MAT Medicine Anthropology Theory

POSITION PIECES

# Unsettling disciplinary frontiers

## An opportunity to address inequities in genetic medicine?

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## Abstract

Recent advances in genetic research provide anthropologists with an opportunity to reconsider the meaning and importance of interdisciplinary research. This piece suggests that interdisciplinary thinking can help to redevelop health policies aimed at improving access to new genetic technology and addressing many health care inequities. Drawing from research on access to genetic testing among women with a breast cancer diagnosis in the United States, I explore how patient perspectives can be used to redefine how policy makers interpret the utility of genetic medicine. Individuals undergoing genetic testing describe how genetic knowledge is translated into salient change in their lives, a view rarely recognized in conventional evaluations of genetic medicine. This work also recognizes how the 'potentialities' of genetic medicine both fuel the engine of ongoing genetic research and motivate individuals to imagine possible future actions to improve health. This reflection is meant to provoke debate and contribute to discussion about how health policies can be designed to improve inequities in access to genetic medicine.

## Keywords

genetics, health policy, breast cancer, uncertainty, interdisciplinary research

## Introduction

In a moment of unprecedented advancement in genetic medicine, genetic technology is increasingly used to treat disease with the aim of improving individual outcomes and

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transforming global health. Anthropologists observe that this evolving landscape of genetic medicine is producing complex challenges and novel opportunities for medical anthropologists to think seriously and reflexively about the meaning of 'interdisciplinarity' (Gibbon, Kilshaw, and Sleeboom-Faulkner 2018; Taussig and Gibbon 2013). Yet, the existing tension between social and life science viewpoints on the merits of genetic medicine often constrain the full integration of multiple disciplinary research perspectives.

Anthropologists are often at odds with the optimistic claims made by clinicians and life scientists working in the field of genetic research. We are hesitant to accept the hope and hype around genetic medicine largely because genetic research has not yet fulfilled its promise to radically transform medicine. Moreover, several anthropologists have emphasized the paradox of global expansion and ethical positioning of genetic medicine, which is framed in terms of social inclusion and justice, and the reality of inequitable and stratified access to this health technology (Gibbon, Kilshaw, and Sleeboom-Faulkner 2018; Taussig and Gibbon 2013). My own research on inequitable access to genetic testing among women diagnosed with breast cancer validates these assessments and is useful in predicting how these inequities might translate to other cases of biotechnology and genetic medicine.

Rather than parsing out disciplinary points of disagreement on the successes or failures of genetic research, 'interdisciplinarity' hinges on our ability to think across boundaries and recognize areas of mutual agreement on the benefits of genetic medicine. Researchers across many disciplines agree that we must address inequities in access to genetic medicine in order to expand the use of this biotechnology. I argue that interdisciplinary thinking is particularly important for reimagining health policies that will improve access to genetic medicine. Interdisciplinary approaches to genetic medicine can become a path towards achieving our common goal of improving population health.

Conventional clinical and health policy perspectives on the utility of genetic medicine assume that the technology will be 'cost-effective' and used to make actionable decisions around disease prevention or intervention. Current policy evaluations on the costs and benefits of genetic medicine do not take into account patient perspectives. In this piece, I explore how these conventional perspectives provide only a narrow view of the benefit or value of genetic knowledge for individuals who experience genetic testing. Developing health policies that include patient perspectives on genetic testing alongside conventional approaches is necessary to fully recognizing how genetic knowledge is translated into salient benefits for individuals and is a critical piece in efforts to broaden access to genetic medicine.

Drawing from my research with women who experienced a breast cancer diagnosis, I reveal how genetic results are a key part of making more informed decisions about breast cancer treatment. Genetic results gave these women greater agency to act in health promoting ways.

This illustrates how genetic knowledge is empowering and diminishes anxieties about the possibility of another cancer diagnosis in the future. Therefore, the value of genetic knowledge does not necessarily stem from the possibility of a measurably improved outcome in health, but rather from the confidence gained in making informed treatment decisions and the opportunity to form a deeper understanding of self.

Emphasizing the immediate utility of genetic medicine also means that, as a society, we run the risk of overlooking how genetic 'discovery' generates new uncertainties and research questions that drive future biomedical research endeavors. Individuals undergoing genetic testing often receive some 'uncertainties' with their genetic results, which is caused when there is insufficient evidence to classify a gene variant as either detrimental or neutral. I find that many research participants perceive these genetic uncertainties as something with 'value' because these results may become clearer and serve a health promoting purpose sometime in the future.

To further interrogate the benefits gleaned from 'uncertainties', I employ the social science concept of 'potentiality', which recognizes how the uncertainties produced from genetic knowledge are a type of insight embedded with latent qualities that may realize future utility (Lee 2013; Taussig, Hoeyer, and Helmreich 2013). Potentiality creates a space to consider how the benefits of genetic knowledge transcend the clinical encounter and retain their value to improve individual health at some unknown future point. Potentiality is not only a useful tool for social scientists to examine how individuals interpret genetic uncertainty, but can also be valuable in creating interdisciplinary insights on the benefits of genetic medicine as something more abstract and not necessarily immediate and measurable. In other words, genetic knowledge offers a form of 'social utility', rather than clinical and immediate utility (Stivers and Timmermans 2017), and can provide an alternative view on what we mean by the 'utility' of genetic testing.

## US health insurance policies and emerging inequalities

For US health insurance companies and other organizational bodies tasked with overseeing policies on the clinical utility of genomic services, a genetic test should lead to measurable changes in prevention, treatment, prognosis, or disease management (Stivers and Timmermans 2017). As an example, oncologists are increasingly using the results of genetic testing for women with new breast cancer diagnoses, along with other diagnostics, to further classify the tumor, fine-tune the cancer treatment strategy, and provide the patient with a 'personalized' therapeutic strategy. This approach is clinically referred to as 'treatment-focused genetic testing' because the results presumably have immediate and direct implications for the management of the disease. Treatment decisions are based on knowledge of specific genetic

variants that are 'clinically actionable' and have approved therapeutic interventions (Narod 2018; Stivers and Timmermans 2017). For example, genetic results can inform decisions such as: choosing between less invasive surgery to remove the tumor (e.g., lumpectomy) and more invasive surgical options (e.g., mastectomy, double mastectomy, etc.); determining chemotherapy options; and developing prognosis and risks for future cancers (Yadav et al. 2018; Katz, Kurian, and Morrow 2015).

In the USA, health insurance is the primary gatekeeper to most health services and treatments and yet previously, very little was known about how different types of plans (e.g., PPO, HMO, Medicaid, etc.) impact access to specific types of health care, including genetic testing. I set out to investigate the role of health insurance in access to care through a case study of access to genetic testing among women diagnosed with breast cancer. My research confirms prior findings indicating the high variability in health insurance coverage of genetic testing across different types of health insurance payers and plans (Phillips 2018). Although genetic testing among newly diagnosed cancer patients can be a cost-effective strategy for treating breast and other forms of cancer (Tuffaha et al. 2018), health insurance payers remain hesitant in changing coverage policies to improve reimbursements for genetic testing. Hesitation on the part of health payers primarily stems from health policy assumptions that new medical technology will be a major source of financial risk. From the health insurance perspective, coverage of genetic tests should only be available for individuals where there is clear evidence that the results are medically 'necessary' and the technology is proven to be 'efficacious' in treating disease.

This constraint can be traced to the rapid advancement of genetic technology, which generates a lag time in the revision of health policies to accurately reflect updates in clinical literature and actuarial data. This lag time in payer coverage policy revisions contributes to the uneven adoption of new coverage policies on genetic technology and unequal diffusion of genetic medicine into clinical practice (Lakdawalla, Malani, and Reif 2017). This disproportionately benefits individuals with higher levels of socioeconomic status who often have better-quality health insurance (i.e., more comprehensive coverage and lower out-of-pocket costs) and are more able to access this genetic testing. Therefore, addressing this problem requires reassessing the evidence used to revise health insurance policies on genetic testing.

## Patient perceptions on the benefits of genetic testing

Patient insights on the advantages of genetic testing are rarely included in clinical literature on the efficacy of genetic medicine that is translated into a clinical setting. Yet, I find that when individuals have the opportunity to reflect on their breast cancer treatment, they have a strong understanding of the 'actionable' benefits provided by genetic results. Most of the women

participating in my study underwent genetic testing after they were diagnosed, including Shonda, who shared:

> After my diagnosis I go see the oncologist, I was thinking chemo the whole way. But he said: 'We need your genetic testing before we can really tell you what type of treatment'. With no family history or anything else, and you know, what do they have to go on other than the genetic testing?

Shonda tested negative for gene mutations associated with breast cancer. Equipped with this knowledge, her oncologist felt she could safely forego chemotherapy or radiation following surgery to remove the cancerous breast tissue.

In other cases involving participants who tested positive for a *BRCA* mutation or other genes with known associations to breast cancer, genetic results supported their decision to take a more aggressive treatment approach. Women with a *BRCA1/2* mutation are at a higher risk of future breast cancer recurrence (Grindedal et al. 2017). Correspondingly, most of the participants who knew they carried a *BRCA* mutation elected a contralateral prophylactic mastectomy as part of their treatment strategy. Savannah had already experienced chemotherapy and a lumpectomy as part of her breast cancer treatment when she received her genetic results. She was 'blown away' by the news that she had a *BRCA1* mutation and, although the thought of having another surgery was almost unimaginable, she also knew what her chances of a recurrence would be if she did not have a contralateral prophylactic mastectomy.

In sum, almost all of the twenty-four participants received genetic testing and felt the experience was an empowering component of their treatment decision-making process. Their perspectives corroborate clinical research demonstrating the efficacy of treatment-focused genetic testing in the improvement of individual disease outcomes (Kurian et al. 2018). Still, health payers remain unconvinced at the medical benefits of genetic testing for all newly diagnosed breast cancer patients. This is likely because clinical research evaluating 'successful' translation of genetic knowledge to clinical utility is based on associated mortality outcomes from breast cancer.

This 'cost-effective' approach to evaluating the clinical benefit of genetic testing for breast cancer patients fails to capture how genetic knowledge facilitates more informed decisions about breast cancer treatment and consequently improves the mental and spiritual wellbeing of cancer survivors. This point is substantiated by women who did not have genetic testing before starting their cancer treatment, all of whom appeared less confident in their treatment choices. For instance, Simone underwent genetic testing at the conclusion of her treatment

and her results indicated that she had a *BRCA2* mutation. Simone commented that if she had known this earlier, she 'probably would have made a different choice and had a double mastectomy'. Comments like Simone's allude to some of the lingering uncertainties felt by individuals who have experienced cancer and associated emotional distress (Clayton, Mischel, and Belyea 2006; Tewari and Chagpar 2014).

## Potentiality and genetic uncertainty: Future benefits of genetic knowledge

Not all of the genetic knowledge gained from genetic testing is used to make 'actionable' and immediate treatment-related decisions. Rapid advances in 'next-generation' genetic sequencing have made it possible for geneticists to simultaneously analyze many different genes and today's new gene panel tests include more than 100 genetic variants (Afghahi and Kurian 2017). These panel tests will include genes like *BRCA1* and *BRCA2* that have clear clinical guidelines, but also incorporate gene variants that are likely unrelated to breast cancer or lack sufficiently robust clinical data to guide patient care (Afghahi and Kurian 2017). Despite this shortcoming of panel testing, cancer geneticists argue that panel testing offers more accurate information about genes associated with cancer and will improve our appreciation for how these other genes are associated with risk of cancer (Kraus et al. 2016). Ultimately, genetic researchers view these limitations of genetic testing as crucial in the advancement of genetic research and are optimistic this approach will yield the most future benefit.

To some extent, study participants who had a gene panel test did say they had some lingering questions about the meaning of their genetic results; however, patients also echoed the voices of geneticists and clinicians who expressed an overwhelming sense of optimism that genetic uncertainties can unlock future health breakthroughs. These women were accepting of uncertainties because 'uncertainty' is a common theme in their breast cancer journey and something that could never be fully eradicated (Hall, Mishel, and Germino 2014). They found more meaning by focusing on the 'potentiality' of genetic uncertainties, opting for a sense of hope that this genetic knowledge will continue producing health benefits in a way that does not yet – and may never – exist (Taussig, Hoeyer, and Helmreich 2013). The potentialities of genetic knowledge open up new territory where individuals hope to 'discover' value from inconclusive genetic results that has yet to be uncovered.

Women who had completed breast cancer treatment viewed inconclusive genetic results as an indication that health surveillance outside of conventional 'post-cure' follow-up care was needed. For instance, Bernice tested negative for mutations in genes that have a known association with breast cancer, but her results indicated a mutation on a gene variant with an unknown significance. Surprisingly, this knowledge brought Bernice comfort because the

result 'made sense' given the extensive history of cancer among members of her family. Bernice was able to construct a self-narrative around *why* she was diagnosed with breast cancer, which resolved some of her prior concerns that she was responsible for causing her cancer (Frank 1995; Kaiser 2008). Bernice regained a sense of control over her health that she had not felt since before her breast cancer diagnosis. She was hopeful she could prevent future cancers by conducting her own research on the gene variant with 'unknown significance' to see if new insights emerge establishing a link to breast cancer.

Winona, an oncology nurse, also tested positive for several genes of 'unknown significance', although her professional experience and extensive family history of breast cancer provoked a strong inclination that her breast cancer was hereditary. Like Bernice, she wanted to increase her knowledge of genomics to better understand her genetic results. Winona continues to schedule semi-annual appointments with her genetic counselor hoping these visits will provide knowledge useful to preventing a future breast cancer diagnosis. She explained, 'I did the full panel [genetic] test and an amazing geneticist' and, because of the inconclusive results of her panel testing, Winona 'really wanted ongoing follow-up. If something else changed and there was some other surveillance or treatment I could do, I would do it'. Winona holds no expectation that genetic knowledge will provide answers overnight and is willing to stay in this 'moment' of potentiality for as long as necessary.

Potentialities of genetic testing are most salient in the ways that women perceive the benefits of genetic knowledge for other members of their family. Even when the results of genetic testing did not have actionable consequences for their breast cancer treatment or were riddled with uncertainties, all of the women I spoke with unanimously agreed that the experience was important because of the implications for their family. Families seeking genetic knowledge from a place of uncertainty again opt for the potentiality of the unknown as a point of departure for action; but, now this action is understood within the family and social context where health and illness is experienced (Stivers and Timmermans 2017).

For some participants, because family members are newly equipped with information about their family medical history, they can access health insurance coverage for genetic counseling and testing services. In addition to immediate family members, participants reported that more relatives, including distant cousins and aunts, accessed testing and in some cases were able to take preventive action to avoid a future cancer (Katapodi et al. 2018). Potential benefits of testing among individuals and families dovetail with clinical research demonstrating that current criteria for genetic testing is insufficient to capture individuals with a predisposition to hereditary forms of breast cancer, which could prevent cancer in other family members (Chen et al. 2018).

I found that individuals describe other ways of finding meaning in genetic uncertainties based on family conceptions of what 'runs in the family' and who in the family is already considered 'at risk'. Prior research interrogating the possibility of genetics to transform understandings of health show that the implications of results extend beyond the individual, especially when results are shared with family, creating new biological ties between family members (Lock et al. 2007). Rather than creating anxiety and tension, ambiguous genetic results have the ability to become socially actionable, which alleviates feelings of concern (Stivers and Timmermans 2016). Social actions are many times similar to those taken by individuals who continue to research and make meaning out of genetic uncertainties. What differs is the communal sense of agency among family members who seek out additional health information that perhaps can lend some insights to the genetic results, if not today, then maybe sometime in the future.

## Conclusions

An interdisciplinary lens shows the value of genetic testing beyond the conventional assumption that results must be clinically actionable. Genetic knowledge provides significant benefits that often go unrecognized by traditional evaluations of the merits of genetic technology. The imperfections and compromises of actual medicine have become a powerful and a motivating force to ongoing investments in human and financial resources towards biotechnology (Mrig and Spencer 2018; see Warren and Addison, this issue). My research shows that the uncertainty of genetic medicine also motivates patients to continue to invest their time and financial resources into making meaning out of their results.

For participants who found the testing beneficial even without 'conclusive' knowledge about the meaning of results for their health, the promise of genetic medicine has already been partially fulfilled. This suggests that individual expectations around the 'success' of genetic medicine is incongruous with how 'success' is perceived by geneticists, clinicians, or even health policy makers.

My research shows that integrating genetic testing into treatment increases the frequency of patient engagement with biomedicine. This recurrent engagement with biomedicine occurs through multiple pathways, including ongoing surveillance and clinical engagement. For this reason, critical social science perspectives remain a necessary component for appreciating the ethical and social implications that come with the expansion of genetic medicine into clinical and global health settings. Still, recognizing interdisciplinary views on the benefits of genetic testing is more than a productive exercise to overcome tensions between life and social science perspectives; it also broadens our understanding of the 'utility' of genetic results for women with breast cancer and deepens our appreciation for the salience of genetic knowledge for individuals and their families. This effort may serve as an innovative opportunity to create

more equitable health policies to increase access to genetic medicine and lead to our common goal of improving global health.

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