Precision medicine and genomics—an opportunity to improve public health?

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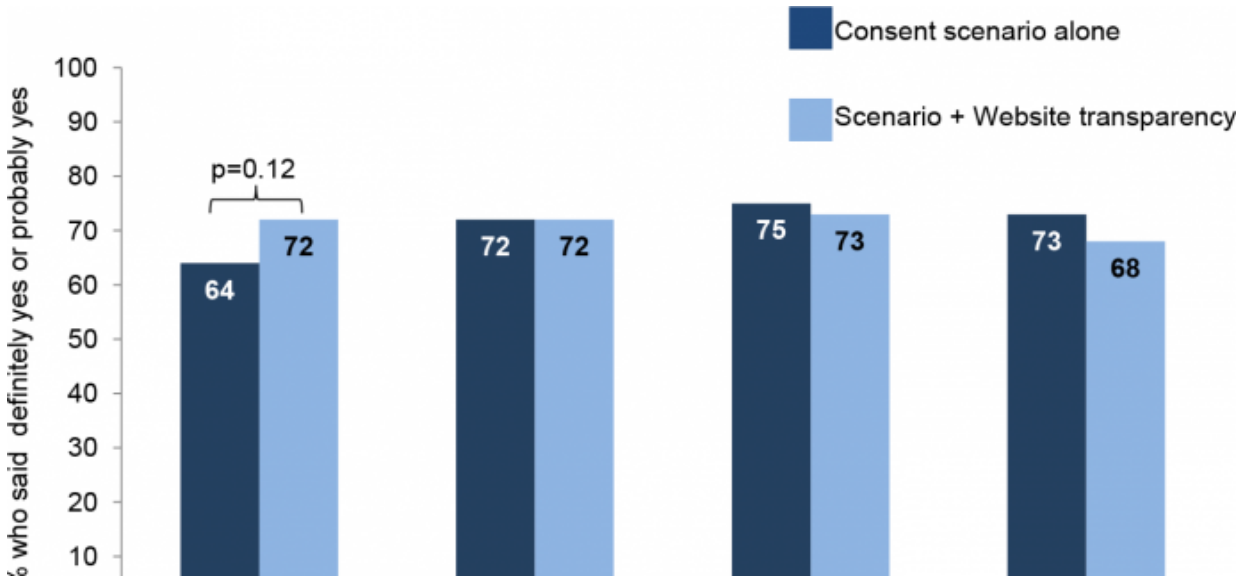

Today, healthcare is portrayed as standing at a crossroads. Precision medicine (also referred to as genomic, personal or individualized medicine) is expected to revolutionize future healthcare. Fundamentally, the promise of precision medicine is that an individual's genetic information will increasingly be used to prioritize access to health care. This model contrasts with the "one-size-fits-all" approach of today, in which disease treatment and prevention strategies are developed for the average person, with less consideration for the differences between

individuals. Advances in genomic and clinical science have created innovative opportunities to further tailor health care to each patient, allowing providers to create optimized care plans at every stage of a disease, shifting the focus from reactive to preventive health care. Basically, precision medicine maintains that medical care and public health will be radically transformed by prevention and treatment programs more closely targeted to the individual patient. These interventions will be developed by sequencing more genomes, creating bigger biobanks, and linking biological information to health data in electronic medical records.

It was with these promises of change that President Obama announced the Precision Medicine Initiative in his 2015 State of the Union address. U.S. government scientists are now seeking a million volunteers willing to share the innermost secrets of their genes and daily lives as part of an ambitious 10-year research project to understand the causes and cures of disease. Those selected to be members of the "precision medicine cohort program" will be asked to provide a detailed medical history and blood samples so researchers can extract DNA. They will also be asked to report information about themselves (lifestyle and environmental factors), so researchers can identify possible risk factors. The project seeks to develop treatments tailored to the characteristics of individual patients. According to survey results published in PLOS ONE, 79% of U.S. adults who responded supported the Precision Medicine Initiative's planned national cohort, and 54% said they would definitely or probably participate if asked. The opportunity to learn health information about oneself from the study appears to be a strong motivation to participate in this study.

False positives and overdiagnosis?

In theory, the progress of precision medicine seems uncontroversial. Who wouldn't want better healthcare for all, with real benefits for the

individual patient with tailored and optimized treatment plan suitable for his or her genetic makeup? Perhaps it is for this reason that the assumptions underpinning personalized medicine have largely escaped questioning. Personalized medicine advocates say that by using specific genetic diagnostic tests, doctors may predict how well a patient respond to treatment for some diseases or conditions and enable them to determine the best dosage and duration of treatment according to the genetic setup of the individual patient.

The problem is that these tests do not yet exist. In addition, this kind of system would mean that a wider group of doctors would now use patients' genetic and other molecular information as part of routine medical care. But many doctors are simply not qualified to make sense of genetic tests, or to communicate the results accurately to their patients. Hence, this precision approach may instead lead to too many false positives and subsequent overdiagnosis and overtreatment. It could also lead to health insurance companies advertising to encourage patients to get annual check-ups to assess their individual risk. These data could be used to justify higher premiums and insurance costs based on unclear "risks."

Would precision medicine benefit a healthy individual? Today, it is highly debated whether annual check-ups for healthy adults are necessary. For instance, a 2012 Cochrane review concluded that there is little chance that these appointments would be beneficial. Even worse, the FDA recently warned women and their doctors that current screening tests for ovarian cancer are unreliable and could lead to false diagnoses. The FDA is especially concerned about delaying effective preventive treatments for women who show no symptoms, but who are still at increased risk for developing ovarian cancer. Despite the risks of false positives, it's unclear that healthy individuals would gain much from precision medicine.

What about public health?

To complicate matters further, there are already clear tensions at the intersection of precision medicine and public health. Instead of a panacea in solving the current and future health challenges, and the impending shortage of healthcare resources, an overemphasis on precision medicine by the scientific community and health systems could pose a challenge to health of populations, by further increasing focus on the individual. This approach downplays the social, economic, and structural drivers of population health, and it comes with the risk of draining resources from important public health projects to fund hypothetical, expensive medico-technical solutions.

Historically, improvements in public health have come either from general improvement in socioeconomic conditions or from programs targeted broadly to entire populations rather than individualized solutions. As an ECR trained in public health, I am accustomed to defend the "causes of the causes" approach and not to be naïve when new medical initiatives are presented. To me, an open discussion is therefore urgently needed to assess the actual potential (and not the promises) of precision medicine. Otherwise we may risk ending up only benefiting the genetic testing companies.

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