a shortage of genetic counselors, insurance coverage limitations, and a lack of data for low-risk populations. To conclude, I provide policy recommendations meant to address each of these issues.

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INTRODUCTION

"The plague of mankind is the fear and rejection of diversity: monotheism, monarchy, monogamy and, in our age, monomedicine. The belief that there is only one right way to live, only one right way to regulate religious, political, sexual, medical affairs is the root cause of the greatest threat to man: members of his own species, bent on ensuring his salvation, security, and safety." – Thomas Szasz, M.D.

One would be hard pressed to argue that diversity does not have a positive impact in many areas of life. In fact, initiatives have now expanded in some spheres from diversity itself towards the provision of an inclusive environment which allows and accepts the open expression of diverse viewpoints.[1] However, I would like to explore one area of public policy where diversity has been a point of contention, with some even contending it has been quelled. Non-invasive prenatal testing (NIPT) has been recently introduced in many countries. This test is seen as a much better alternative to invasive prenatal testing, which has a risk of spontaneous abortion.[2] Before the introduction of NIPT, invasive testing, along with information from ultrasounds, was the sole way for pregnant women to gain information on potential disorders. NIPT uses the blood of the mother to examine fragments of cell-free DNA (cfDNA) from the placenta in her bloodstream. This test is most accurate when used in identifying potential chromosomal disorders, particularly trisomy 13, trisomy 18, trisomy 21, and sex disorders caused by extra or missing copies of the X and Y chromosomes.[3] Trisomy 21, or Down Syndrome, is the condition of possessing an additional chromosome 21. This additional chromosome causes developmental and physical anomalies which vary on a case-by-case basis. Down Syndrome is the most common chromosomal congenital condition, occurring

[1] Stokes, Caroline. "How to Commit and Turn 'Diversity' into 'Inclusion'." *Entrepreneur*, 4 Nov. 2019, www.entrepreneur.com/article/339988.
[2] Halle, Kristine Flo, and Maria Fjose. "Early Prenatal Screening in Iceland and Norway." *Norwegian University of Science and Technology*, 30 Nov. 2016, pdfs.semanticscholar.org/f96b/ccac0d01ed083eae1aa165f0a0be215473b0.pdf.
[3] "What Is Noninvasive Prenatal Testing (NIPT) and What Disorders Can It Screen for? - Genetics Home Reference - NIH." *U.S. National Library of Medicine*, National Institutes of Health, ghr.nlm.nih.gov/primer/testing/nipt.

approximately one in 700 pregnancies and is the chromosomal disorder most often identified by the NIPT test. [4]

When it comes to a probable diagnosis of Down Syndrome, the use of NIPT leads to different outcomes across countries in which it is widely used. An abnormal test result that indicates a likelihood of Down Syndrome has led to differences in abortion rates across countries. In the United Kingdom, the number of women who choose abortion after a prenatal Down Syndrome diagnosis has been estimated at around 92%,[5] and this number has been estimated close to 100% in Iceland.[6] The abortion rate in the US has been estimated to be much lower, falling in the realm of 67%.[7] Many factors involved in higher abortion rates have been linked to the methods surrounding the implementation of NIPT.[10] Since the NIPT test and subsequent abortion estimates are so recent, most research in the area to date is focused on factors surrounding test administration as well as on the countries with higher abortion rates.

Much of what we currently know involves the NIPT test itself. A brief overview of this research is presented below, followed in the Literature Review with a more in-depth summary. NIPT was created as a "screening test;" experts recommend additional invasive testing if an abnormal result is obtained. Additionally, health care professionals recommend that genetic counseling should accompany NIPT.[8] Multiple qualitative studies have been carried out with women who choose to receive the NIPT test. Overall, these women have highly positive opinions about the test and its potential uses, but often are uninformed regarding certain aspects of

[4] "What Is Down Syndrome?: National Down Syndrome Society." *NDSS*, www.ndss.org/about-down-syndrome/down-syndrome/.
[5] Bissell, M.g. "Trends in Down's Syndrome Live Births and Antenatal Diagnoses in England and Wales from 1989 to 2008: Analysis of Data from the National Down Syndrome Cytogenetic Register." *Yearbook of Pathology and Laboratory Medicine*, vol. 2011, 26 Oct. 2011, pp. 333–334, doi:10.1016/s1077-9108(10)79509-8.
[6] Quinones, Julian and Arijeta Lajka. "'What Kind of Society Do You Want to Live in?": Inside the Country Where Down Syndrome Is Disappearing." *CBS News*, CBS Interactive, 14 Aug. 2017, www.cbsnews.com/news/down-syndrome-iceland/.
[7] Natoli, Jaime L., et al. "Prenatal Diagnosis of Down Syndrome: a Systematic Review of Termination Rates (1995-2011)." *Prenatal Diagnosis*, vol. 32, no. 2, 2012, pp. 142–153, doi:10.1002/pd.2910.
[8] Allyse, Megan, et al. "Non-Invasive Prenatal Testing: a Review of International Implementation and Challenges." *International Journal of Women's Health*, 2015, doi:https://dx.doi.org/10.2147%2FIJWH.S67124.

testing.[9] Within the United Kingdom, research has been conducted with health care professionals using interviews and focus groups. Conclusions have focused on potential problems surrounding NIPT's implementation, with some going as far to say that unclear professional standards in the United Kingdom undermine informed choice with respect to subsequent abortion.[10] However, to date there have been no qualitative studies done with doctors in the United States regarding NIPT and Down Syndrome abortion rates.

I aim to address a gap in NIPT research: what implementation factors impact the stark differences in Down Syndrome abortion rates across countries? Specifically, I will use qualitative data from the United States and the United Kingdom as my examples. Out of all of the countries who use NIPT with subsequent high Down Syndrome abortion rates, the United Kingdom offers the best point of comparison to the United States. Currently, the United Kingdom only offers NIPT in the private health care sector, while most other nations utilize a public health care system very different from health care in the United States.[11] I will address the policy structure of both nations in relation to NIPT in the Literature Review. Future research may be able to draw upon my work in order to extend these findings to other nations. This paper will discuss the impact of health care professionals' role in the United States and the United Kingdom regarding the administration of the NIPT test, and the potential abortion of a fetus with a positive result for Down Syndrome. For the United Kingdom, I will be using a sample of three previously conducted qualitative studies. I will compare this research with my own interviews conducted in the United States. My study will be conducted in Illinois, as it offers a similar abortion rate

[9] Halle, Kristine Flo, and Maria Fjose. "Early Prenatal Screening in Iceland and Norway." *Norwegian University of Science and Technology*, 30 Nov. 2016, pdfs.semanticscholar.org/f96b/ccac0d01ed083eae1aa165f0a0be215473b0.pdf.
[10] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[11] "UK National Screening Committee." *NHS Choices*, NHS, www.datadictionary.nhs.uk/data_dictionary/nhs_business_definitions/u/uk_national_screening_committee_de.asp?shownav=1.

baseline to the United States as a whole.[12] I will discuss the data supporting this claim in my methods section. Further research will need to be conducted to extend the applicability to individual states. My study will target a few key aspects of NIPT testing: informational and counseling resources, health care professional opinions regarding the test, and the circumstances in which the test is typically given.

As stated previously, health care professionals recommend NIPT be accompanied with genetic counseling. I will target the following questions with my research: What information is given to pregnant women about early pregnancy screenings and invasive tests? What do most women know before learning about the test? Are pre-test and post-test counseling offered with a specialist healthcare professional? How are results conveyed, and what is the guidance given to women after delivering these results? I hypothesize that women in countries with higher Down Syndrome abortion rates typically receive less information and genetic counseling services, or that the information they do receive is biased. If true, this assumption would indicate that women in the United States receive information and counseling that is more complete than that in the United Kingdom. This hypothesis would be consistent with previous research conducted in nations having higher NIPT Down Syndrome abortion rates.

I will also examine health care professionals' opinions regarding NIPT testing. Specifically, I aim to address the following questions: What are the positive uses of the test? What are the negative aspects? The term "sorting society" is often used in the debate: What do health care professionals think about this term? What do health care professionals think about the future of NIPT? Doctors in the United Kingdom have expressed some concerns regarding the

[12] "State Facts About Abortion: Illinois." *Guttmacher Institute*, Sept. 2019, www.guttmacher.org/fact-sheet/state-facts-about-abortion-illinois.

implementation of the test.[13] I believe that health care professionals in the United States may have fewer apprehensions related to NIPT implementation, as Down Syndrome abortion rates have been closer to expected rates, indicating that they are likely not effecting patient decision-making.

Finally, I will address when and to whom NIPT is offered. In the United Kingdom as well as Iceland, NIPT is offered early in one's pregnancy, leading health care professionals to believe women are more inclined to get an abortion when carrying their child only for a short amount of time.[14] I want to examine if administration of the test is offered at a different point during pregnancy in the United States, and if any discrepancies might effect abortion rates. I also would like to consider to whom the test is offered; if doctors in the United States offer the test to a smaller subgroup of women, this could affect differences in subsequent abortion rates.

The importance of this research lies in the public policy surrounding NIPT testing. Is the implementation of the NIPT test biased, influencing some women to think differently about carrying a pregnancy to full-term than others? I do not aim to provide an answer to the ethical debate of abortion; however, I do seek to identify measures that can be put in place surrounding NIPT testing in order for women make an informed choice about continuing or terminating their pregnancy. If we believe public policy to be designed to promote the best interests of a person, then public policy surrounding NIPT testing should provide women with complete information in order to make an informed decision regarding her pregnancy. My data and recommendations will specifically discuss the United Kingdom and the United States, and future research is needed

[13] Allyse, Megan, et al. "Non-Invasive Prenatal Testing: a Review of International Implementation and Challenges." *International Journal of Women's Health*, 2015, doi:https://dx.doi.org/10.2147%2FIJWH.S67124.
[14] Halle, Kristine Flo, and Maria Fjose. "Early Prenatal Screening in Iceland and Norway." *Norwegian University of Science and Technology*, 30 Nov. 2016, pdfs.semanticscholar.org/f96b/ccac0d01ed083eae1aa165f0a0be215473b0.pdf.

to determine if my findings can be generalized to other nations or applied in a more nuanced way to individual states.

Initially, I will present a Literature Review of previous research on the NIPT test and Down Syndrome, specifically contrasting the United States with the United Kingdom. This will be followed by an overview of my data collection methods and procedures in conjunction with a review of the comparable interview data from the United Kingdom. Next, in my Analysis section, I will contextualize my interview data within the framework of the research previously discussed in my Literature Review. Finally, I will conclude by recommending policy changes that will facilitate accurate and unbiased communication between health care providers and patients.

LITERATURE REVIEW

In this section, I will review three lines of research most relevant to my data: informed consent for NIPT testing, reproductive rights critiques, and disability rights critiques. Since the aim of my research is to ultimately provide policy recommendations, I will conclude with a brief overview of the current policy regarding NIPT in the United States and the United Kingdom. Of note is the fact that NIPT testing itself and related research is all quite new, and additional studies may significantly impact the findings of current research.

Informed Consent for NIPT Testing

Informed consent as defined by the American Medical Association (AMA) is "communication between a patient and physician" which accurately presents "the burdens, risks, and expected benefits of all options" including interventions. Health care professionals also should "assess the patient's ability to understand relevant medical information and the

implications of treatment alternatives and to make an independent, voluntary decision." The AMA defines informed consent in medical treatment as "fundamental in both ethics and law."[15] Given informed consent is a primary area of concern with NIPT testing, what then suffices as informed consent for NIPT?

Invasive prenatal testing requires written consent, as well as extensive doctor-patient communication due to the risk of miscarriage from the procedure.[16] However, informed consent for NIPT is not clearly defined. The procedure involves minimal risk while providing the same information as invasive testing and thus can be just as emotionally impactful on decisions affecting pregnancy.[17] A blood sample is a routine procedure, and one that is not directly associated with prenatal testing. This simplicity creates an additional concern regarding informed consent. Cernat et al. demonstrate that the ease of the procedure "could lead to routinization, pressure to test, and an erosion of informed choice." Pregnant women have echoed this concern, indicating that a routinization of testing could lead to a stigma for those who choose to decline NIPT.[18] In a recent study conducted by Montgomery and Thayer, women in the United States feel social pressure to receive NIPT testing.[19] "Responsible mothers" are those who go to childbirth classes, read all the right parenting literature, and prepare as best they can for the birth of their child.[20] It follows that a routinization of NIPT testing could very well lead to NIPT testing being considered an action only taken by "responsible mothers."

[15] "Informed Consent." *American Medical Association*, www.ama-assn.org/delivering-care/ethics/informed-consent.
[16] Ravitsky, Vardit. "The Shifting Landscape of Prenatal Testing:Between Reproductive Autonomy and Public Health." *Hastings Center Report*, vol. 47, 2017, doi:10.1002/hast.793.
[17] Cernat, Alexandra, et al. "Facilitating Informed Choice about Non-Invasive Prenatal Testing (NIPT): a Systematic Review and Qualitative Meta-Synthesis of Women's Experiences." *BMC Pregnancy and Childbirth*, vol. 19, no. 1, 2019, doi:10.1186/s12884-018-2168-4.
[18] Lewis, C., et al. "Non-Invasive Prenatal Testing for Down's Syndrome: Pregnant Women's Views and Likely Uptake." *Public Health Genomics*, vol. 16, no. 5, 2013, pp. 223–232, doi:10.1159/000353523.
[19] Montgomery, Sophie H., and Zaneta M. Thayer. "The Influence of Experiential Knowledge and Societal Perceptions on Decision-Making Regarding Non-Invasive Prenatal Testing (NIPT)." *Research Square*, 2019, doi:10.21203/rs.2.17104/v1.
[20] Rothenberg, Karen H., and Elizabeth Jean Thomson. *Women and Prenatal Testing: Facing the Challenges of Genetic Technology*. Ohio State University Press, 1994.

However, even as NIPT might not yet be perceived as "routine" in all parts of the United States and the United Kingdom, there are signs that NIPT testing may already suffer a lack of informed consent. In Berit Sjögren's 1988 study, 75% of women who were offered prenatal testing reported that they found it difficult to refuse such testing.[21] Even though NIPT is not widespread, it is perceived by those who know of it as straightforward and risk-free, leading expectant mothers to feel obligated to take the test.[22] In the United Kingdom, there have been reports of an even greater lack of informed consent than in the US with some women believing they were giving only a routine blood sample, and the doctor later presenting NIPT results.[23] Informed consent for NIPT needs to be balanced; health care providers must help support a woman making an informed decision without pushing her to make any specific choice.

Informed consent for NIPT is a complex public policy issue, but some ideas have been generated regarding how to better present the test in order to ensure women understand the test and freely want to take it. Minear et al. discussed changing the abbreviation for "non-invasive prenatal testing" from NIPT to NIPGS for "non-invasive prenatal genetic screening" to emphasize that this is a screening test, which requires additional testing to confirm a positive result. In a recent survey, over half of United States genetic counselors believed that a written consent form, as opposed to simple verbal discussion, should be required for NIPT testing.[24] It is clear not all women feel as if they are given the chance to provide informed consent that the AMA requires.[25] With my research, I aim to address the insufficiency of much current informed

[21] Sjögren, Berit, and Nils Uddenberg. "Decision Making during the Prenatal Diagnostic Procedure. A Questionnaire and Interview Study of 211 Women Participating in Prenatal Diagnosis." *Prenatal Diagnosis*, vol. 8, no. 4, 1988, pp. 263–273, doi:10.1002/pd.1970080404.
[22] Cernat, Alexandra, et al. "Facilitating Informed Choice about Non-Invasive Prenatal Testing (NIPT): a Systematic Review and Qualitative Meta-Synthesis of Women's Experiences." *BMC Pregnancy and Childbirth*, vol. 19, no. 1, 2019, doi:10.1186/s12884-018-2168-4.
[23] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[24] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.
[25] "Informed Consent." *American Medical Association*, www.ama-assn.org/delivering-care/ethics/informed-consent.

consent as well as potential policy solutions to ensure women are able to give true informed consent to NIPT testing.

Reproductive Rights Critiques

Proponents of reproductive rights and most women are generally very pleased with the introduction of NIPT testing. Non-invasive testing, in contrast to invasive prenatal tests, is much less of a risk to the health of a mother and her child, as it is less stressful both mentally and physically. [26, 27] While the test is not diagnostic, within the high-risk population, it is close to 99% accurate for diagnosing Down Syndrome and can be given as early as 9 weeks into pregnancy.[28] This allows women more time to make a knowledgeable decision regarding their pregnancy compared to later invasive testing, normally given around 14 weeks. For those who decide to terminate their pregnancies, they are able to obtain an earlier abortion in areas where termination is limited to under 20 weeks. Additionally, those who decide to keep their fetuses are able to gain more time to prepare for the birth of a child with special needs.[29] It is important to keep in mind that these results have only been validated on high risk pregnancies and cannot be generalized across all subgroups.

Embedded within the question of informed consent is the ethical question: Is it permissible to abort a child with Down Syndrome? Those strongly in favor of reproductive rights answer with a resounding "yes." Karen Rothenberg's and Elizabeth Thomson's *Women and Prenatal Testing: Facing the Challenges of Genetic Technology* outlines the reasoning behind the ethics of aborting a child with a disability. Mothers are inherently inclined to protect their

[26] Dondorp, Wybo, et al. "Non-Invasive Prenatal Testing for Aneuploidy and beyond: Challenges of Responsible Innovation in Prenatal Screening." *European Journal of Human Genetics*, vol. 23, no. 11, 2015, pp. 1438–1450, doi:10.1038/ejhg.2015.57.
[27] Lewis, C., et al. "Non-Invasive Prenatal Testing for Down's Syndrome: Pregnant Women's Views and Likely Uptake." *Public Health Genomics*, vol. 16, no. 5, 2013, pp. 223–232, doi:10.1159/000353523.
[28] Gil, M., et al. "Analysis of Cell-Free DNA in Maternal Blood in Screening for Aneuploidies: Meta-Analysis." *Fetal Diagnosis and Therapy*, vol. 35, no. 3, 2014, pp. 156–173, doi:10.1159/000358326.
[29] Dondorp, Wybo, et al. "Non-Invasive Prenatal Testing for Aneuploidy and beyond: Challenges of Responsible Innovation in Prenatal Screening." *European Journal of Human Genetics*, vol. 23, no. 11, 2015, pp. 1438–1450, doi:10.1038/ejhg.2015.57.

children and to help them attain a happy life. If a mother receives a prenatal diagnosis of Down Syndrome, who better to decide whether that child can have a quality life than the mother herself? Parents are those that can best determine how much time and resources they will be able to give a child with a disability; therefore, they are best suited to determine whether the child will have a quality life and should be brought into the world.[30] This viewpoint and freedom of choice is one that is threatened by policy surrounding the implementation of NIPT testing.

Proponents of reproductive rights believe that reproductive choice is jeopardized if too little or too many policy restrictions are put in place surrounding NIPT. The US Food and Drug Administration (FDA), which will be discussed further in the final section of the Literature Review, is the primary administrative body over NIPT in the United States. If the FDA does not act and implement policy regarding NIPT, some fear that this test could be leveraged to argue for the restriction of reproductive rights.[31] Female health could also be negatively impacted, as increased abortion restrictions will likely drive some women to pursue illegal and more dangerous means of termination.[32] Finally, these critics assert than manipulating and limiting reproductive decisions is an infringement upon a fundamental right. [33] Following this logic, NIPT extends reproductive autonomy, which many believe is an inherent right and should be protected through the use of public policy. I will not provide any recommendations specifically regarding abortion but offer policy recommendations on NIPT implementation that balance the assertions put forth by reproductive rights advocates with the claims of disability rights proponents.

[30] Rothenberg, Karen H., and Elizabeth Jean Thomson. "Women and Prenatal Testing: Facing the Challenges of Genetic Technology." Ohio State University Press, 1994.
[31] King, Jaime S. "Politics and Fetal Diagnostics Collide." *Nature*, vol. 491, no. 7422, 2012, pp. 33–34, doi:10.1038/491033a.
[32] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.
[33] Rothenberg, Karen H., and Elizabeth Jean Thomson. "Women and Prenatal Testing: Facing the Challenges of Genetic Technology." Ohio State University Press, 1994.

Disability Rights Critiques

Advocates of disability rights disagree with how NIPT is typically presented, as well as disagree with many of the previous claims described by reproductive rights proponents. Many argue that NIPT testing itself contains an inherent bias. According to Dondorp, et al., "prenatal screening sends a discriminatory message about the worth of the lives of people living with the relevant conditions."[34] Furthermore, results of testing are conveyed in terms of "risk."[35] Risk has an inherently negative connotation, which makes a statement about the perceived value of one's fetus. Miller and Levine demonstrate that health care professionals "often assume that the quality of life of a disabled individual is inversely proportional to the degree of physical or mental impairment."[36] Disability proponents assert that aborting a child does not "protect it" from what many believe is a low-quality life.[37] Their concern arises from these inherent biases that unfairly predispose reproductive decisions.

There have been multiple studies investigating information presented to women regarding NIPT testing and Down Syndrome. Farrelly, et al. found a large disparity in information provided by genetic counselors to patients about Down Syndrome. 95% of genetic counselors primarily addressed the physical conditions of disability, while only 27% discussed social aspects of disability. Furthermore, termination (86%) was the option most frequently presented by genetic counselors, followed by the less mentioned options of continuing one's pregnancy (37%) or putting the child up for adoption (13%).[38] Roberts, et al. demonstrated that many

[34] Dondorp, Wybo, et al. "Non-Invasive Prenatal Testing for Aneuploidy and beyond: Challenges of Responsible Innovation in Prenatal Screening." *European Journal of Human Genetics*, vol. 23, no. 11, 2015, pp. 1438–1450, doi:10.1038/ejhg.2015.57.
[35] Rothenberg, Karen H., and Elizabeth Jean Thomson. "Women and Prenatal Testing: Facing the Challenges of Genetic Technology." Ohio State University Press, 1994.
[36] Miller, Paul Steven, and Rebecca Leah Levine. "Avoiding Genetic Genocide: Understanding Good Intentions and Eugenics in the Complex Dialogue between the Medical and Disability Communities." *Genet Med*, 16 Aug. 2012, doi:10.1038/gim.2012.102.
[37] Rothenberg, Karen H., and Elizabeth Jean Thomson. "Women and Prenatal Testing: Facing the Challenges of Genetic Technology." Ohio State University Press, 1994.
[38] Farrelly, E, et al. "Genetic Counseling for Prenatal Testing: Where Is the Discussion about Disability?" *Journal of Genetic Counseling* , 21 Dec. 2012, doi:10.1007/s10897-012-9534-6.

women did not receive information on "the positive as well as the negative aspects of giving birth to a child with disabilities." Women were more inclined to terminate their pregnancies if they had less knowledge of disability-related services.[39] In fact, most medical students do not receive clinical training regarding psychological disabilities, and medical schools concede this training is a low priority, leading to health care professionals often feeling uncomfortable delivering a Down Syndrome diagnosis.[40, 41] As NIPT testing is most frequently used to identify Down Syndrome, the imbalance in the type of information provided about the testing could have large effects on the outcomes of Down Syndrome pregnancies.

Disability rights advocates are not fighting for the elimination of NIPT testing altogether, but rather advocating for an improvement in information surrounding the test. In fact, most current mothers of children with Down Syndrome report that they would use the test as a way to gain more time to prepare for the birth of a disabled child rather than to determine whether to terminate their pregnancy.[42] These disability rights supporters believe that health care professionals need to equally present the positive and negative aspects of a Down Syndrome diagnosis. Advocates do not consider Down Syndrome a "disability," but rather point to the problems experienced by disabled persons as "the result of intolerance, poorly conceived social programs, and environmental or communication barriers than can be moved by changes in social policy."[43] Many of the problems surrounding NIPT informed consent, they believe, result from the type of information presented and the tone of presentation. Skotko et al. conducted research

[39] Roberts, CD, et al. "The Role of Genetic Counseling in the Elective Termination of Pregnancies Involving Fetuses with Disabilities." *The Journal of Special Education*, 2002, doi:10.1177/00224669020360010501.
[40] Skotko, BG, et al. Down Syndrome Diagnosis Study Group. 2009. "Prenatal diagnosis of Down syndrome: How best to deliver the news." Am J Med Genet Part A 149A:2361–2367.
[41] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.
[42] Kellogg, Gregory, et al. "Attitudes of Mothers of Children with Down Syndrome Towards Noninvasive Prenatal Testing." *Journal of Genetic Counseling*, vol. 23, no. 5, 2014, pp. 805–813, doi:10.1007/s10897-014-9694-7.
[43] Rothenberg, Karen H., and Elizabeth Jean Thomson. "Women and Prenatal Testing: Facing the Challenges of Genetic Technology." Ohio State University Press, 1994.

in order to provide specific recommendations on how to present a balanced diagnosis of Down Syndrome. Women receiving the diagnosis "should be provided with up-to-date information about what Down Syndrome is, what causes Down Syndrome, and what are the expectations for a child with Down Syndrome living today." Furthermore, health care professionals should discuss all options surrounding pregnancy and emphasize that parents may receive negative comments no matter how they decide to act regarding the pregnancy. Skotko et al. asserts that mothers are more satisfied when the news is delivered with "neutral and nondirective language," not "risk," and when parents are given the opportunity to speak with parents who already have a child with Down Syndrome.[44] I aim to discuss these considerations and recommendations with health care professionals in my research in order to determine how to best mitigate the concerns of disability rights proponents surrounding the implementation of NIPT testing.

Policies Surrounding NIPT Testing

NIPT has only been implemented in the United States and the United Kingdom since late 2011.[45] Despite its advantages, NIPT brings with it the aforementioned concerns surrounding the issues of test accessibility, informed choice, reproductive and disability rights. Lawmakers have begun to address some of these concerns affecting NIPT testing, but the debate continues regarding the best way to structure policy. While my paper discusses the impact on Down Syndrome termination rates, I will solely provide recommendations on the implementation of NIPT testing rather than make any recommendations on abortion policies. Furthermore, abortion regulations differ widely across the United States and the United Kingdom, making the

[44] Skotko, BG, et al. Down Syndrome Diagnosis Study Group. 2009. "Prenatal diagnosis of Down syndrome: How best to deliver the news." Am J Med Genet Part A 149A:2361–2367.
[45] Chandrasekharan, S., et al. "Noninvasive Prenatal Testing Goes Global." *Science Translational Medicine*, vol. 6, no. 231, 2014, doi:10.1126/scitranslmed.3008704.

consolidation of these policies a much broader issue.[46, 47] I will first discuss the current status of NIPT testing in the United States, and follow with a summary of the debate in the United Kingdom.

In October of 2008, the US Congress passed the "Prenatally and Postnatally Diagnosed Conditions Awareness Act" which was enacted in order "to amend the Public Health Act to increase the provision of scientifically sound information and support services to patients receiving a positive test diagnosis for Down Syndrome or other prenatally and postnatally diagnosed conditions." This act specifically aims "to increase patient referrals to providers of key support services for women who have received a positive diagnosis for Down Syndrome," as well as strengthen existing public health support networks and ensure that "patients receive up-to-date, evidence-based information about the accuracy of the test." Specifically, the bill calls for patients to receive information on the range of outcomes that accompany Down Syndrome, and additionally for health care providers to supply information regarding specific support services. [48] This federal law remains unfunded; however, nine states have subsequently passed similar legislation. Illinois is one of 41 states which has not passed and funded a comparable statute.[49] Thus, the aims of the "Prenatally and Postnatally Diagnosed Conditions Awareness Act" are currently not funded throughout most of the United States.

Aside from the US Congress, the other national body that has potential regulatory oversight on NIPT testing is the Food and Drug Administration (FDA). NIPT tests are currently sold in the US by five companies, all who advertise the test as "laboratory developed." Because

[46] "What are the UK's Laws on Abortion?" *BBC News*, BBC, 22 Oct. 2019, www.bbc.com/news/health-19856314.
[47] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.
[48] United States, Congress, "An Act to Amend the Public Health Service Act to Increase the Provision of Scientifically Sound Information and Support Services to Patients Receiving a Positive Test Diagnosis for Down Syndrome or Other Prenatally and Postnatally Diagnosed Conditions." U.S. G.P.O., 2008. 110th Congress, bill Public Law 110–374.
[49] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.

these NIPT tests are "laboratory developed," the FDA is not technically required to regulate them. The FDA currently does not regulate NIPT or other similar prenatal chromosomal tests. However, the FDA has indicated that they are reconsidering their position, as the debate surrounding NIPT continues to intensify.[50] Without governmental oversight in the US, several heath care professional associations have provided their own guidelines on NIPT testing. The American College of Obstetricians (ACOG) provides multiple committee opinions regarding testing guidelines as well as resources to facilitate physicians' discussions of a positive diagnosis of Down Syndrome.[51] The National Society of Genetic Counselors (NSGC) offers support for the test. NSGC stresses that health care providers "should communicate the benefits and limitations" of the test, as well as understand "it may not be the most appreciate option for every pregnancy." Furthermore, the NSGC emphasizes that patients should have the opportunity to have expert post-test genetic counseling.[52] Altogether, other than guidelines put in place by national organizations and unfunded national laws, there are very few formal regulations on NIPT test implementation in the United States,.

The United Kingdom has not passed any national legislation directly affecting the implementation of NIPT. Recently, the UK National Screening Committee (UK NSC), an advisory body over all types of population screenings, recommended that the United Kingdom develop a plan to roll out NIPT testing nationwide in the public health systems.[53] The implementation of NIPT testing itself has not yet taken effect in the public sector, but I will provide a short overview of the national standards promoted by the UK NSC. In the future public

[50] Allyse, Megan, et al. "Non-Invasive Prenatal Testing: a Review of International Implementation and Challenges." *International Journal of Women's Health*, 2015, p. 113, doi:10.2147/ijwh.s67124.
[51] "Women's Health Care Physicians." *ACOG*, www.acog.org/About-ACOG/ACOG-Departments/Genetics/Prenatal-Genetic-Screening-Diagnostic-Testing.
[52] "Prenatal Cell-Free DNA Screening." *National Society of Genetic Counselors*, www.nsgc.org/p/bl/et/blogaid=805.
[53] "UK National Screening Committee." *NHS Choices*, NHS, www.datadictionary.nhs.uk/data_dictionary/nhs_business_definitions/u/uk_national_screening_committee_de.asp?shownav=1.

health initiative, the Committee requires that all women have equal access to NIPT screening early in their pregnancy if they so choose to take the test. However, the UK NSC provides very few other stipulations on the implementation of the test, as the rest of their guidelines concern general information about NIPT itself rather than its administration.[54] As the United Kingdom plans to roll out NIPT in public health care in the near future, health care professionals have criticized the lack of governmental support and training resources.[55] Additionally, the only association of UK health care professionals that have discussed NIPT testing to date is the British Medical Association (BMA), which just recently underwent a study to determine the limits of NIPT test implementation. The BMA currently is consulting with the public as well as relevant doctors in order to potentially instill limitations on the use and implementation of NIPT testing.[56] The lack of any real consensus and few notable investigations in both the United States and United Kingdom justifies the need for my research as well as further study. With medical societies' stances in both nations being so rare and varied, individual opinions of health care professionals in both nations tend to guide the test's use, leading to somewhat haphazard implementation. Comparing the differing professional opinions of Down Syndrome abortion rates in the United States and the United Kingdom assists us in making public policy decisions knowing potential effects these decisions might have on outcomes.

Summary

Overall, neither the United States nor the United Kingdom has clear guidelines for the implementation of NIPT testing, which fuels this controversy. There are many stakeholders in

[54] United Kingdom, Public Health England, "NHS Fetal Anomaly Screening Programme Handbook." *NHS Fetal Anomaly Screening Programme Handbook*, 2018.
[55] "Non-Invasive Prenatal Testing: Ethical Issues: Review of Activities since Publication." *Nuffield Council on Bioethics*, Nov. 2018, nuffieldbioethics.org/wp-content/uploads/Nuffield-Council-NIPT-review-of-activites.pdf.
[56] Iacobucci, Gareth. "Non-Invasive Prenatal Testing: Public and Doctors Should Be Consulted, Says BMA." *The BMJ*, British Medical Journal Publishing Group, 3 July 2018, www.bmj.com/content/362/bmj.k2916.

NIPT testing and the potential implementation of policy surrounding the test. Informed consent, and the best way to attain this, is a large topic of debate surrounding NIPT. Additionally, proponents of reproductive and disability rights often struggle in determining the ethical limits surrounding testing. The current policies impacting NIPT are very narrow in scope, which leaves space for vast differences in test administration, leading to most stakeholders' dissatisfaction. I aim to look at the issue through a "veil of ignorance" from *A Theory of Justice* written by John Rawls. The "veil of ignorance" is a stance from which I examine the policy space without regard to any specifics about myself, my position in society, or my abilities.[57] I could very well be a proponent of reproductive rights, a disability rights advocate, or a health care provider. No matter from which position I were to find myself, I hope to provide actionable public policy recommendations to ensure that each of these stakeholders have their liberties protected.

METHODOLOGY

Within this section, I will discuss the methodology used to collect my data, as well as the methodology employed to collect the comparison data from the United Kingdom. For United States data, I conducted my own series of interviews. Regarding the United Kingdom data, I utilize three different studies. The time constraints on my research as well as geographical limitations did not allow me to obtain first-hand data from the UK. Much of my methodology is focused on collecting United States interview material comparable to the previously gathered United Kingdom data. All of my assumptions are based on previous research regarding NIPT in the US as well as the United Kingdom. My research is entirely qualitative, and I intend to establish a correlative rather than direct causal relationship within my findings. From my

[57] Rawls, John. *A Theory of Justice*. Universal Law Publishing Co Ltd, 2013.

findings, I hope to provide policy and research recommendations that may help guide future analysis around NIPT and Down Syndrome.

As stated previously, I aim to use my research to address three aspects surrounding NIPT testing and Down Syndrome: informational and counseling resources, health care professional opinions regarding the test, and the circumstances in which the test is typically given. Genetic counseling is highly recommended to accompany NIPT.[58] All three United Kingdom data sources that I use express concern regarding the quality of counseling and/or informed consent surrounding NIPT testing.[59,60,61] From this previous research, I assume that the genetic counseling and information given regarding the NIPT test has an impact on the decision to potentially terminate a Down Syndrome pregnancy.

I also make assumptions regarding the impact of health care professionals' opinions and the circumstances surrounding the test. I am assuming that qualitative data is the best way to get health care professionals' opinions as well as obtain information on their interactions with patients. Available quantitative data does not provide any insight into NIPT's implementation. I assume that health care professionals will be forthcoming about the population offered NIPT as well as the point in pregnancy it is discussed. Qualitative means of obtaining data is used in the three studies from the United Kingdom as well as many other studies around the world.[62] I will speak more to the methodology of these studies later in this section. Comparative quantitative statistics are already available from the United States and United Kingdom. I believe that the use

[58] Allyse, Megan, et al. "Non-Invasive Prenatal Testing: a Review of International Implementation and Challenges." *International Journal of Women's Health*, 2015, doi:https://dx.doi.org/10.2147%2FIJWH.S67124.
[59] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[60] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[61] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[62] Potter, Beth K., et al. "Exploring Informed Choice in the Context of Prenatal Testing: Findings from a Qualitative Study." *Health Expectations*, vol. 11, no. 4, 2008, pp. 355–365, doi:10.1111/j.1369-7625.2008.00493.

of interviews will allow me to obtain data beyond that of already existing quantitative statistics, allowing me to obtain additional information to assist in explaining the disparity in statistics.

In my own data collection, I have made assumptions regarding the potential generalizability of the results from the doctors whom I interviewed to the United States as a whole. Due to time and geographic constraints, I am not able to interview health care professionals across the United States. Furthermore, as abortion laws and regulations vary widely across the United States, I chose to collect data from only one state, Illinois. In order to ensure complete generalizability to other parts of the United States, one will need to look at particular state laws surrounding abortion and health care. Also of note is that these professionals self-selected to participate in the study, and I have a small sample size of 15 individuals. These factors could result in data primarily reflecting professionals having a greater interest in the NIPT test, which could be more pronounced due to smaller sample size. This interview method of data collection is similar to other qualitative studies on NIPT with participants self-selected and sample size small.[63] It is difficult to obtain large qualitative data sets, requiring additional research to confirm my conclusions. However, other than some states possessing different legal frameworks than Illinois, there is no apparent reason why this state's health care professionals' opinions and answers would greatly vary from those across the United States. The following figure will display data that shows the comparison of Illinois general abortion rates to those of the United States.

[63] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.

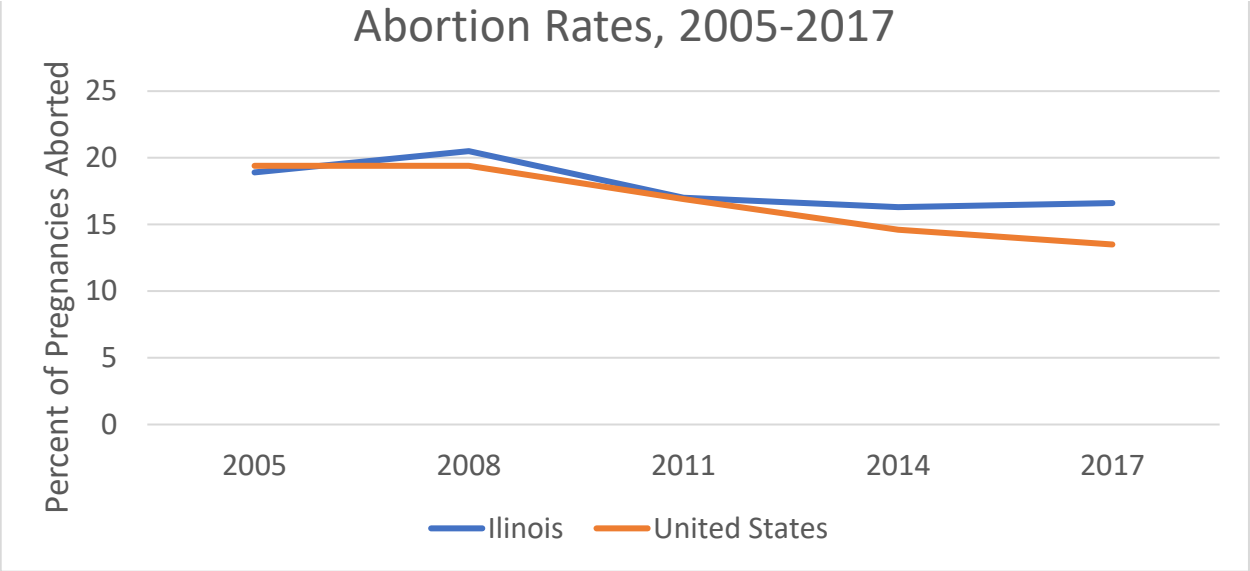

Figure 1: Abortion rates in the United States and Illinois[64]

Figure 1 illustrates that abortion rates in Illinois closely mirror those in the United States. The x-axis represents the year of the observed data, which was recorded every three years. The y-axis shows the percent of pregnancies that ended in an abortion. The blue line traces the abortion rates in Illinois, and the red line shows the rates in the United States. The largest difference in the abortion rates in this dataset occurred in the last recorded year, 2017, where the difference between Illinois and the United States was 3.1%.

I will now discuss the manner of my data collection and construction, first looking at the three data sources I am using to represent the United Kingdom. Again, employing data collected by other researchers is not ideal but practical; I would prefer to conduct interviews in the United Kingdom myself, but time and geography constraints do not allow for this. All of the data collected in the United Kingdom was published between 2013-2014. NIPT has been accessible in the private health care sector in the United Kingdom since 2012.[65] Currently, the United

[64] Data obtained from The Guttmacher Institute
[65] "NIPT in the Private Sector." *Nuffield Bioethics*, nuffieldbioethics.org/report/noninvasive-prenatal-testing-ethical-issues/nipt-private-sector.

Kingdom is rolling out small scale tests to potentially include NIPT testing into the National Prenatal Screening Program.[66,67] This initiative is the only slight change to NIPT testing in the UK subsequent to the studies I am using; however, this initiative will only be fully implemented well after my study is completed and therefore will not affect my results. Although the UK data is a few years old, there is no reason to believe health care professionals and administration of the NIPT test have significantly changed since the time of collection, as there have been no major changes in the United Kingdom's prenatal health care during this time period.

Initially, I will discuss "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives" conducted by Elizabeth Alexander, et al. and released in October 2014. Subsequently, I will refer to this study as "Study 1."[68] This study conducted interviews with 20 prenatal genetic counselors at four different centers in the United Kingdom. The interviewees were selected by contacting lead genetic counselors in those four facilities, who then each recommended six genetic counselors for participation in the study. The first 20 recommended genetic counselors all agreed to participate, and therefore the response rate is 100%. 18 of the 20 participants had direct experience administering the NIPT test. Study 1 looks at participants' experiences with the general use of NIPT as well as NIPT used in cases of fetal sex identification, and discusses the implications of using NIPT to diagnose Down Syndrome. I will use this study as data to illustrate genetic counselors' opinions of NIPT in Great Britain and specifically how NIPT is used to diagnosis Down Syndrome.

[66] Reporter, Staff. "UK Approves Use of NIPT as Part of National Screening Program." *GenomeWeb*, 4 Nov. 2016, www.genomeweb.com/molecular-diagnostics/uk-approves-use-nipt-part-national-screening-program.
[67] McHugh, Annette. "Update on NIPT Implementation - Microarray Testing Technology and Twins." *PHE Screening*, 17 June 2019, phescreening.blog.gov.uk/2019/06/17/update-on-nipt-implementation-microarray-testing-technology-and-twins/.
[68] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.

Just as with my data, Study 1 provides a small sample size of 20 individuals that is not proportionally representative of the entire United Kingdom. This small set is a limitation of using interview-based qualitative data, and so, in order to present a more robust data set on Great Britain, I will supplement the data used here with two additional qualitative studies. Within the data given by Study 1, I will use interview quotes the researchers gathered to support their overall conclusions regarding themes of consensus among the interviewees. As this data is qualitative, I will not make any modifications to the data set, but only identify and focus on themes that can be compared to my own research in the United States.

Secondly, I will discuss "Views and preferences for the implementation of non-invasive prenatal diagnosis for single gene disorders from health professionals in the United Kingdom" conducted by Melissa Hill, et al. and first published in May 2013.[69] Subsequently, I will refer to this study as "Study 2." This study focused on general usage of NIPT, its use in diagnosing single gene disorders, as well as diagnosing Down Syndrome. Researchers obtained qualitative data from a variety of doctors and genetic counselors utilizing the test. Focus groups were conducted with 17 participants, and one-on-one interviews were utilized for 30 participants. Health care professionals were contacted by email detailing the study and requesting that interested participants contact the research team. The study reported a response rate of 39%. These participants were self-selected, and there is a possibility that these professionals represent a segment of health care workers that have stronger than average views on NIPT. However, their responses are consistent with other studies conducted in the United Kingdom, so I believe impact of self-selected participation to be minimal. Data in Study 2 was recorded and subsequently

[69] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.

transcribed. I will use themes and direct quotes from this study which focus on the NIPT's general implementation and specific use diagnosing Down Syndrome. Again, I will only utilize the information specifically relevant to my study.

Finally, I will discuss the United Kingdom study "Will the introduction of non-invasive prenatal testing for Down's syndrome undermine informed choice?" conducted by Caroline Silcock et al. and published in February 2014.[70] Subsequently, I will refer to this study as "Study 3." This study takes the form of a questionnaire, using data from 393 health care professionals and 523 pregnant women. I will only be using the data from the health care professionals and excluding responses from the pregnant women, as my study in the United States focuses exclusively on information obtained from health care professionals. Data was obtained primarily from obstetricians given a questionnaire distributed across three conferences in the United Kingdom. The questionnaire took the form of a vignette, in which health care professionals were asked to imagine themselves discussing prenatal care with a pregnant woman. Multiple choice options as well as spaces were provided to explain one's answers. No response rate was reported for Study 3. This data will be used to supplement Study 1 and Study 2 findings from the United Kingdom, ultimately serving as an additional point of comparison with the United States results.

For my own data set collected in the United States, I spoke with a variety of health care professionals with some form of involvement in implementation of the NIPT test, totaling 15 interviews. As an undergraduate with few connections to the medical community, I initially contacted local hospitals and genetic counseling services. These hospitals and genetic counseling services gave me the names of health care professionals who I subsequently reached through email. I scheduled one-on-one interviews with the health care professionals in their personal

[70] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.

offices. When necessary, due to time and geographic restrictions, I conducted interviews over Skype. These interviews took place from November 2019 through January 2020. All interview data was coded and kept anonymous; therefore, no names will be transcribed in my analysis.

I studied previous research in order to find areas of focus for my interview questions. Much of my intent with these focus areas was to obtain data comparable to the data I am using from the United Kingdom which resulted in my interviews following a semi-structured format. As stated previously, these focus areas are the following: informational and counseling resources, health care professional opinions regarding the test, and the circumstances in which the test is typically given. I entered each interview with a list of questions pertaining to the above topics, and asked follow-up questions if I felt the interviewee could provide additional insight or clarification. A list of these questions may be found in the appendix at the end of this paper. The semi-structured interview format allowed interviewees to reveal additional points of interest that I will discuss further in my analysis section. My referrals had a 100% response rate, though some referred me to more experienced providers of NIPT, resulting in a pool of very qualified interviewees and thus highly credible data. However, this method may result in a lack of data concerning health care providers who have limited experience administering NIPT.

In order to conduct my study, I needed comparable data from both the United States and the United Kingdom. Upon finding three studies of comparison from the United Kingdom, I identified the three focus areas mentioned above. Within my data from the United Kingdom, I was able to find a great deal of supporting information relevant to these focus areas. I then developed the semi-structured interview guide mentioned above to obtain data in these focus areas from health care professionals in the United States. After conducting interviews, I examined all the qualitative data that was obtained from the participants, not just the data that

supported my initial hypothesis, comparing it to the United Kingdom studies. When discrepancies were found, I placed these variances into the larger context of studies from my Literature Review in order to draw potential conclusions. I used these results in order to provide recommendations for future research and policies surrounding the NIPT test and Down Syndrome.

ANALYSIS OF FINDINGS

While many scientific studies have been done thus far regarding NIPT testing, very little policy research has been conducted, and almost none has been performed using the personal opinions of health care professionals. I have interviewed and collected data from 15 health care professionals from the United States which I will use to compare to the previously obtained findings from studies conducted in the United Kingdom. The US participants consisted of six Doctors of Medicine (MD) who administer NIPT and nine genetic counselors. The MDs were composed of a family planning OBGYN specialist, four general OBGYNs, and a pediatrician, all of whom frequently administer the NIPT test. Three of the genetic counselors were hospital employees, five employees of genetic counseling centers, and the other an employee of a NIPT distributor.

My discussion of research focuses on three areas: NIPT implementation, health care professionals' opinions and knowledge of NIPT, and populations who have access to NITP. Large disparities, both between and within the US and UK, exist in how testing is initially presented, the degree of informed consent, and how a positive diagnosis is framed. Health care professionals hold mostly positive beliefs regarding NIPT testing, but also have strong concerns surrounding informational risk, societal pressure, and limited professional resources and training,

especially as NIPT access is expanded. Finally, medical professionals in both nations differ in who they offer NIPT, while insurance restrictions were regarded as a large barrier to access in the United States. US health care professionals also discussed the lack of testing statistics released by testing companies regarding low-risk populations. All of my analysis ultimately pertains to the implementation of NIPT and my policy recommendations. As a result, some of the discussion between sections may converge. I will follow this analysis section with policy recommendations.

NIPT Implementation

Most early adapters of NIPT in the US originally used the screening test to solely identify Down Syndrome. In the eight years since the test has been implemented, many women now come in looking for the test as the "sex text" or the "Down Syndrome test." [71] While pregnant women in the United States did not have much knowledge about NIPT at first, fortunately genetic consolers were readily prepared to both inform and administer this test. Prior to the implementation of the test in late 2011, the majority of genetic counselors in the United States attended a week-long annual genetic counseling conference focused on developing a nuanced understanding of NIPT and its implementation. As a result, many counselors in the US felt very prepared to administer and discuss NIPT with patients in the United States.[72] However, in the United Kingdom, genetic counselors have varying levels of technical understanding regarding NIPT. Furthermore, many of these genetic counselors are unsure how to present "inconclusive" results, and expressed uneasiness due to their lack of experience with the test. One genetic counselor expressed this concern stating,

> "Certainly initially I think a lot of genetic counselors, myself included, felt a bit anxious about this test. It wasn't 100% certain and certainly in the first year or two people

[71] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[72] Burt, Miranda. "Personal Interview." 22 Nov. 2019.

> deciding to carry on [with the pregnancy] on the basis of the test was quite a nerve-wracking time."[73]

The apprehension expressed by the genetic counselors in the United Kingdom likely suppressed the initial recommendation of the test. In the United States, the genetic counselors interviewed did not express any apprehensions about presenting test results or dealing with false negatives, thus likely resulting in a higher rate of recommendation.

Both the United States and the United Kingdom rarely had patients see genetic counselors for specific pre-test counseling. In the United Kingdom, patients might receive pre-test counseling if they were considered to be part of a "high-risk" group.[74] Women in the United States received pre-test counseling if they were high risk or had a lot of questions surrounding testing. This lack of routine pre-test counseling in the United States and United Kingdom is primarily due to lack of resources, which I will discuss in more depth later in my analysis. One US genetic counselor discussed the benefits of providing pre-test counseling to all patients.

> "If there were enough resources and time, theoretically, it would be helpful… But I don't think it'd be feasible to counsel all those patients, especially low risk patients."[75]

Many patients "didn't understand the test beforehand, didn't know they would be receiving this information and now they have it."[76] All genetic counselors interviewed echoed this sentiment, saying that while all patients could benefit from pre-test counseling, this solution is not practical. For those high-risk patients who do receive pre-test counseling in the United States, counselors discuss family history and emphasize that "all testing is optional; it's appropriate to accept or decline."[77] Some genetic counseling agencies use video platforms and visual aids to ensure that

[73] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[74] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[75] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[76] Burt, Miranda. "Personal Interview." 13 Jan. 2020.
[77] Burt, Miranda. "Personal Interview." 22 Nov. 2019.

all patients receive the same pre-test information in an easily understood manner.[78] Genetic counselors believe many of these high-risk women do not know what they will do at the time of the test, but want to "be reassured that things are ok" by receiving an affirmative test result.[79]

US physicians, in contrast to US genetic counselors, emphasized the question "how would you use that information?" if women received a positive result. These physicians generally would encourage women to abstain from taking the test if they did not think they would act on the information.[80,81] Physicians in the United States used a variety of communication mediums from verbal to visual aids. The most common area of concurrence among all US doctors was an emphasis on NIPT being "just a screening test" multiple times throughout their interviews. On the other hand, the line between NIPT as a screening test versus a diagnostic test was blurred in the United Kingdom. As one English health care professional stated,

> "I think once you've moved away from fetal sexing alone and talking about diagnosing [disorders], then my concern about any potential error rate would be even more concerning to me… You know, they say it's never perfect."[82]

Many other health care professionals in the United Kingdom felt similarly, as they described NIPT as having diagnostic abilities.[83] This disparity between the UK and US in viewing NIPT as a diagnostic test was seen throughout my research, particularly with health care professionals in the United States going out of their way to emphasize that NIPT is solely a screening test and should not be used to diagnose any condition.

[78] Burt, Miranda. "Personal Interview." 25 Nov. 2019.
[79] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[80] Burt, Miranda. "Personal Interview." 15 Nov. 2019.
[81] Burt, Miranda. "Personal Interview." 21 Nov. 2019.
[82] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[83] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.

In addition to the disparity in how doctors present and view NIPT testing in the US and the UK, there is also a difference between heath care professionals in the two countries regarding informed consent for NIPT testing. I aim to address many of these problems surrounding informed consent in my Policy Recommendations. Within the US, health care professionals ranged from "no required informed consent to the test"[84] to "everything we do is documented with written consent."[85] One US genetic counselor discussed the benefits of having written informed consent saying,

> "So we have a whole discussion, upon which we touch upon the points of informed consent. But not all providers have the time or the inclination to have that type of discussion. You know, if you think about it, they may only have 15 minutes to see a pregnant woman. And there's a lot of things they need to discuss. Besides, genetic screening is just one thing on a long list of things they need to discuss… I mean, I think a lot of times, informed consent is just used as a paper that you give patients that is designed to protect yourself if something goes wrong… And you may end up limiting access to women who would really benefit from testing if you put a lot of requirements on it."[86]

This sentiment indicates that while in principle written informed consent may be helpful to require for NIPT testing, in practice the implementation of this requirement might be harder to attain. The majority of US health care professionals I interviewed echoed this opinion, agreeing that requiring written informed consent likely will not solve the problem of patients fully receiving and understanding information before consenting to testing. Most US physicians I interviewed believed they personally do a great job obtaining informed consent, and many stated that they discuss every blood test they conduct with their patients. One OBGYN specialist described her discussions regarding blood tests stating,

> "On the initial prenatal visit, you are drawing a lot of blood for a patient. I actually go through and tell them every single blood test I'm ordering, and I show them on the screen what I am ordering. They are able to ask questions and refuse any test."[87]

[84] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[85] Burt, Miranda. "Personal Interview." 21 Nov. 2019.
[86] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[87] Burt, Miranda. "Personal Interview." 12 Dec. 2019.

In the United Kingdom, NIPT is seen as routine and, therefore, a recommended test by doctors. Unlike in the US, some in the UK believe that "written consent for NIPT may help the test stand out from routine blood tests and encourage fuller discussion of the implications."[88] Physicians in the UK do not believe women take a blood test as seriously, and the lack of written consent could lead to ramifications when patients receive results. As one physician stated, "people could get on a path- they don't realize the consequences of the path until they're a long way down it."[89] A majority of genetic counselors in the United Kingdom expressed concerns regarding informed consent for NIPT. One genetic counselor in the United Kingdom described her concerns around informed consent for NIPT,

> "Even if you're explaining it's for this, this, this and this, this is what happens, and I suppose that is informed consent but I think, from a nursing and midwifery background, the woman just puts her arm out and the blood's taken, that is informed consent."[90]

It is clear that there is a distinction between the two countries on what constitutes informed consent, and, furthermore, what actual impact informed consent can realistically have on the implementation of the test.

What happens in a discussion if the results of an NIPT screening test are positive? There has not been much research done on the topic in the United Kingdom, but I studied the language used when delivering a positive diagnosis in the United States. Most often, health care professionals call patients to deliver initial news, then they follow up with a more detailed conversation in-person. US physicians and genetic counselors all stated that they typically use

[88] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[89] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[90] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.

phrases such as “higher risk than average for your baby to have Down Syndrome,” and then they would follow-up with more patient specific test details. Health care professionals in the United States emphasize that the test could be wrong, or might not detect a certain condition.

> “I emphasize that just because this test result isn't normal, it does not mean your baby is normal. And then I tell them that like you can have birth defects and have a chromosomally normal baby. You can have babies that have neither visible birth defects nor abnormal chromosomes but still have an issue like autism, for instance, for which there is no test.”[91]

Of the genetic counselors interviewed, differences became apparent depending on whether one was based at a hospital or based at an agency comprised of solely genetic counselors. Those in hospitals modified their care based on perceived patient understanding, and those within genetic counseling centers tried to give “routine care” to everyone.[92] Clearly, positive test results are presented differently across disciplines in the United States. However, all physicians and genetic counselors did emphasize that a positive test result is merely indicative of a higher risk, and the test is only a screening tool as opposed to a diagnostic test.

After presenting a positive diagnosis, US health care professionals tailor the information they supply the patient to the direction the patient wants to take the pregnancy. The majority of health care professionals that I interviewed (12 of 15) repeatedly emphasized that they would strongly encourage patients to seek invasive testing to confirm the screening results and would not recommend termination on NIPT results alone. One physician discussed how she presents information following a positive result saying,

> “This is a very good screening test, more than 90% accuracy in that patient population if she is someone of advanced maternal age. I would say the screening test is likely to be correct… I still would never say that I recommend termination. I would never confirm a diagnosis, just from the cell-free DNA test.”[93]

[91] Burt, Miranda. “Personal Interview.” 18 Dec. 2020.
[92] Burt, Miranda. “Personal Interview.” 25 Nov. 2019.
[93] Burt, Miranda. “Personal Interview.” 22 Nov. 2019.

The vast majority of US health care professionals reiterated that they would never confirm a diagnosis from NIPT testing. Genetic counselors and doctors first try to determine the patient's experience with Down Syndrome when describing the disorder. "Most people say they know what Down Syndrome is but then if you ask them, it's from what they've seen on TV."[94] One genetic counselor described what she told patients stating,

> "When we talk about intellectual disability, people generally don't know what that means… People [with Down Syndrome] would be expected to learn to walk and talk, dress themselves, but do those at later ages. They would attend school, but may need some special education assistance with school. They would play with friends, but may do that at a later age."[95]

Furthermore, health care providers emphasized that "there is a whole community of people who live with Down Syndrome" and "there are resources and counselors that can help with that population."[96,97] The information that doctors and genetic counselors provide patients primarily depends on the direction they wish to proceed. If patients are leaning towards continuing their pregnancy, most health care professionals will give resources that can connect parents with Down Syndrome parents and support groups. On the other hand, if patients want to terminate their pregnancy, physicians will first recommend invasive testing to confirm the diagnosis. If the patient still insists on terminating their pregnancy, then health care professionals will remind them that NIPT is not diagnostic before connecting them with the appropriate resources.

In conclusion, there are many differences between how NIPT is implemented in the United Kingdom and the United States. Health care professionals in the United Kingdom have received little training on how to implement NIPT, and patients there have expressed more

[94] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[95] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[96] Burt, Miranda. "Personal Interview." 6 Jan. 2020.
[97] Burt, Miranda. "Personal Interview." 17 Dec. 2019.

dissatisfaction with their care surrounding testing.[98] On the other hand, genetic counselors in the US have been given extensive training on NIPT, and health care professionals in the United States endeavor to prompt patients to think how they would use the information from NIPT before they receive it. These differences extend to informed consent, where physicians in the United Kingdom believe requiring written informed consent would help improve patient satisfaction with NIPT. US physicians vary in the degree of informed consent provided, but all physicians I interviewed at the very least made sure to touch upon the points of informed consent in conversations with patients. Finally, while there is little literature surrounding the presentation of a positive diagnosis of NIPT to a patient in the United Kingdom, health care professionals in the US present a positive result as "higher risk" and present only those resources useful in supporting a patient's wishes. Physicians and genetic counselors in the United States strongly recommend invasive testing to confirm a positive result before a woman decides to terminate her pregnancy.

Health Care Professional Opinions Surrounding NIPT Testing

With the introduction of NIPT testing, health care professionals in both the United Kingdom and the United States see many benefits for prenatal care. When asked about the positive facets of NIPT, US health care professionals note that the test has low false positive rates, can be conducted earlier in one's pregnancy, and reduces the number of invasive procedures.[99] One US physician sees few risks specific to NIPT's identification of Down Syndrome, as she believes American society as a whole has taken huge steps towards accepting children with Down Syndrome.[100] In the United Kingdom, health care professionals state that the

[98] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[99] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[100] Burt, Miranda. "Personal Interview." 15 Nov. 2019.

test is often cheaper than invasive testing, but note that it could lead to negative perceptions of Down Syndrome.[101] Professionals in the UK also emphasized that NIPT is much easier both emotionally and physically on patients than previously used invasive procedures.[102]

Health care professionals, both in the US and UK, expressed concern with regard to NIPT's use in decision making. Genetic counselors in the US mentioned that women often come to them after getting an abnormal NIPT result, believing that their baby is definitively diagnosed with a chromosomal disorder. Some patients have expressed that they did not know why the test was conducted until the results were directly in front of them.[103] One US physician said that patients have a "different concept of risk" with NIPT as opposed to invasive testing, and spoke extensively regarding her concerns about the "informational risk" that NIPT poses.[104] Another US physician said due to the quick rollout of NIPT, large portions of the population believe that it is diagnostic. She believes that about half of her own patients do not understand the test.[105] While the specific concerns vary in the US, it is clear that many physicians believe that not all women are receiving adequate information before they undergo NIPT testing. In the United Kingdom, there is also concern regarding inadequate informed consent. One UK genetic counselor spoke of patients saying,

> "If it was 'just a blood test,' in inverted commas, then anyone could basically do the test and probably a wider number of people would be able to order it, and I wonder if maybe less discussion would go into it because of the lack of miscarriage risk. So I think people spend quite a long time thinking about consent for physical procedures, and I wonder if the same level of thought would go into if there was no sort of perceived physical threat to the pregnancy."[106]

[101] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[102] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[103] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[104] Burt, Miranda. "Personal Interview." 15 Nov. 2019.
[105] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[106] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.

Another United Kingdom genetic counselor echoed this concern, this time speaking specifically about informed consent,

> "Some couples will accept whatever test is offered to them, but that doesn't mean to say they are fully informed. I think we have a duty of care to couples and especially to women who are pregnant of giving them the best information that we can."[107]

Concerns in the United Kingdom primarily revolve around the lack of fully informed consent. In both counties, most health care professionals agree that ensuring patients understand NIPT testing is a point of concern.

Increased societal pressure surrounding NIPT testing is also problematic. US genetic counselors are primarily concerned the test might be "done as part of routine care, rather than it actually being a voluntary process."[108] Over half of genetic counselors interviewed in the United Kingdom believed that NIPT could have a stigmatizing effect on disability, as societal expectations can pressure women to test.[109] One UK genetic counselor spoke in more detail saying,

> "I feel that there will be perhaps more pressure on women to have this testing because it will be seen as this great technology that is available and that women might feel that, almost a responsibility for them to use [it]… and having a child with the condition that they could easily have tested for would be wrong when it's so widely available. So, I think that it could have implications from that point of view."[110]

Other physicians in the UK echoed this concern, stating that increased pressure to use NIPT testing could lead to increased pressure to terminate a fetus who receives an abnormal result.[111] Many health care professionals interviewed in the UK have even gone as far as to say that when

[107] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[108] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[109] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[110] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[111] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.

NIPT testing becomes routine, specific written consent should be required and testing should be conducted at a later visit.[112] Although informed consent is a point of concern in both countries, in the United Kingdom almost all health care professionals interviewed thought that NIPT could harm the Down Syndrome population.

Specifically, health care professionals express a number of concerns including misunderstandings of NIPT results. Genetic counselors are concerned with the test's accuracy "because of the potential serious implications including termination of pregnancy or the unexpected birth of an affected child."[113] While there is no direct evidence that United Kingdom physicians and genetic counselors view the test as diagnostic, 50% of genetic counselors in one study think that NIPT can be utilized to replace invasive testing for high risk mothers.[114] This statistic is alarming; if the majority of health care professionals in the UK think NIPT, a screening test, is as good as a diagnostic test in some populations, they may well misrepresent the test as diagnostic to patients. This could lead some women to continue or terminate a pregnancy with information that could potentially be incorrect without the confirmation of a true diagnostic test. Other physicians in the United Kingdom have expressed that even though NIPT provides less information than invasive testing, it will increase pressure on women to abort a fetus with a positive diagnosis.[115] Increased pressure to terminate is a direct misunderstanding of NIPT, as a positive result is only meant to lead to further invasive testing.

[112] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[113] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[114] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[115] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.

As NIPT testing increases in popularity and accessibility, health care providers in the US and UK believe additional issues will arise. In the United Kingdom, genetic counselors are concerned that when more women want the test, the capacity of genetic counselors and doctors will be overwhelmed, and midwives will be called upon to administer it. Furthermore, there are concerns that it would be difficult to provide the necessary training and education to those midwives who would be called to implement NIPT.[116] UK professionals also expressed concern that the test could expand such that it is utilized to screen not only for more disorders, but also for personal qualities such as IQ or eye color.[117] Providers emphasized the need for ongoing and effective training as well as guidelines for NIPT in the United Kingdom.[118,119] In the US, genetic counselors believe that the physicians they work with need additional education as they start to administer more NIPT tests. Genetic counselors feel that many US physicians still do not thoroughly and correctly present NIPT to their patients.[120]

Heath care providers in the US and the UK strongly agreed that patient access to genetic counselors is very important. In the US, there is evidence that the number of genetic counselors is growing, but these counselors are located primarily in urban areas with large populations, limiting access in rural areas.[121]

[116] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.
[117] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[118] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[119] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[120] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[121] Burt, Miranda. "Personal Interview." 21 Nov. 2019.

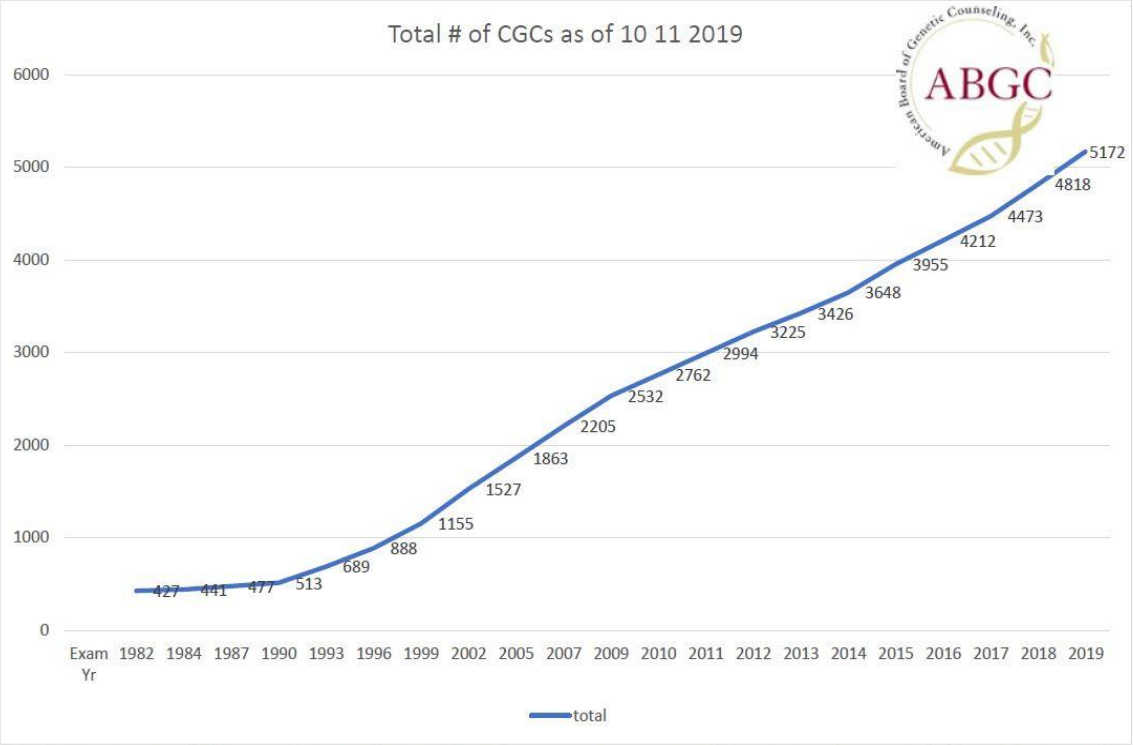

Figure 2: Total number of Certified Genetic Counselors as of October 11, 2019 as reported by the American Board of Genetic Counseling[122]

Figure 2 illustrates the growth of the genetic counseling profession in the US since 1982. However, as recently as 2014, there were no genetic counselors in two states and just three genetic counselors in three US states.[123] I will address the lack of genetic counselors in my subsequent Policy Recommendations. One US physician specializing in fetal medicine spoke of the need for more genetic counselors saying,

> "If anything, I think we've needed their help more, because we have more tests that have different sensitivities and specific specificities. And we're following a lot more of these abnormal screening tests with diagnostic tests that include more than just like a carrier type, because we're doing carrier attire. We're doing snips. We're doing microarray. And some people are starting to consider doing more whole genome sequencing… If anything, I think we've leaned on [genetic counselors] more."[124]

This physician went on to discuss the benefits of genetic counseling before administering NIPT, as well as her preference for having genetic counselors in the same hospital where she was

[122] "Total Number of Certified Genetic Counselors." American Board of Genetic Counseling. 11 Oct. 2019.
[123] Minear, Mollie A., et al. "Noninvasive Prenatal Genetic Testing: Current and Emerging Ethical, Legal, and Social Issues." *Annual Review of Genomics and Human Genetics*, vol. 16, no. 1, 2015, pp. 369–398, doi:10.1146/annurev-genom-090314-050000.
[124] Burt, Miranda. "Personal Interview." 22 Nov. 2019.

based. All physicians interviewed stated that having access to more genetic counselors would help ease testing concerns. Understanding all aspects of NIPT testing "if you do it correctly, takes over 45 minutes" which physicians do not have time to do.[125] One physician had to refer his patients to genetic counselors over Skype because of their facility's location. While he did admit that patients would probably prefer to meet in person if possible, he reported high levels of satisfaction with online genetic counseling.[126] Every physician and genetic counselor in the US interviewed echoed this sentiment. They agreed that access to online genetic counselors, as long as they are not employees of the testing lab, was much better than having no access to a genetic counselor at all. Health care professionals were apprehensive that genetic counselors employed by testing labs would have financial incentives to encourage women to receive testing. One genetic counselor interviewed conducted consultations over the phone spoke of the benefits of telecommunication.

> "Patients can talk in privacy of their own home, on the weekends… It's almost just as effective as in person. People are so appreciative to talk without waiting."[127]

Those in the United Kingdom did not discuss online genetic counseling or genetic counseling by phone, but many were adamant that good genetic counseling was just as important for NIPT as it is for invasive testing.[128] In both countries, the expansion of NIPT needs to be met with expanding patient access to genetic counselors, who are preferred by physicians and patients alike to deliver abnormal test results.

In summary, NIPT offers many benefits to pregnant women who might receive the test. However, there are also many drawbacks, particularly related to decision making. In both the US

[125] Burt, Miranda. "Personal Interview." 18 Dec. 2019.
[126] Burt, Miranda. "Personal Interview." 21 Nov. 2019.
[127] Burt, Miranda. "Personal Interview." 6 Jan. 2020.
[128] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.

and the UK, testing has brought about debates regarding informed consent, patients' understanding of the test, and increased societal pressure to receive testing as well as that pressure's effects upon a woman's decision-making process. Furthermore, there is evidence that health care professionals in the United Kingdom themselves do not adequately understand the test itself. With the many positive aspects of NIPT and its ease of use, it is inevitable that it will continue to be given to a wider population of women in the future. Many in both the US and the UK fear that an increase in testing will not be met with an increase in health care personnel and training. From opinions expressed by health care providers in both nations, it is clear that there needs to be public policy surrounding the ever-expanding effects and implementation of NIPT.

Populations Having Access to NIPT

In both countries, there are specific groups who currently have access to NIPT. At present, the United Kingdom is working on the future implementation of NIPT into its public health care system.[129] The private health care sector in the UK is primarily supported by private health care insurance, "typically funded as part of an employer funded healthcare scheme or is directly paid by the consumer."[130]In the UK, the test is given to most pregnant women who receive health care in the private sector.[131] Health care professionals in the United States differ in whom they offer NIPT. Some offer it to all patients, and others just offer it to "high risk" patients or those in their first pregnancy. All US health care professionals offered NIPT to their relevant patient population on their first prenatal visit so that they might have ample time to respond to an abnormal test result.

[129] "UK National Screening Committee." *NHS Choices*, NHS, www.datadictionary.nhs.uk/data_dictionary/nhs_business_definitions/u/uk_national_screening_committee_de.asp?shownav=1.
[130] Chang, Josh, et al. "The UK Health Care System." *Columbia University*.
[131] Alexander, Elizabeth, et al. "Non-Invasive Prenatal Testing: UK Genetic Counselors' Experiences and Perspectives." *Journal of Genetic Counseling*, vol. 24, no. 2, 2014, pp. 300–311, doi:10.1007/s10897-014-9765-9.

The most prohibitive barrier to NIPT access in the US, according to physicians, is health care insurance. When NIPT was first introduced, no insurance companies covered the test. Now, all insurance companies will cover the test for patients at high risk for a chromosomal disorder detectable by NIPT.[132] If a patient does not fall into a high-risk category, then she would have to pay for the test out of pocket. At present, test companies have dropped their prices to about $300 a test in order to boost purchases and incentivize insurance companies to cover NIPT for all patients.[133] However, the cost remains a barrier for many lower risk patients who cannot afford such a high out-of-pocket fee. "Cost plays a big role in patient decision making," said one genetic counselor.[134] US health care professionals all expressed their desire for expanded insurance coverage, citing out-of-pocket costs as a barrier. One physician stated, "I wish that I didn't have to worry about insurance coverage or billing, so that I could offer what I thought was best today and streamline care."[135]

Another barrier to NIPT access was revealed through some of my interviews with US health care professionals. Many stated that the test has made huge strides in accuracy, especially for high risk patients, since its inception.[136] However, testing providers seem to have been unsuccessful in making the same advances for low-risk populations. I will address this problem further in my Policy Recommendations. One OBGYN discussed the problems with low-risk populations of patients who now come in seeking NIPT testing.

> "I do mention that it is available to all patients. But I warn them that I don't have data for low-risk mothers. I still only have the sensitivity and specificity for women that I know that have a risk factor. I still don't have data for lower score women, many of the people that are routinely ordering these tests… So I can order this test on them, but I don't have a

[132] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[133] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[134] Burt, Miranda. "Personal Interview." 25 Nov. 2019.
[135] Burt, Miranda. "Personal Interview." 22 Nov. 2019.
[136] Burt, Miranda. "Personal Interview." 16 Dec. 2019.

> validated population to base it off of, which to me is kind of fishy. Again, the test has been out for a while, why don't we have that?"[137]

The majority of US health care professionals agreed, citing that low-risk women come in looking for "the test that can tell you the gender," and medical professionals have little data to discuss low-risk sensitivity regarding other risks.[138] Test providers have not provided any justification as to why they have not released any low-risk statistics. As NIPT is provided by similar companies in the United States and the United Kingdom, it is likely that this problem exists in both countries. This lack of data is a disservice to both health care providers and patients who wish to undergo NIPT testing. The lack of useful predictive data poses an ethical question as well: is it ethical to order NIPT with no normative data to interpret it? Without data specific to low-risk women who make up the vast majority of pregnant mothers, health care professionals have no basis to interpret what a positive result really means in the context of their patient's pregnancy. This renders the test practically useless in low-risk populations.

Both the United Kingdom and the United States have barriers to NIPT access. The United Kingdom currently only offers NIPT within the private health care system. Patients in the US and the UK who do not have insurance that covers NIPT must pay out of pocket, a cost that is high enough to limit access to the test for some. Furthermore, there is an overall lack of data for NIPT testing in low-risk populations. From my research, there are no strong indications that patient access, as compared to implementation, has led to the disparity in Down Syndrome abortion rates between the two countries. However, as public policy helps ease the problems associated with NIPT test implementation, it will be important to address and expand access to NIPT for all patient populations.

[137] Burt, Miranda. "Personal Interview." 12 Dec. 2019.
[138] Burt, Miranda. "Personal Interview." 6 Jan. 2020.

POLICY RECOMMENDATIONS

Multiple stakeholders are involved in NIPT testing: women, families, doctors, genetic counselors, and many others who hope to make the best possible decision regarding a pregnancy. Across the United States and the United Kingdom, NIPT testing involves inconsistent care and implementation methods. These differences in implementation most likely have resulted in the aforementioned varying Down Syndrome abortion rates that follow testing. In this section, I hope to provide policy recommendations that will be beneficial to all stakeholders involved and give expectant mothers complete and unbiased information on NIPT testing. As my both my direct data collection and place of residence are in the United States, I will confine my policy recommendations to the US. The United Kingdom provides an excellent point of comparison in my study, but more research will need to be done in order to support these policy implementations if they are to be implemented in the UK or any other nation.

My research supports three policy recommendations that address the disparity in information given to patients as well as limited access to genetic counseling resources. First, I will discuss the use of videos to help obtain informed consent and overcome the time constraints that many doctors face. I will then examine expanding access to genetic counselors through telecommunication. Finally, I will present a legislative proposal that would require more information to be released by NIPT testing providers. If implemented, these proposals will help ensure not only that women have the tools to make an informed decision, but also that doctors will be more prepared and better able to discuss and administer NIPT tests. I conclude by addressing the need and urgency for the implementation of these policy recommendations.

Pre-Consultation Videos

Many health care professionals struggle to communicate all the necessary information about NIPT in a prenatal visit. "That conversation, if you do it correctly, takes over 45 minutes," but most providers only have around 30 minutes to discuss NIPT testing among numerous other matters.[139] If a woman already has a thorough understanding of NIPT, or if a woman does not receive an abnormal test result, the lack of time spent discussing NIPT likely does not have any detrimental consequences. However, health care professionals have seen "a decent number of patients who don't entirely understand why they are getting testing"[140] and others who "didn't understand the test beforehand, didn't know they would be receiving abnormal information, and now they have it."[141] Clearly, the lack of time and disparity in information presented before undergoing NIPT testing creates problems.

Health care professionals and researchers in the United Kingdom have suggested that mandating written informed consent for NIPT testing would alleviate some of the disparity in information that patients receive.[142,143] However, higher patient understanding in the United States was not necessarily due to written informed consent, as many providers do not obtain written informed consent.[144,145,146] Almost all US providers (14 of 15) professed that they did not think that requiring written consent would help alleviate any of the problems surrounding NIPT testing. As one OBGYN discussed, consent issues arise more as a function of the conversation with one's health care provider rather than written consent.

[139] Burt, Miranda. "Personal Interview." 23 Nov. 2019.
[140] Burt, Miranda. "Personal Interview." 6 Jan. 2020.
[141] Burt, Miranda. "Personal Interview." 13 Jan. 2020.
[142] Silcock, Caroline, et al. "Will the Introduction of Non-Invasive Prenatal Testing for Down's Syndrome Undermine Informed Choice?" *Health Expectations*, vol. 18, no. 5, 2014, pp. 1658–1671, doi:10.1111/hex.12159.
[143] Hill, Melissa, et al. "Views and Preferences for the Implementation of Non-Invasive Prenatal Diagnosis for Single Gene Disorders from Health Professionals in the United Kingdom." *American Journal of Medical Genetics Part A*, vol. 161, no. 7, 2013, pp. 1612–1618, doi:10.1002/ajmg.a.35972.
[144] Burt, Miranda. "Personal Interview." 6 Jan. 2020.
[145] Burt, Miranda. "Personal Interview." 11 Nov. 2019.
[146] Burt, Miranda. "Personal Interview." 25 Nov. 2019.

> "What consent actually is, that is expressed to patients as much more of a function of the conversation you have… You can have people sign the paper and that gives you permission, but it doesn't mean the patient is understanding of the risk. That has a lot to do with the actual conversation. And that's something that is hard to encapsulate in a paper form."[147]

From my research, mandating written consent in the United States would do little, if anything, to improve patients' understanding of NIPT testing.

However, health care providers in the United States discussed the positive implications of using videos to help ensure patient understanding. In this context, short videos are given to a patient on a tablet prior to a consultation with a doctor. This allows patients to get a basic and consistent understanding of NIPT testing, and they are then able to ask their health care provider any questions during their visit. Three out of the fifteen health care providers I interviewed already incorporate videos as part of their informed consent process, and the others all reacted positively to the option of implemented pre-consultation videos. As one OBGYN stated,

> "I probably spend about five to ten minutes total discussing carrier screening and genetic screening… showing patients a video of the basics would allow me to assess the patient's knowledge without rushing through everything."[148]

This sentiment on pre-consultation videos was shared by all health care professionals who discussed the videos with me.

Many studies have been conducted previously about the merits of short informational films prior to meeting with a health care professional. These videos, used across a variety of different medical domains, have been shown to help patients achieve a higher level of

[147] Burt, Miranda. "Personal Interview." 23 Nov. 2019.
[148] Burt, Miranda. "Personal Interview." 12 Dec. 2019.

understanding as compared to just talking to a health care provider.[149,150,151,152,153] Furthermore, many patients prefer the video as opposed to simply speaking with their health care professional.[154,155,156] Video informed consent has proven to be successful in helping patients who are part of a minority group or from a low-literacy background.[157,158] While pre-consultation videos have not been adopted as formal policy in the US, very positive outcomes have resulted when implemented. The evidence presented in these studies, along with my research, strongly supports the implementation of pre-consultation videos as formal NIPT policy.

Pre-consultation videos should be overseen and required by the American College of Obstetricians and Gynecologists (ACOG) and the National Society of Genetic Counselors (NSGC). Both of these societies already have issued basic opinions on NIPT testing, so this policy would be an extension of their prior research. The ACOG and the NSGC need to provide elemental requirements and framework for the pre-consultation NIPT video. Hospitals and care facilities would then provide direct and more detailed oversight for video implementation. This system would ensure that both doctors' and genetic counselors' concerns are addressed in the primary video components. Additionally, oversight at the facility level allows for flexibility in implementation based on different factors such as patient population and facility resources.

149 Armstrong, A.w., et al. "Portable Video Media for Presenting Informed Consent and Wound Care Instructions for Skin Biopsies: a Randomized Controlled Trial." *British Journal of Dermatology*, vol. 163, no. 5, 2010, pp. 1014–1019, doi:10.1111/j.1365-2133.2010.10067.x.
150 Fureman, I., et al. "Evaluation of a Video-Supplement to Informed Consent: Injection Drug Users and Preventive HIV Vaccine Efficacy Trials." *Europe PMC*, 31 July 1997.
151 Rossi, Michael J., et al. "Video Informed Consent Improves Knee Arthroscopy Patient Comprehension." *Arthroscopy: The Journal of Arthroscopic & Related Surgery*, vol. 21, no. 6, 2005, pp. 739–743, doi:10.1016/j.arthro.2005.02.015.
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153 Migden, Michael, et al. "The Use of High Definition Video Modules for Delivery of Informed Consent and Wound Care Education in the Mohs Surgery Unit." *Seminars in Cutaneous Medicine and Surgery*, vol. 27, no. 1, 2008, pp. 89–93, doi:10.1016/j.sder.2008.02.001.
154 Armstrong, A.w., et al. "Portable Video Media for Presenting Informed Consent and Wound Care Instructions for Skin Biopsies: a Randomized Controlled Trial." *British Journal of Dermatology*, vol. 163, no. 5, 2010, pp. 1014–1019, doi:10.1111/j.1365-2133.2010.10067.x.
155 Fureman, I., et al. "Evaluation of a Video-Supplement to Informed Consent: Injection Drug Users and Preventive HIV Vaccine Efficacy Trials." *Europe PMC*, 31 July 1997.
156 Migden, Michael, et al. "The Use of High Definition Video Modules for Delivery of Informed Consent and Wound Care Education in the Mohs Surgery Unit." *Seminars in Cutaneous Medicine and Surgery*, vol. 27, no. 1, 2008, pp. 89–93, doi:10.1016/j.sder.2008.02.001.
157 Olanrewaju, Muhammed. "Multimedia Informed Consent Tool for a Low Literacy African Research Population: Development and Pilot-Testing." *Journal of Clinical Research & Bioethics*, vol. 05, no. 03, 2014, doi:10.4172/2155-9627.1000178.
158 George, Sheba, et al. "Using Animation as an Information Tool to Advance Health Research Literacy among Minority Participants." *AMIA Annual Symposium Proceedings Archive*, 16 Nov. 2013.

While additional analysis needs to be done to determine the required components of the video, a few strong elements emerged from my research and should be included. Health care professionals emphasized that NIPT is a screening test, stressing its difference from a diagnostic test. Furthermore, heath care professionals maintained that patients should be very aware of all of the conditions that NIPT screens for, and, if this information is not desirable, patients should understand that they are able to decline testing. When the health care professionals interviewed did encounter problems with NIPT, most revolved around NIPT being perceived as a diagnostic test and the lack of knowledge about the information NIPT testing provides. Pre-consultation videos should emphasize the difference between screening and diagnostic testing, clearly define the information one may receive from the test, and reiterate that women may decline this testing. Further research needs to be done in order to determine video specifics, but these elements are essential in addition to any other concerns ACOG or NSGC may raise.

Genetic Counseling through Telecommunication

Many patients throughout the United States do not have access to genetic counseling services due to their geographical location or the insufficient number of certified genetic counselors. The number of people receiving NIPT testing continues to increase, furthering emphasizing the insufficient availability of genetic counseling. As one general physician stated, "I hope the number of genetic counselors increases faster than the rate of people taking the test. Right now we don't even have enough."[159] When asked about the lack of genetic counselors, all doctors interviewed expressed that if it were feasible for every patient to see a genetic counselor, they would refer each patient to one. One doctor elaborated on this sentiment.

> "I think it would be helpful if all of our patients had genetic counseling as part of their initial prenatal visit so they can understand what these tests are and what they can mean. I

[159] Burt, Miranda. "Personal Interview." 11 Nov. 2019.

> know that that's just the ideal world… I think we're not good at streamlining or presenting available information for all patients."[160]

While all doctors and genetic counselors interviewed desired more genetic counseling assistance when administering NIPT, some "have to send patients too far and it takes too long" to see genetic counselors, or only had a few genetic counselors available relative to a large number of patients.[161]

Some physicians have combatted this problem through connecting their patients to genetic counselors by phone or through video platforms. One physician and three genetic counselors interviewed used telecommunication for increased genetic counseling access. While each said that in-person genetic counseling consultations would be preferred over telecommunication, the benefits of long-distance genetic counseling were clear. One genetic counselor discussed some of the benefits telecommunication has over in-person appointments.

> "Patients can talk in privacy of their own home, on the weekends… It's almost just as effective as in person. People are so appreciative to talk without waiting."[162]

All fifteen health care professionals interviewed agreed that increased access to genetic counselors via telecommunication would help ease the implementation burdens associated with NIPT testing. However, some did discuss the constraints with telecommunication. One genetic counselor who speaks to some patients about NIPT over the phone discussed these limitations.

> "I think it's always a more limited assessment. They are not gathering the patients' full medical history. Also, you don't have the option to follow-up. If you want to do an amniocentesis, you can't schedule it for the patient and facilitate further testing."[163]

[160] Burt, Miranda. "Personal Interview." 6 Dec. 2019.
[161] Burt, Miranda. "Personal Interview." 11 Nov. 2019.
[162] Burt, Miranda. "Personal Interview." 6 Jan. 2020.
[163] Burt, Miranda. "Personal Interview." 22 Nov. 2019.

Even with the limitations of telecommunication, the same genetic counselor expressed that it can be beneficial "for patients who would not have access to a genetic counselor or who wouldn't be able to get timely access to a genetic counselor."[161]

Many studies have been conducted regarding telemedicine. Genetic counseling by phone has been shown to be "as safe and effective" as compared to in-person counseling for cancer related genetic testing.[164] Access over the phone or through video platforms has been shown to have outcomes comparable to those in-person, produce benefits that outweigh potential problems, and increase access for patients who would otherwise be without genetic counseling services.[165,166,167] Furthermore, these services elicit a high level of reported patient satisfaction and increase in patient knowledge. [168,169] Some reviews have expressed concern about the economic viability of telemedicine; however, these concerns have been primarily related to physicians rather than genetic counselors.[170] Overall, the research around telemedicine has produced very promising and positive results. These studies, along with my personal research, support extending access to genetic counseling resources through telecommunication.

I am proposing that the American College of Obstetricians and Gynecologists (ACOG) require all practicing physicians to provide contact information for genetic counseling services to anyone who receives NIPT testing. For physicians who lack direct or easy access to genetic counseling services, this contact information should be for genetic counselors who are accessible

[164] "Hereditary Cancer." *XRAY: Genetic Counseling by Phone or Face-to-Face*, 29 Nov. 2017, www.facingourrisk.org/XRAYS/genetic-counseling-by-phone-or-face-to-face.
[165] Cohen, Stephanie A., et al. "Analysis of Advantages, Limitations, and Barriers of Genetic Counseling Service Delivery Models." *Journal of Genetic Counseling*, vol. 25, no. 5, 2016, pp. 1010–1018, doi:10.1007/s10897-016-9932-2.
[166] Sangha, Karan K., et al. *Journal of Genetic Counseling*, vol. 12, no. 2, 2003, pp. 171–184, doi:10.1023/a:1022663324006.
[167] Hjelm, N M. "Benefits and Drawbacks of Telemedicine." *Journal of Telemedicine and Telecare*, vol. 11, no. 2, 2005, pp. 60–70, doi:10.1258/1357633053499886.
[168] Sutphen, Rebecca, et al. "Real World Experience with Cancer Genetic Counseling via Telephone." *Familial Cancer*, vol. 9, no. 4, 2010, pp. 681–689, doi:10.1007/s10689-010-9369-y.
[169] Vrečar, Irena, et al. "Telegenetics: an Update on Availability and Use of Telemedicine in Clinical Genetics Service." *Journal of Medical Systems*, vol. 41, no. 2, 2016, doi:10.1007/s10916-016-0666-3.
[170] Ekeland, Anne G., et al. "Effectiveness of Telemedicine: A Systematic Review of Reviews." *International Journal of Medical Informatics*, vol. 79, no. 11, 2010, pp. 736–771, doi:10.1016/j.ijmedinf.2010.08.006.

by telecommunication. Providing contact information, but not requiring a genetic counseling consultation, allows patients to have the option for genetic counseling without making the service compulsory. Furthermore, simply providing contact information will not likely further saturate the demand for a limited number of genetic counselors, as a number of patients will presumably decline contacting a genetic counselor prior to testing or if they receive a normal result. For patients who do receive an abnormal result, a genetic counseling consultation should be strongly encouraged by the ACOG. The positive predictive value of NIPT, a measurement of how likely patients with a positive result actually have a child with Down Syndrome, ranges from 20% to 99%.[171] When patients are making a decision that may involve discontinuing a pregnancy, it is important that they understand the difference between screening and diagnostic tests as well as their relative risks and predictive value. Physicians believe genetic counselors are especially helpful at explaining the "different sensitivities and specificities with abnormal screening tests."[172] When a patient does have an abnormal result, strongly encouraging and providing access to genetic counseling will provide patients the opportunity to make a decision based on full and accurate information. I do want to emphasize that a higher number of genetic counselors are also needed in the United States. However, that is a much longer-term solution than using telecommunication to provide access to genetic counseling; nonetheless, more individuals are needed in genetic counseling roles.

Release of Low-Risk Statistics

More and more women are receiving NIPT testing, and the majority of this increase in demand is from "low-risk" patients. This increase in testing has not been accompanied by an increase in informational statistics for the low-risk population, and the lack of data has made it

[171] Burt, Miranda. "Personal Interview." 13 Jan. 2020.
[172] Burt, Miranda. "Personal Interview." 6 Dec. 2019.

difficult for health care providers to discuss relevant benefits and limitations with these women. As stated previously, one OBGYN discussed the lack of data for low-risk women.

> "I do mention that it is available to all patients. But I warn them that I don't have data for low-risk mothers. I still only have the sensitivity and specificity for women that I know that have a risk factor. I still don't have data for lower score women, many of the people that are routinely ordering these tests… So I can order this test on them, but I don't have a validated population to base it off of, which to me is kind of fishy. Again, the test has been out for a while, why don't we have that?"[173]

Other health care providers agreed with this sentiment, and expressed that the lack of data affects their ability to provide women with information to make a decision from these test results. Testing providers have not addressed the inadequacy of release of low-risk data. As previously stated, the positive predicative value of NIPT can range from 20% to 99%.[174] Without data, low-risk women do not know if their abnormal result might only have a 20% chance of being correct or is closer to being 99% correct. With many low-risk women making pregnancy decisions on the basis of an NIPT test result, it is imperative that these women have accurate data with which to interpret their test results. Until normative data is obtained, testing of low-risk worm should be prohibited.

The lack of data for these low-risk populations might purely be a financial decision for test makers. Many low-risk women do not receive insurance coverage for NIPT testing and pay for the test out of pocket. If a positive result for a low risk woman is much closer to only having a 20% chance of being correct, than many women would likely decline testing, leading to a significant revenue loss for testing companies. Physicians repeatedly emphasized the disparity in data between high and low-risk testing populations.

> "NIPT testing is well studied in patients who are high risk, but it is not a diagnostic test. I think we implemented it quickly without fully understanding the ramifications of its use,

[173] Burt, Miranda. "Personal Interview." 12 Dec. 2019.
[174] Burt, Miranda. "Personal Interview." 13 Jan. 2020.

> especially to the general population. The test is often misrepresented to low-risk populations."[175]

From my research, much of this misrepresentation can be corrected by a simple increase in available data for low-risk populations.

Testing providers should be required to release annual data on testing sensitivity and specificity for low-risk patient populations. Since multiple testing companies offer NIPT services and releasing data poses a financial risk, the policy would need to be executed legislatively to ensure compliance. Testing providers have the capability to make this data accessible, as providers already release these statistics for high-risk populations. These providers would be able to use similar collection methods and processes to assemble data for low-risk testing populations. The mandated release of data would be aggregated and non-identifiable as to not harm patient confidentiality. These statistics will help health care professionals better discuss and contextualize NIPT results with low-risk patients. Ultimately, low-risk patients will be better equipped to make an informed decision following test results.

The Urgency of Action

Since the expedited rollout of NIPT testing in late 2011, no public policy has been created to directly address its implementation. While some professional bodies and legislation have attempted to impact testing (see Policies Surrounding NIPT Testing in the Literature Review for more information), the reality is that NIPT implementation is largely unguided and unregulated. Lack of physicians' time, genetic counseling resources, as well as low-risk population data have led to the most glaring problems that arise from NIPT testing. These obstacles especially harm women who are uncertain if they want to receive testing, or those who

[175] Burt, Miranda. "Personal Interview." 6 Dec. 2019.

are attempting to make a decision regarding their pregnancy based on test results. As more women, especially those who are deemed "low-risk," undergo testing, these implementation problems will be exacerbated. The time for policy action is now. My policy recommendations directly address the root problems associated with testing, and these recommendations are based on proven research and interview data. To fully realize the benefits of NIPT testing, all parties need to have full and accurate information.

CONCLUSION

The development of non-invasive prenatal testing (NIPT) has allowed for pregnant women to receive more information earlier in pregnancy while offering little risk to the mother and fetus. However, the expedited release of testing has left NIPT with very limited oversight and regulation. Implementation varies across the US and the United Kingdom. These nations also have varying subsequent Down Syndrome abortion rates. NIPT is a screening test as opposed to diagnostic test, making the differences in implementation more alarming when discussing whether to continue or terminate a pregnancy. Various stakeholders—pregnant women, families, health care professionals, disability rights advocates, and reproductive rights advocates—will all benefit from implementing policy to guide NIPT testing. Without oversight, women will continue to make decisions on their pregnancies based on incomplete or inaccurate information.

Testing providers in the United Kingdom seemed to suffer most from a lack of training and knowledge surrounding NIPT testing. In the United States, continuing education courses and educational sessions conducted by testing providers were common. Closer examination may reveal that expanded provider education in the United Kingdom could help alleviate some of

their implementation problems. Focus must also be directed towards the United Kingdom when the UK National Screening Committee (UK NSC) presents their plan to implement NIPT testing in the nation's public health care system. Particular attention should be given to the impact of testing regulations on subsequent Down Syndrome abortion rates, which may help determine the impact that test implementation has on subsequent decision making. If these testing regulations are successful in increasing patient and provider satisfaction, the policy implemented in the United Kingdom could ultimately serve as a model for further policy decisions in the United States.

I believe that my policy recommendations will address the problems with NIPT implementation in the United States. Problems associated with informed consent, access to genetic counselors, and limited statistics for low-risk women will persist if not directly addressed. While my research has led to overarching policy recommendations, further research will need to be conducted in order to accurately detail these policies and determine if they will also be applicable in the United Kingdom or other countries. One area of further research in which I was not able to obtain sufficient data is the written materials provided by health care professionals to their patients regarding NIPT. Not all professionals I spoke to were willing or able to provide this information, but it is very likely that differences in written information provided may lead to differences in subsequent Down Syndrome abortion rates. This is an area that has yet to be researched, and may provide further insight into differences in NIPT implementation.

Many will attempt to make NIPT testing into a question of disability vs. reproductive rights. While one may hold personal opinions regarding subsequent abortions and NIPT testing, the ultimate problem lies in implementation. Until patients who undergo NIPT testing have

information that is consistent, unbiased, accurate, and complete, neither disability nor reproductive rights groups can clearly address subsequent abortions. Medicine should be about the patient; we need to hold health care professionals and test providers to a higher standard in order to safeguard patients who wish to make decisions using NIPT test results. Unless policy is enforced to guide NIPT testing, we will continue to be unsatisfied with inconsistent and substandard test results, and with the decisions that are based upon these results.

APPENDIX

List of Base Interview Questions

1. What is your current role with ____(place of work)? What led you to this position?
2. How long have you been a ____ (health care professional)? How long have you had experience with NIPT, and can you describe this experience?
3. To whom do you offer NIPT?
    a. Should this test only be offered to 'high risk patients' or everyone?
4. What week of pregnancy do you normally administer the test?
5. What information do you give to the pregnant women about early pregnancy screenings and invasive tests? What do most women know before you talk to them about the test?
    a. Do you tell them they are getting the NIPT test, or is this just part of blood sampling?
    b. Do you think patients know as much what they are going to do with the results of the NIPT test as those who undergo invasive testing?
6. Should the test be administered on the same day or on a return visit?
7. How much do you talk about the consequences of the test before administering it?
8. Do you offer pre-test counseling with a specialist healthcare professional?
9. Do you offer post-test counseling with a specialist healthcare professional?
10. Should women receive the same amount of counseling for NIPT as invasive testing?
11. What is your understanding of the expectations of the women? What do most women really want to know?
12. What is your personal opinion about administering the tests earlier?
13. What are the positive sides to the test?

    a. Are there any negative sides?

14. The term “sorting society” is often used in the debate. What do you think about that? Is it part of the debate here?
15. What do you think about the future with this test?
16. Do you think the small number of genetic counselors poses a problem for more widespread use of NIPT?
    a. What are your opinions on giving patients access to genetic counselors over the phone or through a video platform?
17. What do you say when delivering results?
    a. How do you deliver the result of an abnormal test? How much do you tell patients about the accuracy of a diagnosis?
    b. Do you direct patients to a follow-up test? If so, are women inclined to get another test?
18. What is your opinion on NIPT and Down Syndrome?
    a. Do you think this has become a “target” disorder?
    b. Do many women get tested specifically to look for Down Syndrome?
19. Do you follow-up with patients?
20. Do you have partnerships with other clinics or hospitals in the area?

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