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Will Precision Medicine Improve Population Health?

Muin J. Khoury, MD, PhD and
Office of Public Health Genomics, Centers for Disease Control and Prevention, Atlanta, Georgia.

Sandro Galea, MD, DrPH
Boston University School of Public Health, Boston, Massachusetts.

Announcement of the precision medicine initiative has led to a variety of responses, ranging from enthusiastic expectations[1] to explicit skepticism,[2] about potential health benefits, limitations, and return on investment. This Viewpoint discusses whether precision medicine is unlikely or likely to improve population health, aiming to forge a consensus that bridges disparate perspectives on the issue. The potential of precision medicine to improve the health of individuals or small groups of individuals is not addressed here because it involves a different question with different metrics.

Precision Medicine Is Unlikely to Improve Population Health

There are 3 fundamental reasons why precision medicine might not improve the health of populations. First, disease pathogenesis, especially for common non-communicable diseases, is extraordinarily complex. Abundant evidence has demonstrated this for the association between the multiplicity of specific genes and conditions, including obesity, hypertension, or certain cancers. Additionally, it is known that genetic associations have, in most instances, small effect sizes in contrast with more robust contributions of behavioral and social factors.

Second, a central promise of precision medicine is the identification of predictors of disease that can help guide interventions. This may prove to be the case for some diseases, especially cancer, but is unlikely to be the case for most other complex diseases. The challenge arises from the mathematical foundations of genetic epidemiology. Although large population studies can identify associations between genotypes and phenotypes, resulting associations have limited capacity to predict phenotype in individuals, which is the ultimate goal of precision medicine. It would take substantially stronger associations–several orders of magnitude greater than have been identified so far–to provide sufficient evidence to improve disease prediction in individuals.

Third, an assumed potential benefit of precision medicine (predicated on accurate and meaningful risk prediction) is that disease in the population can be avoided or forestalled by

Corresponding Author: Muin J. Khoury, MD, PhD, Office of Public Health Genomics, Centers for Disease Control and Prevention, 1600 Clifton Rd, Atlanta, GA 30329 (muk1@cdc.gov).

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large numbers of individuals who, when provided with accurate risk prediction, will change their behavior to mitigate their personal risk. Although this may seem intuitively plausible, current data suggest that individuals do not change their behavior much even when they become aware of being in a high-risk group.[3]

Overemphasis on precision medicine by the scientific community and health systems could pose a challenge to the health of populations for other reasons. First, the United States faces extraordinary challenges to the health of its population. Over the past 30 years, the United States has fallen behind other high-income peer nations in health attainment on many metrics, including life expectancy and infant mortality, and there are persistent gaps in health outcomes by income and race/ethnicity.[4] The solution to these challenges is probably not an increased focus on the individual, but rather involves focusing on the social, economic, and structural drivers of population health that are ubiquitous and inevitably linked to health achievement as a country. The centrality of the precision medicine effort to the US national health research agenda may distract from efforts to remedy the foundational causes of ill health such as poverty, obesity, and education. Without addressing these causes, there will be little, if any, success in reversing the trends of poor achievement in US population health.

Second, precision medicine could (and to some extent has) led to a shift from which projects are funded by health research agencies. Funding for grants with a population health or public health goal has declined over the past 10 years at the National Institutes of Health, whereas funding for *-omic* research has increased substantially. This shift in funding may lead to an emerging generation of health scientists who see the world through an individualist lens and may not engage in factors that can improve the health of populations.

Third, the promise of precision medicine may lead to other promises such as the recently announced cancer “moonshot,” which may echo previous efforts that have not lived up to expectations. The hype, which could become unrealized health benefits, could lead to disillusionment in the goals of health science, with potential lasting consequences affecting public confidence and investment in medical research.

Precision Medicine Can Improve Population Health

By contrast, there are 3 fundamental reasons that advances in precision medicine might improve population health. First, population health could improve by applying complementary individual and public health approaches to health care and disease prevention. A focus on the wider environmental and social determinants of health is of great importance in addressing health inequities. However, pitting the health of individuals against the health of populations risks widening an unnecessary divide between medicine and public health. Population health planning requires directing efficient use of resources toward those most at risk. Stratification of populations into risk groups for multiple chronic diseases could provide more efficient and effective prevention and treatment strategies and potentially reduce cost of care. For example, a recent large population study suggested that use of age and polygenic risk score (even though individual genetic variants have small effect sizes) may be more effective in stratifying women into risk categories for breast cancer screening than use of age alone.[5]

Second, substantial evidence indicates that a genetically targeted approach to health has demonstrated a population health benefit. For instance, newborn screening is the largest established precision medicine public health program in the United States. Other opportunities for near-term benefit on population health involve implementing evidence-based tier 1 genomic interventions (those with evidence-based recommendations for use)[6] such as for common genetic conditions associated with preventable premature death from cancer and heart disease. These include hereditary breast and ovarian cancer syndrome, Lynch syndrome, and familial hypercholesterolemia. In aggregate, an estimated 2 million people in the United States have one of these conditions, and most are not aware of their risk.[6]

Third and most important, precision medicine is not limited to genes, drugs, and disease. The same technologies and big data that are propelling precision medicine forward are leading to a new era of precision public health that goes beyond personalized treatment of individuals affected by disease.[7] Precision in the context of public health has been described as improving the ability to prevent disease, promote health, and reduce health disparities in populations by (1) applying emerging methods and technologies for measuring disease, pathogens, exposures, behaviors, and susceptibility in populations; and (2) developing policies and targeted implementation programs to improve health. One potentially important application of precision public health is the use of genomics in the investigation and control of infectious diseases. Pathogen whole-genome sequencing is rapidly changing both clinical and public health microbiology. In addition, genomics promises to become central to other economic sectors such as the environment, agriculture, animal health, biotechnology, and alternative energy.

Conclusions

There are clear tensions at the intersection of precision medicine and public health. There are, however, ways forward in which precision medicine could enhance collaborations between medicine and public health to address population health problems and disparities. Much of the current focus of precision medicine involves developing new drugs for personalized treatment of cancer and other diseases. Moving forward, health professionals need greater emphasis on joining biological with social and environmental determinants of health to develop precision approaches to interventions in individuals and populations. For example, biological knowledge of genetic susceptibility to environmental and occupational exposures could lead to population-wide policy protection based on thresholds determined by the most susceptible individuals in the population rather than individual genetic testing with exposure avoidance only in susceptible individuals.[8] A fundamental concern for population health must be with measuring and addressing health inequities. As with all new technologies, genomic technologies have the potential for widening the divide between the haves and the have-nots.

A major challenge for the future is how to use emerging information from multiple levels—from reductionist molecular markers (genomics, omics, etc) to holistic macrolevel risk factors (behavior, environment, policies)—to develop a better understanding of determinants of health. Precision public health relies on evidence that links population data to measurable outcomes in subpopulations, stratified by persons, place, and time, capitalizing on emerging data and new technologies. Even with millions of points of biological data collected from individuals, it may well be that population-level interventions affecting housing, nutrition, poverty, access to resources, and education may have more benefit on health than individualized interventions. It is, in fact, more likely that a combination of approaches–ranging from population-wide interventions to specific interven-tions tailored to higher-risk groups–will be required to efficiently improve population health and narrow health disparities.

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