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The Genomic Era: What MUST Public Health Do?

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In a recent paper proposing "a vision for the future of genomics," representatives of the US National Human Genome Research Institute (NHGRI) suggested that, "with the completion of a high-quality, comprehensive sequence of the human genome…the genomic era is now a reality."[1] The underlying assumption, that in this new era medical innovations born of genomics will lead to significant improvements in human health, is a safe one. Though unlikely to give rise to a panacea for genetically transmitted disease and dysfunction, our ever clearer understanding of the genetic underpinnings of disease will most certainly have a substantial impact on health care. As the NHGRI group went on to concede, however, the widespread application of genomics to health is some years away.

Those years provide a narrow window of opportunity for the creation and execution of public health initiatives that address the challenges inherent in bringing genomics into the clinic. To a limited extent, genomics has already begun to enter the public health sphere, as evidenced by the screening of newborns for various genetic conditions (eg, cystic fibrosis), and the use of genetic testing to identify carriers of heritable mutations that confer significant predisposition to certain forms of cancer (eg, BRCA1-linked breast cancer). With tests now clinically available for more than 600 genetic diseases from albinism to Williams syndrome,[2] much broader applications of genomics to health care are clearly imminent. Indeed, the time has come to move from suggestions of what *should* be done for public health to incorporate genomics into its purview to discussions of what *must* be done.

The many practical and ethical challenges facing public health in this grand and necessary endeavor have been explored to considerable depth elsewhere.[3] This brief article will focus on 2 of these challenges, the resolution of which will be of particular importance in allowing public health to fulfill its overarching mandate to seek and implement means of providing for and ensuring the collective well-being of the public.

The first and perhaps most pressing challenge is to foster significant improvements in "genetic literacy," which "includes knowing about benefits, risks, and limitations of genetic screening and testing, as well as the implications of genetic information."[4] Owing in large part to the somewhat esoteric nature of the science of genetics, a substantial segment of the public could presently be considered genetics illiterates. True, there are few people in developed countries who are completely

unfamiliar with DNA *per se*. However, just as knowing one's ABCs does not allow one to understand a book, knowledge of the ACGTs of DNA does not equate to understanding its potential medical applications and implications. Moreover, "genetic illiteracy" is a hurdle not only for members of the lay public but also for physicians, many of whom received their medical training before the clinical use of genetics made such literacy necessary.

Genetic literacy for *all* is essential to secure maximal health benefits for the public in the application of genomics to health care. Physicians, ultimately responsible for the clinical use of genetic technology, must take the lead in ensuring that it is used to bring about actual improvements in the health of their patients. They must possess a sufficient level of understanding to appropriately advise their patients about the possible risks and benefits of increasingly numerous diagnostic and therapeutic options made possible through genomics. They must also knowledgeably field the inevitable questions from patients curious about genetic testing, or they will risk, among other things, losing patients' trust in their ability to provide competent care.

That said, patients with a basic understanding of genetics will be empowered to make informed decisions with respect to their care, where less-informed patients may forgo testing due to ignorance, misunderstanding, or fear. Hence, the best outcomes of genetic testing and treatment will likely result from patient-physician interactions in which the genetic literacy level is high for both parties.

Public health initiatives must therefore seek to raise genetic literacy if the promise of genomics is to be realized. This necessity has not gone unacknowledged; numerous groups, including the US Department of Health and Human Services, have deemed it a top priority. Prioritization, however, is a long way from accomplishment, with the 2 bridged by action. For example, state medical boards, some of which already mandate continuing medical education (CME) content, could raise the genetic literacy of practicing physicians by imposing minimum requirements for CME in genetics. Unfortunately, however, none presently do.[5] This or other broad-scoped policy-based approaches will be necessary to ensure that all physicians keep abreast of developments in health care genetics.

Effectively reaching the general public will prove more difficult. For example, since approximately 60 percent of Americans access the Internet, with 80 percent of them using it occasionally to search for health information,[6] patient-centered websites (eg, www.nationalhealthcouncil.org) could disseminate information on health care applications of genetics. Such an approach, however, would fail to reach the 40 percent of Americans who are not online. Similarly, coverage of health care genetics by television and print media reaches only those who choose such news items in favor of "The Simpsons" or the sports section. Therefore, innovative efforts must be undertaken to see that *all* members of the public are informed.

This raises what I see as the other great challenge to the successful integration of genomics into public health: ensuring public accessibility to benefits in the context of a health care system compromised by disparity. Unlike the patient-physician relationship, in which the obligations of the physician extend primarily to the individual patient, the raison d'être of public health is to seek and ensure the collective well-being of *all* members of the public. The single most important role for public health in ushering in the genomic era is to ensure that its benefits reach everyone.

With health care costs rising seemingly unabated, it is likely that medical applications of genomics, presently very expensive tools, will exacerbate the already troubling disparities in health care. Truly, the potential for a "genomic divide" exists not only between developed and developing nations,[7] but also between the socioeconomic strata within those nations. Thus, existing and foreseeable disparities must be assessed and must then be explicitly addressed by any policies instituted to govern the public health applications of genomics. Finally, the public must be unhesitatingly given demonstrable assurance that these considerations will be paramount and that genetic technologies will be made available to anyone for whom they hold the promise of improved health.

If the genomic era is to be one in which genomics, used wisely and effectively, achieves significant improvements in human health, public health must hold genetic literacy and avoidance of disparity as primary goals; without genetic literacy, implementation will be practically impossible; with disparity, implementation will be inadequate and unethical.

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