

# Precision medicine, health disparities, and ethics: the case for disability inclusion

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Precision medicine research (PMR) is gaining momentum across the health-care landscape in the United States. The 2015 announcement of the Precision Medicine Initiative accelerated this process, making PMR a priority on a national level and committing significant budgetary allocations to such programs. Aside from advancing scientific knowledge, a major promise of precision medicine (PM) is that it will curtail the deep health disparities that have plagued the country. Yet, while efforts are ongoing to ensure inclusivity of racial and ethnic minorities,<sup>1</sup> the views of persons with disabilities about PMR have not been well studied. Although several studies have explored the perspectives of patients and the lay public in the United States on genomic research, deriving generalizable disability responses from these studies is impossible because disability is not a required category for reporting. Still, persons with disabilities—a commonly used umbrella term, which, following the US census and Convention on the Rights of Persons with Disabilities, encompasses persons with long-term physical, mental, intellectual/developmental, and sensory (e.g., blindness) impairments—merit attention. Although some disabilities are associated with health conditions that require ongoing treatment, disability and good health are not mutually exclusive. Patients and healthy persons with disabilities may thus have different views and needs for participating in PMR. Involving these populations in PMR is necessary for fulfilling PM's scientific goals and promise of health equity, and for upholding the equal opportunity of persons with disabilities—22% of US adults, as the Centers for Disease Control and Prevention reported in 2015<sup>2</sup>—to enjoy the benefits of PMR.

## JUSTIFICATIONS FOR DISABILITY INCLUSION

Precision medicine is a newly emerging approach to health care. Its long-term goal is to replace the “one-size-fits-all” approach, which offers health-care strategies based on the average person, with more customized preventative and therapeutic options that take individual genetic, environmental, and lifestyle variability into account. Although still in its infancy, advances in PM have already led to new scientific and therapeutic discoveries, e.g., translating pharmacogenomics findings into treatment decisions for patients with lung cancer.<sup>3</sup> In the future, tools of PM may enable tailored

treatment for other health conditions, such as cardiovascular and psychiatric disorders, foster better understanding of population health, and allow the development of strategies to reduce health disparities.<sup>4</sup> Precision medicine research with sufficiently powered inclusion of ethnic/racial subgroups is key to achieving these goals and for identifying genetic variants that contribute to disease risk or affect drug response within and across populations.<sup>1</sup>

Persons with disabilities comprise a necessary group for achieving these endeavors. First, some impairments have genetic underpinnings for which effective treatments are currently unavailable. Notwithstanding disagreements within and outside the disability community about how to delineate disability (e.g., members of the Deaf community do not view deafness as a disability), and whether and which impairments should be treated,<sup>5</sup> progress on understanding, improving, and developing tailored interventions for any of these conditions will be impossible unless persons with disabilities are included in PMR. Second, the prevalence of comorbidities among subgroups of persons with disabilities is invaluable in advancing PM's research priorities, such as oncology. For instance, studies found that, compared with nondisabled peers, persons with schizophrenia had an increased risk of lung and breast cancers but a lower risk of other cancers (e.g., malignant melanoma).<sup>6</sup> Including patients and healthy persons with disabilities in PMR could thus illuminate the complex underlying biological and other mechanisms that link impairments to increased, or reduced, risk for comorbidities. Third, because disability is present across age, sex, race, and ethnicity groups, the inclusion of healthy persons with disabilities in PMR could amplify the genetic and other heterogeneity that is needed for meaningful analysis of variability within and between populations. Finally, studies demonstrate that, as a group, persons with disabilities, especially women,<sup>6</sup> constitute the largest—hitherto unrecognized—health disparity population in the United States: they are less likely to have access to preventative and community-based, quality health-care services, more likely to be poor, and they fare worse than their nondisabled peers across a broad range of health indicators and outcomes.<sup>7</sup> Including this population in PMR can improve understanding of gene-environment interactions and epigenetic processes that

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impact health outcomes and inform strategies for curtailing health disparities in this population.

### BARRIERS FOR DISABILITY INCLUSION

Without concrete efforts, the reasons for the limited participation of persons with disabilities in health research and clinical trials (compared with their nondisabled peers)<sup>7</sup> will probably extend to PMR. These include: difficulties in obtaining informed consent—long observed in genomic research—that are amplified for people with intellectual disabilities or those who are cycling in and out of psychiatric crises; the use of comorbidities as exclusion criteria in clinical trials,<sup>8</sup> which may have a particularly strong impact on patients with disabilities; researchers' lack of knowledge of how to design studies in disability-accessible formats, which precludes participation<sup>8</sup> and denies especially healthy persons with disabilities from even learning about research opportunities; and over-protectionism of institutional review boards, e.g., requiring demonstrable benefit to participants with disabilities, which increases the risk of noninclusive decisions on research participation also in minimal-risk PMR (and other) studies.<sup>9</sup>

Social determinants contributing to health inequalities between persons with disabilities and the general population may further impede research participation. For instance, inaccessible health-care facilities and equipment (e.g., examination rooms) may discourage healthy persons with disabilities from interacting with such institutions.<sup>10</sup> Patients with disabilities, who are already more likely than nondisabled peers to forgo needed medical appointments owing to cost and inadequate transportation,<sup>7</sup> are even more likely to refrain from attending research meetings that do not have immediate relevance. In addition, overall, higher rates of poverty, unemployment, and low levels of education among the general disabled population, especially women,<sup>6</sup> along with less access to the Internet<sup>7</sup>—a primary tool for communication and ongoing engagement in PMR—make involvement in research especially challenging.

The history of disability and genetic research may further negatively affect interest in and the trust required for participating in PMR. The study of how genotypes are expressed has been historically preoccupied with those deemed as “deviant phenotypes” (i.e., people with disabilities), and aside from notorious incidents of abuse (e.g., the Nazi regime's genetic research on persons with intellectual and psychiatric conditions), it has been mobilized by a “therapeutic imperative,” which tends to regard impairments as leading to lives of misery, hence, in need of prevention or cure—often regardless of the personal narratives of those living with disabilities.<sup>5</sup> The routinization of genetic practices (e.g., prenatal genetic testing) and the development of gene editing technologies (e.g., CRISPR/Cas9) have similarly raised concerns. As disability voices have highlighted, focusing on genetics to explain impairments reinforces a medical approach that views disabilities as personal traits in need of fixing while distracting from the societal and environmental

barriers that disable individuals from fully participating in society. Moreover, the aforementioned genetic practices increase the risk of a devaluation of persons with disabilities and efforts to eradicate natural human differences.<sup>5</sup>

Whether such views indeed discourage participation in PMR is currently unknown and is worth investigating. However, PM may well be positioned to address these concerns. Because PMR encompasses more than genetics and includes environmental and lifestyle factors, it can be more attuned to the social model of disability that holds the latter factors as important as genetic or biological ones in causing disability. PMR also moves away from the contentious focus on curing and eradicating disabilities (however delineated) in offering therapeutic benefits for comorbidities. Furthermore, the partnership required for the success of PMR could alleviate concerns about objectification in genomic research and resonate with the disability-rights slogan of “nothing about us without us.” Thus, although criticism of certain uses of genetics (e.g., prenatal screening) may remain, persons with disabilities may find the broader PM enterprise to be worthy of consideration.

### MOVING FORWARD

How to ensure that persons with disabilities are included in PMR is as yet unclear, and studies in this population across disability types and health needs are necessary, as a first step, to better understand the barriers—and possible solutions—to participation. Some issues are relatively straightforward. For instance, the provision of accessible information—key for enrolling all participants into PM cohorts—must encompass alternative formats for subgroups of persons with disabilities (e.g., Braille: blind/low-vision community). Methods for obtaining consent from, enrolling, and retaining ongoing engagement of this population in PMR require further consideration. The latter can include representation on institutional review boards and governing bodies of PM projects and sharing the results of studies in disability-accessible formats<sup>9</sup>—significantly, bearing in mind that the Internet may not reach persons with disabilities to an equal extent. However, collaborations between researchers and persons with disabilities are essential for feedback and additional insight on these (and other) proposed measures. Such practices are compatible with the spirit of PM while not compromising the protection of research subjects.

Without addressing these gaps, PMR runs the risk of developing only limited scientific knowledge and exacerbating, not curtailing, health disparities in the United States. To fathom the issues at stake, and to implement the spirit and goals of PM, it is essential to conduct studies with persons with disabilities to explore their views and to engage them in the planning process. People with disabilities have firsthand knowledge of their needs, and they know best what measures are required to overcome the barriers to their participation.

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The author declares no conflict of interest.

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