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The Ethical and Social Implications of Rhetorical Reform in Genomic Medicine

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Since the dawn of the Human Genome Project in the late 1980s, the human genetics and genomics research community has been promising to usher in a "new paradigm for health care"—one that uses molecular profiling to identify human genetic variants implicated in multifactorial health risks. Patricia Baird eloquently captured their rationale in her 1990 manifesto when she wrote, "We need to see our own genetic individuality as a potential origin of disease. We are all different—we are all genetically unique—which means our risk for disease is different one from another. Progress depends on realizing this and applying the knowledge to prevention."[1]

After the completion of the HGP in 2003, a wide range of stakeholders became committed to this "paradigm shift," creating a confluence of investment, advocacy, and enthusiasm that bears all the marks of a "scientific/intellectual social movement" within biomedicine.[2] As in many revolutionary movements, however, the stakeholders' shared frustrations with the status quo do not always translate into exactly the same vision of the future. As that vision evolves, so do the movement's ethical and social implications.

Since 2011, we have conducted semistructured interviews and ethnographic case studies to analyze how proponents of this movement understand and pursue its goals in order to anticipate the ethical and social challenges they may encounter as the revolution proceeds. Proponents usually offer four ways in which their approach to medical diagnosis and health care improves upon current practices, arguing that it is more "personalized," "predictive," "preventive," and "participatory" than the medical status quo.[3] Initially, it was the first of these virtues—personalization—that seemed to best sum up the movement's appeal,[4] and efforts to translate the tools of genomic analysis into the clinical setting have been widely promoted across scientific, clinical, governmental, and commercial settings as advancing "personalized" genomic medicine.[5] Although the term "personalized medicine" carries different connotations for different stakeholders,[6] it has become one of the most visible biomedical banners of the millennial decades, joining "translational clinical science" and "evidence-based medicine" to headline biomedical initiatives of all shapes and sizes.

By 2012, however, even as the clinicians, editors, and lobbyists we interviewed continued to herald the ways in which personalized genomic medicine would revolutionize health care, powerful opinion leaders were abandoning "personalized medicine" as a usefully descriptive name for their cause in favor of a new label: "precision medicine."[7] Since then, a wave of rebranding and rhetorical reform has swept the field, with this new label "trending" in the names of institutional programs, job titles, scientific headlines, and journal articles.[8] There have been occasional dissenters,[9] but with the U.S. President's State of the Union address in

January 2015, a decisive seal of approval was given to the new label when President Obama unveiled plans for a national "precision medicine initiative" to promote the development and use of genomic tools in health care.[10] The subsequent use of the label by National Institutes of Health (NIH) leadership[11] in launching a portfolio of federal grant-making in support of the PMI has made "personalized medicine" suddenly sound quaintly old-fashioned.

In this paper, we report results from interviews with 143 proponents of personalized genomic medicine (PGM)—including scientists, translational researchers, commercial and nonprofit developers, research funders, clinician-researchers, clinicians in private practice, health professional educators, medical journal editors, and health insurers—to help explain this rhetorical shift and the "rebranding" of the movement.[12] Although most of the stakeholders we studied seemed unaware of the shifting nomenclature when they were interviewed in 2011 and 2012, their backstage reflections on the "personalized" label unveil key tensions that drove the shift toward "precision" and signal ethical and social implications of the changing rhetoric.

To look ahead, our interviews highlight two ideological shifts in the emerging practice of genomic medicine that the movement's rebranding both marks and masks. The first is a turn away from "patient empowerment" and toward expert-mediated decision-making in the clinical setting, reviving debates over medical paternalism that long seemed settled, at least in clinical genetics. The second is to broaden the movement's focus from "individualizing" treatments for particular patients to using genomic profiling on behalf of the interests of extended families, minority groups, and national populations. Both shifts are realistic correctives to the early rhetoric of personalized medicine. However, they also have important implications for the moral priorities that propel this field and, by extension, for the ethical orientations of the professionals and institutions that embrace it. Because these changes in the application of genomics represent a significant departure from the individualistic ethos that initially facilitated public and political support for the genomic medicine movement, they will be important to follow and assess as the genomic revolution unfolds.

The Problems with "Personalized"

As a label for a genomic approach to diagnosis and prevention, "personalized medicine" has had detractors. Physicians have defended traditional medical practice as already thoroughly "personalized," in the sense that good clinicians have always valued knowing each patient's unique health history, social context, and subjective complaints during both diagnosis and treatment.[13] Genome scientists have warned that reducing "personalization" to molecular profiling may, ironically, carry the risk of making health care more *im*personal.[14] Public health advocates chafe against the label because it seems to dismiss or downplay environmental, social, and systemic approaches to prevention.[15] Social scientists, historians, and bioethicists have complained about the hollowness of the label's implied promise to put patients more in control of their health care, as well as its congeniality with neoliberal efforts to relieve society of collective responsibilities for health care equity.[16]

Resistance, however, is a rite of passage for new social movements, and these external criticisms seem to have been largely ignored as personalized genomic medicine has gained

momentum. More significant have been emergent internal tensions within the movement over what "personalized" health care might really mean. On one hand, our interviewees repeatedly cited the ideal of "individually tailored" medicine as the movement's ultimate promise. In the words of one genetic counselor, "It really means using information from genetic results and from DNA testing to personalize a health plan for a patient, whether that's in the area of prevention or treatment options. So really just customizing health care and prevention based on what individuals' DNA makeup is" (I37).[17]

At the same time, our interviewees noted that, for the foreseeable future, genomic medicine will be less about developing unique prescriptions for individual patients and more about categorizing patients into different classes of genetic risk and therapeutic efficacy based on what is known about the subsets of the population with their genotypes. As one senior editor of a genomics journal said,

> Some people equate "personalized" as being equal then to "personal," and I don't think that we can ever, ever really become truly personal and truly individualized because there are so many variables in our environment …. So the way I look at personalized medicine is whereby we can stratify patient groups respective of ancestry, ethnicity, into individuals who are more likely to respond using novel technologies, which may be genomics, proteomics, etc., in combination with environmental factors. So I see a way of being able to subphenotype disease, subphenotype individuals in the way they're going to respond to drugs, and that's what I see as personalized medicine. So I don't see it as individual. (I83)

While a few respondents were optimistic that truly individualized care might one day occur, the overwhelming sense among respondents was that genomic medicine would remain at the group level, stratified by empiric genomic disease risk associations and, to lesser degrees, generalizations from racial and ethnic ancestry. As the president and CEO of a sequencing company told us,

> The hope of course here is that we'll be able to subcategorize people into ever smaller groups that can be more targeted in terms of how a diagnosis is done, what a diagnosis means, what treatment would apply, what side effects that subgroup might experience from a particular therapy, and as we learn more and more about the genome, that those subgroups get smaller and smaller. The day will likely never come where each individual has something specifically done for them that is done to no one else. But I think these groups or subgroups will get increasingly smaller as we learn and have higher resolution to the genomic information. (I55)

The problem for "personalization" is that the statistical logic of genomic information can only really illuminate the health risks of groups, thereby leaving genomic medicine to, at best, classify individuals as members of those groups.[18] This ambiguity was publicly acknowledged when the movement reached political water-sheds involving reports by high-level professional and science-policy bodies. [19] In assessing the "priorities for personalized medicine," the 2008 report by the U.S. President's Council of Advisors on Science and Technology explained that "personalized medicine … does not literally mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify

individuals into subpopulations that differ in their susceptibility to a particular disease or response to a specific treatment."[20]

As the widespread recognition of this point within the translational genomics community was echoed in more public assessments, momentum grew to reform the movement's labeling to better acknowledge its classificatory approach. Scientist Maynard Olsen commented, for example, "I think 'personalized medicine' was perhaps a useful rubric with which to launch this activity, but it sends a misleading message—actually both to ourselves and the broader community."[21]

One early proposal, coming out of the commercial sector already familiar with the notion of "stratified markets," has been to relabel and promote the new paradigm as "stratified medicine." "Some call this approach to proactively testing and selecting populations for specific treatments 'personalized' medicine, but," advocates of the proposal argued, "we believe a more useful description is 'stratified medicine.' In stratified medicine, a patient can be found to be similar to a cohort that has historically exhibited a differential therapeutic response using a biomarker that has been correlated to that differential response."[22]

However straightforward this proposal appears, the reframing of "personalized medicine" as "stratified medicine" has not gained much traction in the United States. For our interviewees, this label evoked notions of resource stratification by both race and income, raising complex and unappealing problems of genetic discrimination, access to health care, and social injustice. Such problems were noted, for example, by an editor of a genomics journal and the director of an academic medical center's personalized health care program:

> This is a really complex social problem but if we deal with genomic and genetic information related to different populations, to different ethnic groups, we should think before we all have this data: What should we do? How can we handle these social problems? How can we make sure that … there will not be a discrimination of those different populations? (I85)

> You might be able to stratify people in some populations or groups that may predict worse outcomes, and I think a lot of those individuals are worried that their insurance rates will go up or they wouldn't be hired or they wouldn't be promoted in a certain job because of the concern of their employers or of their insurance companies that they may be more expensive long term. (I7)

To avoid triggering the social and political concerns raised by the connotations of "population stratification," promoters of genomic medicine in the United States needed a label with more neutral, if not positive, connotations in the public mind. They found their alternative in the writing of economist Clay Christensen, who together with Jerome Grossman, coined the label "precision medicine" in their 2009 book *Innovator's Prescription*.[23] That label's use in the title of an influential 2011 National Academy of Science and Institute of Medicine report, *Toward Precision Medicine*,[24] effectively launched it as a new banner for the movement.

Enter “Precision Medicine”

Precision medicine” was chosen by the IOM committee to convey its sense that genomics and other emerging biodata sciences could improve medicine’s clinically defined nosology. Redefining clinical disease entities in terms of specific molecular causal factors could allow clinicians to diagnose more precisely, with presumable benefits for therapy and prevention when different molecular diagnoses indicate different responses. Charles Sawyer, the cochair of the IOM committee that produced that report, explained that “[w]ith the term ‘precision medicine’ we are trying to convey a more precise classification of disease into subgroups that in the past were lumped together because there wasn’t a clear way to discriminate between them.”[25] On the surface, refining disease classifications does not seem like the same thing as stratifying patients into different subpopulations. But they amount to the same thing, to the extent that the science that associates particular molecular markers with different risks, outcomes, and clinical indications is population based to begin with.[26]

Moreover, shifting the gaze to classifying *diagnoses* rather than *patients* allows “precision medicine” to exploit the popular appeal of “unique tailoring” without giving up the statistical evidence on which measurements rest. As a result, the term “precision medicine” has become ubiquitous as a synonym for “personalized medicine” and is popularly defined as the same approach. As one of its exponents writes, “Call it what you will—personalized medicine, genomic medicine, precision medicine. It’s an approach that emphasizes the ways in which your disease risks are unique and different, just like your other more obvious characteristics. Those disease risks are based on the predispositions written into your genome at birth, combined with your lifestyle and environment.”[27]

“Precision medicine” has other rhetorical virtues as an aspirational label for the goal of translational genomic research as well. First, it helps the movement retreat from its early hyperbolic promises about “individualized therapies” while keeping its central focus on the molecular profiling of individual patients. In doing so, it allows the movement to avoid antagonizing those traditional clinicians who already claim to personalize their care and helps disambiguate it from other holistic “wellness” movements that also exploit the “personalized medicine” label.[28] As Duke’s Geoffrey Ginsburg explains, although “personalized medicine” was always intended to refer to the notion of using genomic information to guide therapy for disease, “[a]t the same time the patient and patient-centeredness have appropriately become central to health care. And with that movement has [*sic*.] come debate and perhaps confusion as to the real meaning of the phrase ‘personalized medicine.’ Concomitantly with the rise of diverse molecular/sequencing and digital/mHealth and eHealth technologies and the recognition of molecular heterogeneity of individuals the term ‘precision medicine’ is being used more and more to reflect the evolution of the field.”[29]

Second, as this quotation indicates, “precision medicine” provides a mantle under which other forms of “data-intensive” interventions in biomedicine—such as electronic medical records research, longitudinal epidemiological studies, crowd-sourced health-data tracking, and environmental health research—can be assimilated in order to broaden the movement’s appeal beyond genomics. This allows “precision medicine” to avoid charges of genomic

exceptionalism and offers an attractive alliance to the spectrum of other "disruptive" data-mining approaches that aspire to revolutionize medicine, from electronic medical records research to demographic and geographic "Big Data." As Dan Roden, assistant vice chancellor for personalized medicine at Vanderbilt University, has been quoted as saying, "The twinkle in many people's eyes has been that you'll be able to marry this idea of dense phenotypic data to genomic data, transcriptomic data, economic data, sociocultural data—all those things that may determine how someone responds to treatments and to disease."[30]

Finally, "precision medicine" even allows the movement to harness the public appeal of contemporary military metaphors such as "precision bombing" and "surgical strikes" and their echoes in the "tumor targeting" language of oncology. Francis Collins memorably observed in a 2016 television interview that, unlike traditional, "one-size-fits-all medicine," "[t]his is much more precise. It's a smart bomb."[31]

At the same time, this rebranding has coincided with two other shifts in the development and clinical integration of translational genomic tools: a renewed insistence on professional gatekeeping in the clinical application of genomic medicine and an increased interest in the public health uses of population-level conceptualizations of genomic variation. To some extent, these shifts are only coincidentally related to the rebranding: they have been spurred as much by the clinical introduction of new genomic sequencing tools and population-level genomic variation research as by any resistance to the rhetoric of personalization. However, these shifts are observable in the literature over the same period as the rebranding, and they each have important ethical implications for the future directions of the genomic medicine movement.

Abandoning Personal Empowerment? Echoes in Practice Trends

One of the virtues often associated with PGM was its putative ability to empower individual patients to actively participate in their care by personalizing risk assessments and health management plans. As attractive as this virtue has been as a selling point for the marketing of genomic medicine by government officials, health care institutions, and commercial labs to potential consumers, the empowerment PGM can foster is relatively illusory. In fact, as we have documented elsewhere,[32] the net effect of most medical uses of genomic information is to provide more authoritative medical justification for clinicians to go one way rather than another in response to symptoms or to reduce disease risk. As one genomics journal editor explained, the ultimate aim of the revolution is simply to make genomic testing another instrument in the physician's toolbox:

> What we just hope to do is provide a tool the physician can use like any other diagnostic test, where … rather than image the brain with an MRI, we're imaging the genome with many, many pixels, so we're getting the whole sequence and providing some variants that may be useful to the clinician to help them better see how to diagnose their patient and potentially treat them based on their individual genome. (I87)

Similarly, even though the Food and Drug Administration's proposal for "Paving the Way" for PGM defines personalized medicine as "tailoring medical treatment to the individual

characteristics, needs, and *preferences* of each patient," it focuses on what personalized medicine can bring to professional clinical judgment: "Our current lack of ability to predict an individual patient's treatment success means that clinicians have no choice but to follow a less than optimal approach …. The goal of personalized medicine is to streamline clinical decision-making by distinguishing in advance those patients most likely to benefit from those who will incur cost and suffer side effects without gaining benefit."[33]

"Precision medicine" is an apt way to characterize this shift because, at a metaphorical level, the operation of "precision" equipment, large-scale "data-mining" activities, and the targeting of "smart bombs" are implicitly the domains of professionals, not amateurs. In fact, with the shift to "precision medicine," *patient-driven* decision-making seems almost completely jettisoned as the revolution's signature virtue. For example, the University of California, San Francisco, in publicizing its effort to "Drive the Precision Medicine Revolution," makes a selling point of the prospect that patients may have *no* role at all in negotiating their diagnosis and treatment: "Imagine a time when you could have a simple blood test on the way to your doctor's office and arrive at the appointment to find your doctor fully prepared with a diagnosis. Imagine that the ideal medicine would already be identified based on your genetic makeup and perfectly formulated to avoid side effects for which you are susceptible…. That is the goal of precision medicine."[34]

Delivering the Data Tsunami

Data from our interviews with PGM promoters suggests a number of reasons for this retreat toward professional gatekeeping. Among the most important is an attempt to control the impact of the "data tsunami" that comes with genomic medicine's emerging abilities to analyze multiple genomic loci simultaneously, either through multiplex testing, genome-wide scans, or clinical exome or whole genome sequencing.[35] Our respondents were keenly aware of the dangers of overloading patients with information that they have no way to interpret and of the need for better ways to validate, distill, and deliver the information that genomic tools can provide. One PGM provider put it this way:

> So just handing someone a sheet of paper that says, "You're homozygous for the ApoE4 variant," and saying, "I'm so sorry," and, "Goodbye," I don't think is meaningful, and to me, taking genetic information and translating that to something with true clinical utility is what I've really cared about. I think that's got to be one of the things that's critical in the future. If genetics are going to play a role in clinical medicine, somehow all of those data and piles and you know gigabytes of information have got to be translated in the physician's office between a physician, genetic counselor, [or] someone, and that patient so that they really understand what this is all about. (I40)

To a large extent, the professional concern over the interpretation of overwhelming amounts of genomic data has been animated by the rise of the direct-to-consumer genetic testing industry and its appropriation of the empowerment rhetoric for marketing purposes. In reaction to concerns over the reliability and utility of the genomic information being offered directly to patients by direct-to-consumer commercial labs[36] academic and medical

promoters of PGM have encouraged health professionals to (re)claim more traditional gatekeeping roles in the clinical provision of genomic information.[37]

Among clinicians pioneering genomic medicine, the sentiment has been that the "fire hose" of information available through genomics needs to be wielded by parties with the best interests of patients in mind, even if that means assuming a more traditional medical gatekeeping role than the open-access ethos some "consumer genomics" enthusiasts would endorse.[38] As one PGM provider told us, this is a better way to achieve patient empowerment than "information on demand" because, when it comes to the implications of complex genomic test results, patients "want to be coached…. They want a partner in their health, and you have to be partners" (I138).

Like other visions of the therapeutic relationship grounded in "shared decision-making" between clinicians and patients, however, this partnership is not meant to be as equal as a shared business ownership. It is a fiduciary relationship in which the professional is privileged to "coach" patient partners on the basis of expert knowledge and to make decisions that advance the partners' best interests.[39] For some professionals, acting as the fiduciary facilitator of patients' genomic empowerment also means accepting the authority to identify and enforce patients' genomic responsibilities. The founding director of an academic medical center's personalized medicine research program explained, for example,

> I can only hold you so responsible for what you do as a participant in the process, but if I know by various markers how you should respond to this therapy, I can hold you more responsible and I can say, "Well … we know that if we give you this particular medication, this is what the outcome should be, but … if you don't stop smoking, you know you now move out of the 80 percent successful to the 30 percent successful with this medication." … I think as a country and as a society, we ought to hold you somewhat more responsible for that…. [I]f you're not willing to do your part of that, well then maybe you ought to pay a little more for health care, or you … ought to, you know, have something that helps you assign some responsibility to the process. (I06)

This provider takes it to be his professional mandate to hold the patient accountable for her lifestyle choices and to discipline those choices. This is not an entirely unusual clinical posture, but it does take a step back from the "patient-centered" individualism expressed by early PGM promoters and their lay supporters.

First, Do No Harm

In addition to clinical interpretation and what we might call "responsibility coaching," a third emerging feature of the clinician's role in genomic medicine is the obligation to withhold genomic information that has no medical utility, whether the patients think they want it or not. The nature and limits of this editorial obligation in the context of genomic testing have been thrown into relief by the advent of clinical sequencing technologies and by the need to make decisions about which DNA sequencing results to analyze and return to patients. Against the efforts of direct-to-consumer genomic testing companies to cultivate public interest in direct access to their raw genomic data, some clinicians are appealing to

their traditional duty to “first, do no harm” to censor the disclosure of uninterpretable, uninformative, and clinically irrelevant information that could serve only to confuse the patient, just as they would ordinarily omit irrelevant remedies from their nongenomic treatment recommendations.[40]

Of course, withholding information that *could* be used to prevent avoidable harm is also problematic on these grounds, even when that is what the patient wants. The next step down the road toward more paternalistic genomic medicine is to argue that the principle of nonmaleficence also supports overriding patients’ disinterest in learning their genetic risks in the first place. In the words of a senior editor of a genomics journal,

> We also have examples from our whole exome and whole genome sequencing studies where we’re getting all this unexpected data, and how does one convey that? So think of it almost as “collateral data.” So we’re looking for a cause for a particular syndrome and we know what to do if we find that, but we also get a bunch of data from pharmacogenetics and all these other things as part of it. So what do we do? Do we tell people that they are going to get muscle pain from statins should they ever take it, or what do we do? And so that’s been a real, you know, a real ethical problem. How do we take that forward? (I82)

The most prominent flashpoint for this issue was the debate over the 2012 recommendations of the American College of Medical Genetics, which prescribes a list of mutations that should be opportunistically sought and disclosed to patients whenever clinical sequencing is undertaken, regardless of the patient’s wishes. The ACMG felt this was necessary due to uneven laboratory reporting standards, leaving a chance that laboratories might either report clinically unactionable findings or fail to report unanticipated “secondary” findings that would reveal preventable genetic conditions.[41] The ACMG therefore solicited and compiled expert opinion to generate a list of fifty-six genes associated with twenty-four health conditions that met this criterion and recommended that laboratories routinely screen for pathological variants in these genes and report positive findings to ordering clinicians whenever clinical sequencing is conducted. This would put clinicians in a position to warn their patients of the risks they face, even if they were not risks that the patient was seeking to clarify through sequencing. This, they concluded, would ensure that clinicians fulfill their professional duty to prevent harm for patients and their families, even at the expense of patient autonomy.[42]

The upshot of these four moves—from *clinical interpretation* to *responsibility coaching* to *informational censoring* to *involuntary genetic testing and disclosure*—is a significant departure from the traditional ethos of clinical genetics. Clinical genetics has historically been one of the medical specialties stressing the primacy of the patient’s role in decisions to seek diagnoses and learn health risks. In part, this tradition has historical roots in the reaction of post-World War II medical geneticists to the excesses of their eugenic predecessors. However, it also reflects an important strategy for dealing with the predictive and moral uncertainties of the decisions that geneticists and genetic counselors help their clients make.[43] The practical result of this orientation has been a strongly client-centered ethos that, historically, anticipated by twenty years the rise of “patient autonomy” in the ethics of other medical specialties.

Some see the resurgence of paternalism in medical genetics as a retrograde retreat from the field's commitment to respecting patient autonomy, while others see it simply as the healthy normalization of genomics as a medical specialty.[44] Either way, it signals that if genomic information is to be used as a clinical tool, the world of precision medicine will demand renewed attention to our models of the doctor-patient relationship and the patient role in negotiating what patients can expect to learn about themselves in the clinical encounter.

Finally, it is ironic that *clinical* genomics is moving away from the rhetoric of patient empowerment just as the basic genomic *research* enterprise, in the form of the precision medicine initiative, has doubled down on empowerment rhetoric in promoting its plan to recruit one million Americans into its longitudinal cohort. Although the PMI emphasizes that the main health benefit of its research will be to give "*medical professionals* the resources they need to target the specific treatments of the illnesses we encounter,"[45] the working group charged with operationalizing the PMI emphasizes in its recommendation to the NIH that "[r]espect for individual autonomy and rejection of paternalism is a paramount concern of the PMI-Cohort Program and is a motivation underlying the participatory model."[46] Indeed, the "Principles of Privacy and Participation" that are being used as a guiding framework for the initiative's development at the NIH embrace the individualistic language of the direct-to-consumer genomics companies verbatim—they promise PMI volunteers direct access to their genomic information and research results, personal control over the research conducted with their samples, and a voice in the governance of the overall initiative.[47] If this ethos can be sustained as the PMI is implemented over the next years, it will create an interesting difference between what people might expect to encounter in the genomic research context on the one hand and in the actual delivery of clinical genomic services on the other.

From Individualized to Population-Level Thinking

While the rhetoric of "personalized" genomic medicine encouraged early advocates to look forward to the day when group risk classifications could be abandoned in favor of completely individualized risk assessments,[48] today's thought leaders admit that this is unlikely. The chief medical officer of a sequencing company, for instance, told us,

> We can't get personal in medicine. I can measure what's happening on an individual, but deciding what happens with them, they have to be in a subgroup, because I have to show statistically that this subgroup behaves different than other subgroups. By definition you're no longer personal. So what this really needs to be called is "genetic subgroup medicine." (I206)

But the acknowledgment that genomic medicine is as much about defining different human genetic subgroups as it is about individualized care has also opened new opportunities for the precision medicine movement. Collective and group health risks are part of the traditional domain of public health, and advocates of "public health genomics" have pointed out that this makes the extension of "precision" approaches to population-health problems both apt and important. As Muin Khoury and colleagues recently put it,

> Could the same technologies that propel precision medicine usher in a parallel era of "precision public health" beyond treatment of sick individuals? If precision medicine is about providing the right treatment to the right patient at the right time, precision public health can be simply viewed as providing the right intervention to the right population at the right time. More accurate methods for measuring disease, pathogens, exposures, behaviors, and susceptibility could allow better assessment of population health and development of policies and targeted programs for preventing disease.[49]

In fact, Khoury suggests that precision medicine's logic makes the pursuit of these "precision prevention" goals not just parallel to, but prerequisite for, the success of precision medicine because achieving the clinical goals of individualized care will require the development of the population-level genomic information public health seeks in order to target its interventions.[50]

Population-Based "Precision Prevention"?

To operationalize the idea of precision prevention, its advocates must make an important conceptual move that is not strictly necessary within a narrower vision of "precision medicine." That is, they have to equate genetic health risk groups across which individual patients might be stratified with the kinds of human groups of concern to public health officials and policy-makers: visible groups with names, locations, and legitimate claims on public resources, for two reasons: First, genetic marker groups relevant to stratifying patient risk are relatively invisible subpopulations before their members' risks are realized, making it difficult to know how to precisely target any preventive interventions in advance. The only way to preemptively identify those groups in advance of their health problems would be through universal population genomic screening, currently being debated in the context of newborn sequencing,[51] and preventive genomic sequencing for adults in the clinical setting.[52] But universal "one size fits all" screening for genetic risks would presumably lose all the gains in efficacy, efficiency, and harm reduction promised by the "precision prevention" approach. To pursue "precision prevention," public health needs to be able to associate the genetic health risks it targets with more visible, phenotypic group characteristics, just as it does in attempting to reduce behavioral health risks through educational interventions tailored to people in visibly different social circumstances.

Second, and more importantly, genetic risk marker subgroups are not the kinds of human groups that public health is designed to serve. For reasons of justice, effectiveness, practicality, and political necessity, public health must define the collective targets of its preventive services along socially discernable lines, as constituencies rather than statistical constructs. Among the most relevant of those discernable characteristics for *genetic* risk prevention are the social categories that we would expect to overlap with patterns of genetic inheritance in the population, like family, ancestry, community identity, ethnicity, and race. As a result, the kinds of public health problems that offer the best opportunities for population health assessments aimed at "precision prevention" are those that segregate along those social lines.[53]

One of the most prominent population-health problems segregated along social lines in the United States are the disparities in health and health outcomes between racially and ethnically defined groups. As a result, a dominating theme of the new “precision public health” discourse has been the use of population genomic information to address this problem. One instructive example of the results is a Request for Application released in the summer of 2015 by the NIH’s National Institute on Minority Health and Health Disparities (NIMHHD), calling for research taking “precision medicine” approaches to the elimination of health disparities between populations.[54]

This solicitation begins by asserting that the major causes of health disparities are structural and systemic factors related to the disadvantaged social status of particular groups and by defining “precision medicine” broadly enough to encompass empirical measures of these social determinants. But the “first priority” of the RFA is to find better ways to correlate such measures with biological risk factors through population genomic studies of the ethnic minorities and disadvantaged social groups they define as “disparity populations.”[55] Thus, they call for research that identifies genomic differences that might account for the disparities these groups experience, that translates those differences into “minority-specific therapies,” and that reduces “community-level and or cultural or societal” barriers in these groups.

It is difficult for agencies like NIMHHD to avoid the political realities of group identity when dealing with public health problems like outcome disparities. But that is because these problems are fundamentally social and political. It seems like a conceptual non sequitur to look for genomic differences between constituencies in order to explain the effects of their unjust social situations. At the very least, it drags the efforts at “precision prevention” into the heart of the debate over how best to correlate human genetic variation with human social identities and the wisdom of reifying categories like “race” in genomic terms.[56]

On the whole, genomic thought leaders have a strong record of cautious, sophisticated, and nuanced participation in this debate.[57] As some of those on the frontlines of population genomic research write,

> The use of social group labels such as African American, Hispanics, and Asians are likely to be insufficient to get us to where we need to be as we strive towards individualized medicine…. If we use genomic information correctly, we will simultaneously describe our similarities and differences without reaffirming old prejudices. More importantly, the careful unbiased study and interpretation of the human story coded in our DNA will enable us to appreciate the fact that individuals cannot be treated as a representative for all those who physically resemble them or who share some of their ancestry.[58]

Rather than “precision prevention” at the group level, the hope has been that, once the entire spectrum of human genomic variation is mined for its health implications, the racial and ethnic categories that framed its collection and analysis could fall away, and the DNA markers could be used directly for individualized risk assessment in a race-neutral fashion.[59] As precision medicine’s population-based foundations get extrapolated into public health initiatives, however, the mounting weight of clinical and epidemiological research framed

against societal minority group membership and “self-identified race” seems to be creating a politically irresistible temptation to follow suit in genomics. As a result, the concepts of race and ethnicity and their links to health disparities remain badly tangled with the logic of genomic risk stratification in precision medicine’s promotional discourse and public health initiatives.[60]

From Collective Need to Individual Obligation

Beyond conceptual confusion, attempting to address health disparities through genomics also brings its own downstream ethical and social challenges. First, as others have pointed out, it carries the risks of being misinterpreted to unfairly “blame the victims” of structural and systemic injustices, unnecessarily imposing group harms like stigmatization, and diverting public health resources from efforts to address the underlying social determinants of the health disparities that different constituencies face.[61] Equally problematic, however, are the uses of such associations to impute group-based obligations to participate in targeted screening activities, like the early efforts under the National Genetic Disease Act of the 1970s to promote preconception carrier screening for sickle cell disease among African Americans[62] or like the famously “successful” public health programs to reduce the incidence of hemoglobinopathies in Sardina and Cyprus.[63]

While the moral merits of these episodes are still debated, their lessons are what helped bring about U.S. public health policies that promote adult and reproductive genetic screening programs as strictly voluntary opportunities for individual risk reduction rather than collective expectations for the common good.[64] When genomic population health assessments are framed as efforts to address the visible health disparities experienced by particular families, communities, or kin groups, however, the pendulum begins to swing the other way. For those with moral commitments of solidarity, loyalty, or service to “their people,” participating in such precision prevention assessments is likely to begin to feel like a matter of social responsibility rather than an optional opportunity for personal risk reduction.[65]

Taken to its logical conclusion, finally, the same line of thinking could be used even to support expectations that everyone has a duty to one’s own social subgroup to become engaged in a genomic research initiative like the PMI. Some, like the philosopher Rosamond Rhodes, have already made this argument in vivid terms:

> The expectation is that researchers will learn a great deal more about the human genome and the human microbiome and that this new knowledge will allow medicine to tailor treatments to individuals. The studies, however, will require the development of biobank and sample bank repositories with the participation of a tremendous number of subjects. To reap the rewards, broad public participation will be required. Furthermore, to the extent that any group abstains from participation, their members will be less able to share in the rewards precisely because their genetic and microbiomic samples are absent from the pool …. I want to point out that existing injustices can only be exacerbated by members of these groups refusing to participate in research. If your group does not participate in studies that

> assess health disparities, no one will know that health disparities of the sort that negatively affect you exist.[66]

For Rhodes, unjust health disparities between groups, ostensibly the publicly obvious phenotypes that genomics might help obliquely explain, are no longer even detectable without genomic research, presumably because they have been reduced to the intergroup genomic differences themselves. Since only genomic research participation can illuminate these differences, the responsibility for rectifying the negative health of a given disadvantaged population falls heavily on that very population.

As scientifically confused and socially dangerous as this essentialist thinking is, it is an effective marketing strategy for translational genomic research. But it does depart from the tradition of insisting that individual decisions to participate in biomedical research should be free and voluntary. Some would view this departure as a laudable corrective to an excessively atomistic way of thinking about human autonomy and an overdue recognition of the importance of communitarian values like solidarity in the genomics research setting.[67] Others would worry that, because of the inevitable mismatch between our social group identities and our genetic risk classifications, the group identities reinforced by genomic difference claims would only exacerbate unjust social divides that already plague us.[68]

Moreover, as experience with other constituency-framed population genomics initiatives has shown, fomenting social groups' investment in "their" genomic differences also spurs groups to assert a variety of other interests in research governance, from group harm protections to community engagement, data gatekeeping, and benefit sharing. We have already seen families, communities, and nations attempt to protect and advance these interests under a variety of ownership concepts, from family "legacy"[69] and group "patrimony"[70] to national "genomic sovereignty."[71] As legitimate as such claims may be coming from socially acknowledged political entities, when they are advanced on behalf of particular currents in the global human gene flow, they face all the conceptual and ethical problems displayed by commercial and scientific claims to genomic information ownership.[72]

Beyond implicit appeals to national patriotism,[73] the new U.S. PMI has not yet taken the step of appealing to notions of subgroup solidarity in order to acquire the range of genomic variation it needs. The promotional rhetoric of the PMI and its "Principles of Privacy and Trust" show strong commitments to the primacy of individual autonomy in both research enrollment and genomic information management, even in the face of clinical practice trends in the other direction. But if its effort to "empower every citizen to volunteer" does not yield a suitably representative research cohort, the pressure to use people's group memberships to encourage participation could build. Just as the PMI's professed aspirations to empower its individual research volunteers have the potential to conflict with the clinical trends toward professional gatekeeping, they also risk coming into tension with the social pressures created by group-based "precision prevention" discourse.

A New Set of Concerns

The weakness of "personalized genomic medicine," as a promissory label for what genomics might bring to health care, is that it promises more than genomics can actually deliver—both in terms of increased patient empowerment and in terms of the individualization of care. Although "precision medicine" correctly takes the focus of translational genomics off of the individual patient in both ways, the clinical and public health trends associated with this new label bring other ethical and social concerns. First, to the extent that it connotes "precision equipment" and privileges medical expertise and training, it encourages professional claims to authority and threatens the medical ethos of shared decision-making, by entitling clinicians to make and act on moral judgments about the propriety of their patients' choices, allowing clinicians to edit the information about patients that they share, and encouraging clinicians to seek out and share information that patients might have chosen not to know about themselves.

Second, as the movement's focus expands to include "precision prevention" framed in terms of the visible constituencies of public health, it encourages the genomic reification of people's social affiliations and ethnic identities, which risks privileging group over individual interests at the same time that it reinforces social divisiveness in the name of health equity. To the extent that "precision medicine" echoes "precision bombing" and targets specific human social groups as the key units of analysis for and beneficiaries of genomic medicine, it encourages population-based essentialism that elevates group allegiances in ethically suspect ways.

Both of these trends are visible in the currents of the genomic medicine movement and must be negotiated transparently, no matter which banners it marches under. At a minimum, their confluence calls into question the translational potential of the individualistic ideals of the U.S. PMI and opens the movement up to charges of false advertising, as programs and providers continue to exploit the rhetoric of personal empowerment to promote what are evolving into increasingly conventional medical services and public health interventions for collective benefit. If taken further, they could even threaten the level of personal autonomy that our historical experience with other state-sponsored genetics movements has helped people to gain, in both health care and research settings. Meanwhile, as the field of bioethics independently rebalances *its* paradigms toward relational autonomy, solidarity, and more nuanced understandings of shared decision-making, it can help genome science be more precise in shaping its agenda, without undercutting that history's hard-won moral progress.

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