Difference in the making: Biomedical Futures and Health Equity

Ramya Rajagopalan
Life Sciences Foundation / UW-Madison

Abstract

Science and culture are in constant, inseparable dialogue; theories of human biological difference emerging from scientific practice shape and are shaped by cultural and political frameworks for making and doing difference. Historically, certain groups have been subjugated, dominated and disadvantaged as a result. But difference has an epistemic politics. How difference is coded, framed, and understood is always contested, modulating the role that differences come to play in political and cultural spaces.

In recent decades, biomedical research has greatly expanded its focus on DNA, developing conceptual frameworks that link differences across culture and biology, via diseases and genes. The turn to "precision medicine," tailored treatment based on individualized genetic information, has amplified the focus on DNA differences. This turn offers a fruitful analytic site to consider what kinds of difference remain salient in biomedicine, and with what implications. How can precision medicine be used to build responsible, equitable and just frameworks for talking about genetic and health differences, without reifying, stereotyping and marginalizing groups along common sociocultural axes? How can we ensure that the benefits of new tools for personalizing medicine are equally shared? And with increasing floods of "big data," how can we ensure that genome information is accessible and empowering to patients, rather than disenfranchising? Decentering dominant discourses around disease and difference can bring to light more nuanced frames for addressing health disparities and health rights, especially for those most marginalized from the benefits of health advances.

Keywords:

Race, precision medicine, scientific practice,

Introduction

Media reports in the last several years have been full of hope for new advances in medicine, with predictions that this, the "century of biology," will finally see the defeat of debilitating chronic diseases like cancers and diabetes. New medical devices, mobile health technologies, and DNA analysis promise leaps forward in clinical care, to a future time when our medical treatment will "truly" become personalized." Precision medicine is animated by the idea that

information about our genomes will transform clinical care, that it will tell us about our disease risks, and help us find the best treatment for each individual.

These promises are so tantalizing they have even attracted the attention and endorsement of heads of state. Earlier this year, President Barack Obama announced his administration's $215 million commitment to a new initiative for personalized health in his January 20 2015 State of the Union Address:

> I want the country that eliminated polio and mapped the human genome to lead a new era of medicine—one that delivers the right treatment at the right time. Tonight, I'm launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes — and to give all of us access to the personalized information we need to keep ourselves and our families healthier.

Precision medicine is predicated on the idea that each individual has unique disease susceptibilities, and responses to drugs, which are partially mediated by their unique genome sequence. Marshalling this idea, human geneticists have focused on genomic similarities and differences in particular ways, constructing theories and knowledge of "human genetic variation" that is shaping the ways in which we think about human diversity. These developments offer a fruitful analytic site to consider what kinds of differences remain salient in biomedicine, and with what implications.

"Science," as a social, political and cultural endeavor that attempts to make sense of the world, does so through a set of established tools, methods, norms and standards of practice. Science authorizes what types of knowledge carry weight in the important policy decisions of the day. Its imprimatur determines what kinds of knowledge about the "human" and about "nature" count. Knowledge produced by science interacts and often collides with other kinds of knowledges, and these encounters shape the governance and trajectories of our social, political, legal and even cultural institutions. Given the authority that science commands, it is useful to ask how knowledge and "facts" are produced by scientific activity in different historical moments. How is this knowledge mobilized to do "work" in the world? What are the

consequences, and for whom? These are questions that have long formed a locus of interest within STS.

Of critical interest in STS is how contemporary science constructs knowledge about human difference. Societies "make," "do," and "act" difference, in different ways. For example, there is broad consensus (although not absolute) across the social sciences that the concept of "race" is a sociocultural one, built through historically situated and contextually localized political and social processes. Race identification is fluid, both institutionally and individually. Census race categories and definitions of race and ethnicity have changed dramatically through the decades in countries like the United States, Brazil and South Africa. The way that individuals identify by race is also dynamic, partly informed by these institutional changes, and partly by changes in what the various terms and categories come to mean over time.

In contrast, some corners of science have through the centuries maintained an interest in explaining human race differences in terms of biology, and have carried into their science the political and social influences of their time. There are ample historical examples that illustrate how "scientific facts" have been used to support sociopolitical logics of difference. Race-based human classification schemes were first created by the natural historian Linneaus and his student Blumenbach in the 1700s. They organized people by continent of origin, and the traits and characteristics they assigned to each continental race clearly valued white Europeans as being the human ideal. Both were respected men of science, and their work lent a mantle of scientific authority to cultural assumptions about race difference. Their science was both informed by, and used to justify, European dominance and systems of slavery during the colonial expansions taking place at the time. Two centuries later, political regimes in Nazi Germany and Apartheid South Africa used similar value-based classifications of humans based on anatomy, cranial structure and even hypotheses about intelligence were to authorize their systems of white superiority and genocide. Elsewhere, scientistic ideas about racial inferiority informed eugenics campaigns, for example in the early-to-mid 20th century in the United States.

These forms of science promulgated a logic of biological essentialism, with respect to human difference. They suggested that race classifications, rather than being a dynamic set of ideas produced by the activities of and embedded in human social institutions, were biologically innate, natural and timeless. They effectively absolved human social institutions of any complicity in the making of human difference, and in the value judgments that were attached to these differences were also "naturalized," that is, made to seem natural and immutable.

Today, science continues to play a significant role in what has become a longstanding debate about the very nature of human difference. New developments in biomedicine and genetics are once again positioning difference at the center of debates about the "human," this time, in the context of health and disease. Although the views from science are more nuanced, and bear in mind the historical injustices perpetrated in its name, there remains much scope for critical reflexivity about their implications. Contemporary views about genetics and human difference seek to distance and distinguish themselves from past injustices but, without care, could in the process solidify present ones.

Our concepts of difference, whether they be census race categories or ideas of human biological variation, are inseparable from the "epistemic politics" that create them. How can STS approaches help illuminate the "epistemic politics" of contemporary difference making in genetics? Like all science, contemporary genetics practices shape and are shaped by common sociocultural understandings of the worlds we inhabit. Understanding these processes is critical to the project of findings ways to intervene, and ensuring equitable access to the benefits of new medical technologies, instead of concentrating their harms in groups who have long been shouldering the burden at the margins.

To that end, ethnographic studies of the spaces in which researchers enact these knowledge-making processes can reveal how genetic understandings of health and human diversity are made, by whom, and under what assumptions. The particular configuration of knowledge-making practices employed affects the kinds of knowledge made. In other words, process

affects product. The ethnographer can open the "black box" of process, to expose contradictions between what is done and what is reported to have been done, and analyze what is claimed as a finding, among the many possible frames for interpreting the data. They also open the possibility of imagining alternative frames for studying, understanding and addressing human health, in the hope of achieving more equitable outcomes. The analyses in these paper are based on ethnographic fieldwork I have done since 2008 in laboratories that use genomics to study clinical medicine and human disease in various circumscribed population groups.

What is Precision Medicine?

The concept of "precision medicine" (also known as "personalized medicine" and "genomic medicine") began to gain traction after the completion of the Human Genome Project (HGP). The HGP employed thousands of scientists in North America, Europe and the East Asia, all focused on producing a single human genome sequence, an open resource to aid the research community's search for the genetic factors involved in diseases. Sequencing the human genome during the HGP cost an estimated $1 billion USD and took thousands of researchers about ten years to complete. Today, with the latest DNA sequencers and desktop computers, a human genome can be sequenced for a few thousand dollars, in a few days. Within a span of twenty years, DNA sequencing efficiency has skyrocketed while costs have plummeted to the point where the "clinical genome" has, we are told, come within reach.

The clinical genome would work something like this: at some to-be-determined point in the future, it will be possible for you to walk into your doctor's office and have your DNA sequenced on the spot. As part of your routine blood tests, your DNA would be sampled and sent to a benchtop genome sequencer for immediate analysis. Your entire genome would, almost in real-time, be uploaded as a permanent part of your electronic health record for you and your physician to consult, a compendium of predictions about your disease susceptibilities and drug tolerances, and accompanying guidelines for prevention or treatment.

The clinical genome has gained so much traction it has now routinely discussed in laboratory and clinical arenas and the popular press as an inevitability. But the clinical genome remains largely an idea, still under development. It is predicated on a future which stubbornly continues to evade the reach of genetics, a future in which the clinical significance (if any) of the different parts of the genome are known and actionable, and can be used to guide treatment. But so far, genetics has had little success in explaining the causes of most diseases. Still, the allure of "precision medicine" approaches remains a dominant force in the search for ways to revitalize the American health care system and improve the general health of the population. The Obama administration's Precision Medicine Initiative, as it has come to be called, is a relative latecomer to the stage, just one project among many large-scale efforts since 2008 to harness the power of DNA sequence and big data analytics in medicine. Population DNA sampling is underway at large biobanks being built by private health systems, research hospitals and clinics, and biotechnology companies, often working collaboratively.

As a result of these efforts, "precision medicine" has become a catchword that is being applied to a whole range of tools, methods and technologies that plug genetic information into other kinds of health information to predict, prevent or treat disease. Some examples include: genetic tests for predicting susceptibility to cancer and other diseases; genetic tests for drug intolerance; and networked "big data" repositories, interfacing medical health histories with real-time data uploads from mobile "wearable" technologies which monitor and measure heart rate, breathing and sleeping patterns, and activity.

Precision medicine has been widely heralded as an approach that will "democratize" medicine, giving patients access to their entire genome. New DNA-based technologies promise to empower patients with information about their molecular selves. Armed with all of that genomic sequence information, patients are promised more choices, more input into the decision-making processes that will guide their health care and treatment options. At the same time, precision medicine has emerged from genomics studies that have sought out biological

differences at the level of DNA, and in certain corners has organized these differences along human groups that are often interpreted as race groups. As described in the next section, these ways of organizing human variation have consequence for how we think about the nexus of health, disease and human difference.

Making and Doing Difference in Contemporary Genomics

Precision medicine's promises pivot around the idea that every individual has a unique set of disease susceptibilities, and that these can be uncovered by triangulating risk elements in their genetic makeup and in their daily environment, be it diet, physical activity, medications etc. (Some epidemiologists, but not usually geneticists, consider a more expanded repertoire of environmental stimuli, including work conditions, toxic exposures, etc., and some are active against ideas of biological or racial essentialism, showing instead how experiences of racial inequality and discrimination, and not race itself, are correlated with poor health outcomes (Geronimus and Thompson 2004, Krieger and Bassett 1993, Krieger 2011)).

Biomedical geneticists argue that genetic variation is medically important knowledge, that it will help explain why chronic diseases affect some people and not others, why some patients respond better to specific treatments than others. In order to be able to understand how *your* genome works, researchers have spent much time, effort and money, trying to understand to what extent humans differ at the level of their DNA, and what the clinical significance of these differences are, if any. This so-called genetic variation has been extensively catalogued in a few people from around the world, but what any of this DNA sequence information means for human health and disease is still largely to be determined. Thus the turn to "precision medicine," tailored treatment based on individualized genetic information, has amplified the focus on DNA differences.

Over the last several years, biomedical geneticists have conducted studies to examine possible links between DNA and disease on an unprecedented scale, using newly developed DNA

genotyping and sequencing technologies to search the genome. These techniques have also allowed researchers interested in human genetic variation to begin to catalogue the range of genetic similarities and differences that exist across individuals from around the world, through for example the International Haplotype Mapping Project.

Precision medicine efforts are trying to leverage these data on human genetic variation in order to tailor and individualize diagnoses and treatments for use during routine medical care. To this end, researchers have focused on characterizing genetic differences between individuals and groups. Researchers refer to the genomic sites where these genetic differences occur as SNPs (single nucleotide polymorphisms) and CNVs (copy number variations). These "genetic markers" of human variation are thought to comprise only a tiny proportion, around 0.5 percent, of the roughly 3 billion bases that make up a human genome. Still, the prevailing argument is that these small differences will prove to be medically relevant.

Some individuals may share genetic markers in common, such as a particular genetic variant that might be related to a particular disease. One challenge researchers have faced is how to select, delineate and circumscribe "groups", and categorize the genetic variation within those groups, in ways that might be meaningful for medical purposes. They have done so within the context of the dominant discourses and knowledge-making practices within medicine and science, which have produced and accumulated specific ideas about the nature, diversity and extent of human differences and their significance for health and disease.

Focusing on SNP differences, geneticists have mobilized theories of human evolution which hold that early in human history, small groups of people migrated out of Africa and onto other continents, where they expanded in relative isolation from other humans. Over millennia, groups tended to reproduce mostly with their neighbors, creating gradual patterns of variation across the world. They argue that these differences persist as signatures in human genomes, although different methods make visible different, sometimes contradictory, patterns.

As medical geneticists have tried to locate explanations for diseases in DNA, and specifically in the small proportion of the genome where DNA differences occur, they have mobilized these theories and practices of circumscribing particular groups. In their efforts to link genetic differences to disease, some researchers have claimed they can also distinguish differences in DNA that are attributable to what they call "geographic ancestry" – that is, the geographic locations where a person's ancestors came from. Some, like NIH-director Francis Collins and genomics entrepreneur Craig Venter, as well as other prominent medical geneticists, argue that these differences represent the complexities of a person's ancestry, and are not diagnostic of a person's "race." The conclusion from these quarters is that DNA differences do not correspond to the crude and homogenizing topologies of racial categories. But a minority of genetics researchers argue that these geographic differences do resemble membership in different groups defined by continental race. This view has gained currency in the popular press and even in so-called DNA ancestry testing companies that purport to be able to tell an individual their percentage of each continental ancestry from America, Europe, Asia and Africa.

Such claims, which attempt to link genetics, race, and disease, present troubling challenges for sociological understandings of race as a set of historically-situated political and cultural processes created and used by societies for difference-making. When ancestries are described in terms of continent of origin, for example, European, African, and Asian, these labels are based on socioculturally defined U.S. categories of race and ethnicity, such as white, black, and Asian. They are not "natural" descriptors "out there" in the genome, but are projected on to it. On the one hand, some geneticists argue in liberal democratic fashion that the genome will take us "beyond race" to other forms of group-making based on our genes and genetic susceptibilities. On the other hand, some argue that the genome bears the imprint of race, a move which might serve to naturalize and reify race as somehow biological and immanent in our DNA, rather than as a set of sociocultural constructs humans use to organize their worlds. Contemporary genetics, then, like all scientific activity, echoes the varied ideological strains of the prevailing social and political moment.

The Problematics of Genetics and Race

To some within the genetics research community and to scholars who study the activities and practices of this field, links between genetics and race are tenuous. But race is of broad cultural interest, and, according to many geneticists, media reports often over-interpret scientific reports in order to generate bigger headlines. They read race in, where none necessarily exists, further embedding an essentialist conception of race in the popular consciousness. Science writers, like Nicholas Wade, have misread genetic science and popularized the unsupported view that races are genetic, and further, that these genetic differences can explain why some nations have industrialized while others have not. These writings in the popular press feed into the perception of science as an authority on human difference, and endanger our collective ability to understand genetic difference and race differences as distinct (and different) sociocultural and sociotechnical constructions. The translation of science is left to the media, rather than to the authors of the studies, whose interpretations of the findings are usually more nuanced and complex. Indeed, geneticists would agree that no single pattern of genetic variation is completely diagnostic of affiliation with any particular race or ethnicity for any particular individual. However, some argue that in the aggregate, genetic variation is patterned in a way that resembles race, and they assert that these correlations might be useful for developing biomedical and clinical interventions.

Deconstructing how this knowledge is generated reveals that the links between genetics and race are not at all as clear-cut as media reports might have us believe. First, several studies have shown extensive genetic variability among individuals who self-identify with any particular group defined by sociocultural or census race categories. In effect, racialized groups are very genetically heterogeneous. That genetics can "diagnose" race, or that there is a 1:1 correspondence between race and genetics, is unfounded according to most geneticists' interpretation of the data from genetics itself.

Second, genomic studies typically use a relatively small subset of individuals who claim membership within any particular continental ancestry or race grouping. Conclusions about genetic patterns and any perceived resemblance to race based on small sample sizes cannot be extrapolated with any reliability to a large, genetically heterogeneous race-based grouping of people. Any extension of study findings to others who identify within these broad groupings could be fraught with problems of accuracy and precision.

These and other lines of evidence strongly indicate that there is no neat correspondence between genetic variation and one's assumed race or ethnicity (Fujimura et al, 2014). In other words, genetics does not map onto race, and vice-versa.

The disagreement is an interpretive one. No one disagrees that there are genetic differences among humans. There are even genetic differences between parents and children, and among siblings. The question is, how should we think about and conceptually organize such differences? What are alternative ways to code genetic differences? Is there any justification for coding genetic difference by race? Or is simply convenient to do so, given the prominence of race differentiation in social and political life? Race's troubled history, and the view among many genetics researchers and social scientists that genetic differences do not correspond well to race categories, are compelling reasons to discontinue raising the spectre of race in genetic research.

This decoupling of race and genetics becomes especially urgent in light of current evidence regarding the nature of disease. When it comes to the common and complex diseases of current interest in medical genetics, genomics studies have so far found no easy links between race, DNA and disease. Indeed, compelling associations between DNA and most of these diseases, leaving race out of the equation, has been scant. This may not be surprising, given that these "complex" diseases have long been suspected to be closely tied to one's environment—factors such as diet, exercise, smoking and drinking habits, air and water quality, standard of living, etc.

The biological essentialisms underlying the conflation of race and DNA have stark implications for genetic medicine going forward. Many worry that the new technologies being used to develop precision medicine may also become technologies that are used to define "genetic signatures" for, or "genetic stereotyping" of, different racial or ethnic groups. This aspect of precision medicine, if developed and nurtured into broader clinical use, will popularize the misconception that it is possible to infer underlying genetic makeup from an observer-defined or self-reported race or ethnicity, when even proponents of using race in genetics research argue that this will not be possible. Still, this scenario recalls some of the past attempts to link race and biology, e.g., the eugenics movements of the early twentieth century.

Indeed, race is not new to medical decision making in the U.S. For decades, it has been common practice among American medical clinicians to use race as one factor among many when deciding among possible diagnoses and treatments. There is a long-standing view in medicine that certain diseases travel more frequently in certain racial groups and a belief that certain groups may respond better (or worse) to certain treatments than others. However, epidemiological evidence of racial differences in disease incidence is not evidence of race-specific genetic susceptibility to disease. Many studies have shown that social factors that differ across race, including socioeconomic status and experiences of discrimination, contribute significantly to racial differences in disease incidence.

Many generalizations about the relationships between race and genetics, now part of popular understanding and medical training programs, grew out of late-20th medical genetics studies. For example, medical school and college biology curricula continue to propagate the idea that some single-gene, highly heritable diseases, like Tay-Sachs disease or sickle-cell anemia, are prevalent in only certain groups—as in Jewish and African American groups, respectively—than other groups. What is not well-known is that Tay-Sachs has also been observed at high prevalence in non-Jewish groups in Quebec, Canada and that sickle-cell and other hemoglobin disorders are common in many groups around the world. The misconception that a particular

disease like sickle-cell is specific to African Americans may lead to patients being misdiagnosed, or diagnosed too late in the progression of disease simply because they are not of the ethnic group "marked" by the disease. There are concerns that findings in precision medicine, similarly based on genetic difference, if organized by race groups, may have adverse medical consequences, and precisely for those groups most marginalized, disenfranchised or constrained by the inequalities endemic to American healthcare.

Will these new genomics technologies be used to support the idea that differences among individuals, when grouped along racial or ethnic categories, are medically relevant? That is, will contemporary genomics give race a new prominence in medicine? Using genetic patterns as readouts of one's "real" ethnicity or race makes no sense when racial and ethnic groups are social categories constructed within specific historical and cultural situations and not based on genetics. Methods now exist whereby genetic variation may be interrogated at the level of individuals. Why use these methods to harden the lines between racial groups, especially since race groups are not genetic groups, and then use these groups to organize "precision medicine"? This defeats the aim of personalized medicine, which is to tailor treatment so it is more effective for each individual's specific combination of health factors.

Parsing disease susceptibilities by continental ancestry or race groups and using race as a proxy for underlying genetic variation in medical decision making may have negative consequences for individual health and well-being. For example, it could lead doctors to incorrectly treat patients who they assume to be part of the group(s) with genetic predisposition to the disease in question. Such generalizations may also be detrimental to groups not directly implicated. For example, associations between specific diseases or genetic signatures and particular groups may result in a higher chance that the correct diagnosis of the disease in question will not be made for patients assumed not to be part of these groups.

For these reasons, many medical professionals and scholars are worried about the implications of the use and misuse of race in the new clinical pathways being pursued by advocates of

precision medicine. Using race as a lens through which to determine treatment decisions could (and has) opened the door to instances of misdiagnosis or incorrect treatments for individuals. If race becomes the organizing variable for genetic variation, the potentially democratizing new tools of personalized medicine could become instead the instruments of a new means of stereotyping groups of people, legitimized by the perceived authority of genetics. This in turn could exacerbate existing inequalities.

In addition to concerns about the conflation of race and biology, there are concerns about the accessibility of precision medicine tools. Given a history of uneven access to medical care for some groups, there are also concerns about how democratic, accessible and inclusive precision medicine will be. Will new genomic technologies remain out of reach of those without adequate insurance coverage, or the financial means to pay out of pocket? Currently, DNA genotyping and sequencing technologies are only available at a few elite health systems across the country. New health technologies, if not deployed equitably, might exacerbate pre-existing socioeconomic divides and lead to a health system that is further fractured in terms of quality and access, with tiered levels of care based on a patient's means.

Given that the pricetag of a full genome sequence is a couple of thousand dollars, insurers have been hesitant to cover the costs, especially when the chances of finding medically actionable in the genome are low. In a few instances, and for those who can afford it, genome information has been extremely informative, and has helped identify previously unknown, rare, highly heritable, familial diseases. But insurers have been unwilling to reimburse genome sequencing as a preventative measure (for example, when an individual has a family history of a complex disease in which genetic factors are not believed to be significant enough to warrant the cost of testing).

The question of accessibility and deployment extends not just to patient care, or to access to the new genomics methods for genotyping and sequencing DNA, but to new information technologies that are part of the precision medicine toolkit. Precision medicine claims it will

democratize health, putting health information in patients hands. As the floods of "big data" increase, how can we ensure that genome information is accessible, empowering and democratizing, rather than disenfranchising? For example many new technologies touted under the banner of precision medicine require access to digital technologies: laptops, tablets, and smartphones, and broadband internet. According to a 2015 Pew Research Center Report, poor or elderly Americans are less likely to have access to home internet connections, bank accounts or health coverage than affluent and well-educated Americans. How widely available can precision medicine technologies be if they rely on other technologies that are already unequally distributed across the "digital divide"? How can these divides be bridged? These are important questions to consider as precision medicine advocates make decisions about where to focus efforts and how to guide future health policy.

Decentering Genetics: Alternative Views of Disease

Because of its strong genetic focus, precision medicine ideologically places the burden of disease causality on DNA. But since genetics cannot account for most diseases, a decentering of this focus could help widen the scope of study and understanding, and help to find workable solutions for addressing health disparities.

A focus on trying to find genetic differences between race and ethnic groups, in order to explain differences in disease incidence, will likely divert needed attention and scarce resources away from other more significant factors leading to differential disease susceptibilities among sociocultural groupings, such as quality of life and standard of living, socioeconomic status, neighborhood, and access to health and education.

Increasingly, geneticists studying these diseases are taking the role of environmental factors more seriously, re-focusing their attention on interactions between genes and the environment. They argue that perhaps genomics has been unable to account for most diseases so far because our genomes do not operate in a vacuum. Perhaps, they argue, environmental

factors "trigger" genetic factors in ways that the current exclusive focus on genetics alone cannot capture. In this way, they are making efforts to widen the perspectival terrain, which might help to veer biomedical research away from the limited vantage of genetic explanations for disease. This also makes possible an accounting for the very real bodily effects of socioeconomic disparities in biology and health. Differences in environmental factors may contribute to differences in disease incidence among different race and ethnic groups, owing not to genetic predispositions but to correlations with socioeconomic disparities that exist between different race and ethnic groups in the U.S.

This approach alone is not likely to be sufficient. While many genomic studies have purported to find biological and genetic differences along race, the evidence strongly suggests that socioeconomic variables have much stronger causal relationships to disease than either biology or race. This would suggest that the best interventions from a public health standpoint would be to invest resources in strengthening access to healthy environments across all communities. This might include improving housing, employment and educational opportunities, improving nutrition and access to healthier living conditions, addressing institutional factors that contribute to psychosocial stresses, and intervening in other contexts of inequality, especially for those most at-risk of falling through the cracks of the system.

Although the evidence strongly suggests that socioeconomic or "environmental" factors play a far greater role in disease and health than any genetic factors proposed to date, this data remains consistently under-appreciated in the decision-making processes that determine which types of research get funding. The allure of genetic explanations, for everything from human disease to human behavior and human psychology, remains compelling, firmly embedded in our research infrastructures, funding regimes, and even in popular opinion. In addition, there is little institutional momentum to redirect resources to improving socioeconomic conditions for those most disenfranchised, and is too often de-prioritized or not even considered in policy decisions.

Conclusion: Reimagining Health Futures

As contemporary genomics reveals, it is not easy to separate the "scientific" from the "cultural," the "social," or the "political" within the practices of making and doing difference. How do various actors and institutions locate, delineate and ascribe meaning to difference? On what basis? In order to understand why and how different differences comes to matter in different contexts, we must understand how these knowledges are constructed. This is the point at which STS approaches can help to crack open the fissures and reveal the continuities and exchanges that traffic between "science" and "society," blurring the binary distinction.

Precision medicine has brought American healthcare to a crossroads. Genomic testing has in a few cases proven effective at diagnosing critical health information in time for prevention or treatment, and new insights may well help inform better health outcomes for those with access. But the subtle and overt ways in which the race-as-genetic theme continues to animate discussions in the field present an obstacle to the stated goals of precision medicine. If these persist, then the tools of the enterprise may be misused to sustain old beliefs about racial differences, yoking them to supposedly "natural" differences that lead to poor health and disease susceptibilities. This racial essentialism in turn may fuel a genetic essentialism, the view that our genetics establishes an innate, definitive roadmap of our future health. However, recent studies of hundreds of common complex diseases suggest that genetics has only a small part to do with our susceptibilities to these diseases. If precision medicine is to bear out its name and become truly "personalized," then a focus on racial differences at the level of the genome constitutes a step off-path, with many ramifications, including the possibility of exacerbating racial and ethnic stereotyping and discrimination during routine medical care and the attendant social and ultimately health consequences.

Importantly, difference always carries with it an epistemic politics. How difference is coded, framed, and understood is always contested, and the relationships of power among different

actors modulate the role that differences come to play in political and cultural spaces, of which the laboratories of science are a part. This begs the question, is it possible to think/talk about difference without reifying it? What sorts of epistemic (or ethical) limits could or should be invoked or operationalized?

A social justice agenda for healthcare might investigate how precision medicine can be used to build responsible, equitable and just frameworks for talking about genetic and health differences, without reifying, stereotyping and marginalizing groups along common sociocultural axes. It might explore how we can better use the new tools to address the gross health disparities in our medical systems. Given that genomic approaches to health have established a strong foothold in clinical research, how can we ensure that the benefits of new tools for personalizing medicine are equally shared?

A multi-pronged approach is needed. Too often, social science scholars and "publics" are viewed as "outsiders" to the biological sciences and medicine. Such public engagement often only takes place after the path to a particular biomedical end has already been chosen and embarked upon, not at the critical upstream junctures at which decisions are made regarding the nature of the problem to be addressed and possible means to address it. More robust and inclusive policies for determining health funding priorities and for distributing the benefits of new genomics technologies are needed, but will require significant momentum from various stakeholders: patients, patient advocacy groups, regulators, hospital administrators, as well as clinicians and researchers. More inclusive methodological frameworks for generating biomedical knowledge would see a greater diversity of voices at the decision-making table. Patients themselves may be able to contribute in significant ways. They are closest to their own health and disease, and in many instances they know best how their bodies respond to various stimuli. Reimagined forms of engagement for involving various publics in the discussions and debates around the uses and applications of new technologies can help to widen the perspective on what the proximate causes of disease may be and how to treat them. These approaches can also help to make patients participants in their own health, as precision

medicine promises. Practices for interactive knowledge-making and truly "patient-driven" medicine would allow collective assessments of new genomics technologies and medical needs and priorities with greater reflexivity. Indeed, some of these efforts are ongoing (see for example Montoya, 2015). Such interventions might bring us to new conceptions of what it means to "do science" and to be an expert, and even redefine what constitutes authorized scientific knowledge.

In addition, as discussed above, practitioners within the precision medicine space could explore ways to take seriously the various *environmental* onslaughts that individuals are subjected to, and how they interact with both the biological and the genetic to produce health or disease. Decentering dominant discourses around disease and difference can bring to light more nuanced frames for addressing health disparities and health rights, especially for those most marginalized from the benefits of health advances. For example, "big data" might be usefully leveraged in ways that help to develop an attention to human bodies that captures their full richness, instead of employing reductionist frames refracted through the lens of genomics. By decentering the focus on genetics, and instead studying disease causation through an interactional lens, new perspectives on how the biological and the social together produce ill health could allow societies to truly create transformative medicine.

The genomics of disease is still in its infancy, and the medical relevance of genomics findings remains uncertain. Efforts to achieve personalized medicine in clinical settings would do better to focus on how genomes and environments together produce disease, rather than trying to find genetic correlates for race and ethnicity. Genomics researchers have a responsibility to be aware of the ways in which they draw boundaries around groups based on genetics, and to communicate the caveats associated with their findings to the media, various publics, other health researchers, medical practitioners, and clinicians. They need to guard against their findings being misinterpreted as supporting genetic determinism with respect to disease, and biological determinism with respect to race. In the push for precision medicine, such approaches will be vital in preventing new waves of genetic determinism or new practices of

"genetic stereotyping" around disease and race. Doctors and medical decision makers must also be educated on the limitations and caveats associated with findings from genomics studies. In addition, research funding agencies need to be sensitized to the problem of erroneously conflating genetics and race, and use these insights to guide funding priorities and decisions, and establish guidelines for how findings are reported in genomics studies.

If we really are living in the "century of biology," then we need to take seriously our responsibility to be mindful of the legacies of inequality that still persist, and to ensure that new tools and technologies do not simply recapitulate these inequalities. Precision medicine is still a few years away from becoming a clinical reality. Currently, findings from genomics research are still not ready to be extended to individual patient care reliably and accurately, which remains for the most part an elusive enterprise. The knowledge emerging from genomics is still some way from being able to reliably inform decisions about the most effective therapy, treatment or course of action to improve a patient's health and well-being. That is, precision medicine is not yet part of routine clinical practice. This presents an opportunity to realize new forms of civic participation in health care, engaging communities in contributing to understanding their own genetic health risks, in the context of other socioeconomic factors. If precision medicine is to find a footing in the clinic in fair and equitable ways, care should be taken to ensure that genetic differences are not essentialized, naturalized, or biologized as race, and further, that such conflations do not then become the lever by which precision itself comes to be defined in medical care.

References and Further Reading:

Duster, Troy. 1990. *Backdoor to Eugenics*. Psychology Press

Fujimura, Joan H., Deborah A. Bolnick, Ramya Rajagopalan, Jay S. Kaufman, Richard C. Lewontin, Troy Duster, Pilar Ossorio, and Jonathan Marks. 2014. "Clines Without Classes: How to Make Sense of Human Variation." *Sociological Theory* 32 (3): 208–27.

Fujimura, J.H. and R. Rajagopalan. (2011) "Different differences: The use of ancestry versus race in biomedical human genetic research." *Social Studies of Science*, 41(1): 5-30.

Geronimus, Arline T., and J. Phillip Thompson. 2004. "To Denigrate, Ignore, or Disrupt: Racial Inequality in Health and the Impact of a Policy-Induced Breakdown of African American Communities." *Du Bois Review* 1 (02): 247–79.

Krieger, Nancy, and Mary Bassett. 1993. "The Health of Black Folk." *The "Racial" Economy of Science: Toward a Democratic Future*, 161.

Krieger, Nancy. 2011. *Epidemiology and the People's Health: Theory and Context*. Oxford University Press New York.

Montoya, Michael. (accessed July 2015). The Community Knowledge Project. http://www.communityknowledgeproject.org/

Rajagopalan, R. and J.H. Fujimura. (2012) "Making History via DNA, Making DNA from History: Deconstructing the Race-Disease Connection in Admixture Mapping." In *Genetics and the Unsettled Past: The Collision between DNA, Race, and History.* (ed. K. Wailoo, C. Lee, A. Nelson), Rutgers University Press, New Brunswick, NJ.

Biographical Note:

Ramya Rajagopalan studies the social and political dimensions of recent developments in the life sciences, including human genomics and personalized medicine, and their implications for understandings of race, identity and difference. She has a background in received her PhD in Genetics from MIT, and trained in Science and Technology Studies as a postdoctoral fellow at UW-Madison. Her co-authored article "Different Differences" won the 2013 David Edge Best Article Prize from the 4S.