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. Initially, it was the first of these virtues—personalization—that seemed to best sum up the movement’s appeal, 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. Although the term “personalized medicine” carries different connotations for different stakeholders, 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.” 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. There have been occasional dissenters, 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. The subsequent use of the label by National Institutes of Health (NIH) leadership 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. 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. Genome scientists have warned that reducing “personalization” to molecular profiling may, ironically, carry the risk of making health care more impersonal. Public health advocates chafe against the label because it seems to dismiss or downplay environmental, social, and systemic approaches to prevention. 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.

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” (L37).
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, 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 watersheds 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 ‘person- alized’ 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...”

**“Personalized medicine” has had detractors, but emergent internal tensions over what “personalized” health care might really mean have been more significant.**

...
a discrimination of those different populations? (185)

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. (17)

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-centered

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. (187)

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 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
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, academic and medical promoters of PGM have encouraged health professionals to (re)claim more traditional gatekeeping roles in the clinical provision of genomic information.

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. 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".

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. 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. . . . If 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.

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.

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,
what do we do? Do we tell people that they are going to get muscle pain from statins should they ever take it, or do 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. 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 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.12

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.33 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 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 sub-
group behaves different than other
subgroups. By definition you’re no
longer personal. So what this really
needs to be called is “genetic sub-
group medicine.” (I206)

But the acknowledgment that
genomic medicine is as much about
defining different human genetic
subgroups as it is about individual-
ized care has also opened new oppor-
tunities for the precision medicine
movement. Collective and group
health risks are part of the traditional
domain of public health, and advo-
cates 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 treat-
ment 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, be-
haviors, and susceptibility could
allow better assessment of popu-
lation health and development of
policies and targeted programs for
preventing disease.59

In fact, Khoury suggests that pre-
cision medicine’s logic makes the
pursuit of these “precision preven-
tion” goals not just parallel to, but
prerequisite for, the success of pre-
cision medicine because achieving the
clinical goals of individualized care
will require the development of the
population-level genomic informa-
tion public health seeks in order to
target its interventions.59

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 “precis-
ion 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 re-
sources, 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 iden-
tify those groups in advance of their
health problems would be through
universal population genomic screen-
ing, 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 char-
acteristics, just as it does in attempt-
ing 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 collec-
tive targets of its preventive services
along socially discernible lines, as
colleagues rather than statistical
constructs. Among the most relevant
of those discernable characteristics
for genetic risk prevention are the so-
cial 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 popu-
lation health assessments aimed at
“precision prevention” are those that
segregate along those social lines.53

One of the most prominent
population-health problems segre-
gated 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 “pre-
cision public health” discourse has
been the use of population genomic
information to address this prob-
lem. One instructive example of the
results is a Request for Application
released in the summer of 2015 by
the NIH’s National Institute on Mi-
nority Health and Health Disparities
(NIMHHD), calling for research tak-
ing “precision medicine” approaches
to the elimination of health dispari-
ties between populations.54

This solicitation begins by assert-
ing 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 em-
pirical measures of these social de-
determinants. But the “first priority”
of the RFA is to find better ways to
correlate such measures with biolog-
ical risk factors through population
genomic studies of the ethnic minori-
ties and disadvantaged social groups
they define as “disparity popula-
tions.”55 Thus, they call for research
that identifies genomic differences
that might account for the disparities
these groups experience, that trans-
lates 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 Americans62 or like the famously “successful” public health programs to reduce the incidence of hemoglobinopathies in Sardinia 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

As bioethics independently rebalances its paradigms toward relational autonomy, solidarity, and more nuanced understandings of shared decision-making, it can help genome science shape its agenda, without undercuts the state-sponsored genetics movements.
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.

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.

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.

Moreover, as experience with other constituency-framed population genomics initiatives has shown, fostering 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” and group “patrimony” to national “genomic sovereignty.” 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.

Beyond implicit appeals to national patriotism, 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 promotional 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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Notes

17. Quotations of interviewees are identified in parentheses by the number we assigned to them; “I37” refers, for example, to interviewee 37.

20. President’s Council of Advisors on Science and Technology, “Priorities for Personalized Medicine,” 1.


48. For example, in 2010 Francis Collins wrote, “The goal for personalized medicine must be to move as swiftly as possible toward the identification of individual risk factors, be they environmental or genetic, that play a direct role in disease risk. Racial profiling in medicine, even if well intentioned right now, should recede into the past as a murky, inaccurate, and potentially prejudicial surrogate for the real thing” (Collins, The Language of Life, 163).


52. J. P. Evans et al., “We Screen Newborns, Don’t We?: Realizing the Promise of Public Health Genomics,” Genetics in Medicine 15, no. 5 (2010): 332-34.


55. “These include ‘Blacks/African Americans, Hispanics/Latinos, American Indians/Alaska Natives, Asian Americans, Native Hawaiians and other Pacific Islanders,’ as well as ‘rural’ and ‘socioeconomically disadvantaged populations’ (National Institute on Minority Health and Health Disparities, “NIMHD Transdisciplinary Collaborative Centers for Health Disparities Research Focused on Precision Medicine [U54]).”


73. The White House publicity for the PMI warns that “[t]ranslating these successes to a larger scale will require a national effort: to push this effort forward we will need all hands on deck, including patients, hospitals, industry” (L. Holst, “The Precision Medicine Initiative: Data Driven Treatments as Unique as Your Own Body,” The White House (blog), January 30, 2015, https://www.whitehouse.gov/blog/2015/01/30/precision-medicine-initiative-data-driven-treatments-unique-your-own-body).