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Genomics and Medicine

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How Hard Could It Be?

The Integration of Personal Genomics into Medical Practice

Personal genomics carries incredible potential to revolutionize the way patients, physicians and institutions deal with health, and to exponentially improve the health of patients themselves. If patients use their sequenced genome effectively, they can take preventative action against diseases to which they have genetic predispositions, such as type II diabetes. By using personal genomics, patients take the most effective medications for their conditions because “genetic variations/mutations in genes that encode drug transporters, drug metabolism enzymes, and receptors are key factors in altering drug responses in some individuals” (Schneider 7). Even aside from the promise of personal genomics to improve and streamline health care itself, in the long term the integration of personal genomics into medical practice would greatly reduce costs. Many diseases and conditions are brought on largely by environmental factors, and if personal genomics can tell a patient they are at high risk for such a disease, he and his physician can work to avoid the disease with lifestyle management at a significantly lower expense than treating the disease (or more likely, its symptoms) once it has developed. Also, if personal genomics can tell a patient that they metabolize a certain drug too quickly for it to be effective, then that patient can use a different drug and avoid the pain of paying for medication that does not work—or worse, is harmful.

These concepts have been integrated with others to create P4 (predictive, preventive, personalized and participatory) medicine. Many, such as the P4 Medical Institute, are embracing these ideas and realize that “personalized, predictive, preventive, and participatory medicine—that is, prospective care— has been receiving increasing attention as a solution to the U.S. health care crisis” (Snyderman 1). As personal genomics is indispensable to this innovative and progressive type of care, one would hope that hospitals, physicians, health insurance companies and government organizations could not wait for its inclusion in day-to-day medicine.

Unfortunately, many issues must be resolved before personal genomics can be effectively integrated into medical practice. These include, but are not limited to, physician resistance, government regulation and involvement with hospitals and insurance companies, and ethical issues involving cost and access.

Physician resistance: why and how to change it

Physician resistance against personal genomics as a part of daily medical practice has several origins. One may be anxiety over their patients’ reactions to the results of a genomic test. This is especially understandable when one considers that the many companies that offer genomic sequencing offer it directly to the individual and do not, in all cases, require a physician’s oversight or meetings with a genetic counselor. If patients are not properly educated (or fail to understand the material provided to them by the personal genomics company they used) on the meaning of and how to interpret the results of their genetic test, those results can be not only useless but misleading or harmful to the patient.

“To make matters more confusing, personal genomics could predict risks of very weak genetic associations while leaving out powerful predictive mutations” (Magnus 2), which is to say that a patient could receive a false sense of security from some of these genome sequencing companies, but actually get the disease they are supposedly at low risk for. In addition, many patients, especially the elderly or those less educated, will not understand the benefits of having their genome sequenced and so will resist personal genomics even if it is offered. On the other hand, some over-zealous patients (likely those with a little too much time spent on the internet) may order the genetic test from a company like 23andMe and engage in self-diagnosis, which is likely to be misdiagnosis, and demand treatment. If individuals are to use their sequenced genomes to effectively manage their health, it is clear that physician oversight is needed.

Of course, physician oversight will mean very little if physicians are not adequately educated on how best to use the results of their patients' genomic tests. This certainly explains some of the physician resistance against the integration of personal genomics into medical practice. As Magnus, Cho and Cook-Deegan astutely recognize, many “physicians know little about the studies that the new personal genomics services use to make their predictions; a lot more education of the medical community therefore also needs to take place” (Magnus 2). The American Medical Association seems to be taking a positive stance toward personal genomics, and is clearly aware of their physician's need for new information, as evidenced by their published physician's Q&A on “Direct-to-Consumer Genetic Testing.” But online references or even trained genomics counselors working in hospitals alongside physicians will not be enough to give doctors the knowledge and confidence they need to personalize their medical treatments with a patient's genome in

hand. As Schneider recognizes, “accurate content about genetics, genomics, and proteomics is essential in curricula for undergraduate medical students, training programs for residents, and continuing professional development programs for practicing physicians”(11). Some medical schools, including Stanford University School of Medicine, now include personal genomics in their curricula. But many have yet to add this, which means that not only all the currently practicing physicians, but most of those graduating in the next few years will have no formal education in what will quickly become a milestone in medical care and an incredible advantage to their patients. With this in mind, it is reasonable to predict that within a few years, the American Medical Association will firstly require all medical schools to educate their students in personal genomics and secondly launch a continuing education campaign for practicing physicians to understand and work effectively with personal genomics.

Into the institution: government regulation, hospitals and insurance companies

In order to integrate fully into medical practice, personal genomics must function as a part of the major health institutions: hospitals, health insurance companies and the federal government, mainly the FDA.

Currently, commercial genomic tests such as those administered by companies like 23andMe and Navigenics are subject to very little federal regulation. That regulation is confined to the very basic quality control of laboratories through CLIA and the approval of test kits by the FDA (Magnus). It is challenging, however, to regulate personal genomics because it does not fit cleanly into the FDA’s existing classification of medical devices and

tests, so there is little precedent for regulation in its case. Still, the “Secretary’s Advisory Committee on Genetics, Health & Society, which advises the US Department of Health and Human Services, has identified significant gaps in the oversight and enforcement of genetic testing, especially nutrigenomic and direct-to-consumer tests” (Magnus 2). Personal genomic testing should be regulated by the FDA for accuracy and safety. This should not be in debate—because the genetic information is useless or harmful if it is not accurate—but further government regulation of personal genomics takes consideration. Some states, such as New York and California, have placed more stringent regulation on personal genomics companies, including laws requiring companies to qualify for a license to administer genetic tests (Magnus). The California Department prohibits offering genetic tests to consumers without a physician’s order (Magnus), which may account for some of the inconsistency seen between the policies of different personal genomics companies, and certainly helps explain the tendency of some companies, such as 23andMe, to list only association, not causal, genes, and to avoid reporting the presence of serious and untreatable conditions, such as Huntington’s. Given the ease of access of information across borders via the internet, however, it is clear that at least national—and eventually international—standards of regulation should be established to protect personal genomics patients everywhere.

This anxiety over direct-to-consumer genomic tests is reminiscent, although in reverse, of the approval of pharmaceutical companies’ DTC advertizing. While the costs and benefits of medical tests and treatments being offered and advertized for sale to the American public without the mediation of a trained professional are debatable, the consequences of pharmaceutical DTC advertizing sheds light on another source of

physician resistance against personal genomics. If anyone can buy a 23andMe home genomic test and discover their predispositions toward a range of diseases and disorders, will everyone be clamoring at their doctors for a range of drugs and treatments they don't need? Or worse, will they receive them? While the clear benefits of personal genomics outweigh these unsightly consequences of patient self-diagnosis and action on anxiety, some regulation of personal genomics companies will be necessary to avoid those consequences and effectively integrate personal genomics into medical practice.

On that note, I would suggest that personal genomics companies be required to administer their health-related genetic sequencing packages only through, and in cooperation with, a physician. While this poses several problems, including initial economic loss by personal genomics companies because doing this would at first be more difficult and less profitable, I believe that in the long-term, this would be beneficial for the personal genomics companies, hospitals, physicians, health insurance companies, and—most importantly—patients. If personal genomics companies were to design comprehensive assimilation plans explaining how the integration of personal genomics into medical practice would benefit the involved parties economically and (for the patients) health-wise and then take those plans to health insurance companies and major hospitals, I would not be surprised to see health insurance companies not only completely covering physician-integrated genomic testing, but actually requiring their clients to have their genome sequences on record. Because in the long term, the benefits of personal genomics used effectively with P4 medicine not only grant patients improved health, but saves all involved parties, especially insurance companies, a lot of money.

Ethical obstacles: cost and access

From an ethical and social point of view, the improved health for a cost poses serious problems. Currently, good health correlates with wealth. It is possible that with the introduction of personal genomics into everyday medical practice that this correlation will become even stronger. It is possible that the cost or, for now, the novelty and complexity (and lack of public awareness) of having the genome sequenced may act as a barrier towards some against this new technology and the better health care it provides. Many factors other than just income could cause this, including “lack of access, lack of trust, assumptions and stereotypes, lack of culturally and linguistically appropriate care, environmental factors...[and these factors] are relevant to the future implementation of personalized health care” (Schneider 10). So relevant, in fact, that I believe the close relationship between personal genomics companies and health insurance agencies or hospitals outlined in the previous section would partially, but not completely, solve this problem.

Even if health insurance companies required all their clients to have their genome sequenced and that genome to be an integral part of their doctor’s diagnosis and treatment (especially preventative treatment), we are faced with an overwhelming portion of the American public that does not have health insurance. Assuming these people do not have health insurance because they can not afford it or do not understand its necessity, it is reasonable to assume that neither would they elect to have their genome sequenced because it would be too expensive and/or they do not understand its uses and applications. Depending upon how successful and philanthropically inclined the budding personal genomics companies become, a possible (though albeit not particularly plausible given the

political manhandling it would require) solution would be a government mandate that these companies devote some percentage of their income to sequencing the genomes of those Americans who can not afford it otherwise, using a hospital and physician as an intermediary. Of course, this would present a range of new problems, such as what government body would manage such a feat and what hospitals or physicians would be willing to participate.

On another ethical and practical note, I see a parallel between this budding genetic sequencing industry and the roaring pharmaceutical industry. “Orphan drugs” occur in the pharmaceutical industry when a drug is dropped before it reaches the market because it will not turn out enough profit, either because the disease it treats is very rare, or the people afflicted by the disease it treats can not afford the medication at all. Is it possible, then, that in the future we will be confronted by the problem of “orphan genes”?

Conclusion

Personal genomics as a part of P4 medicine would provide enormous health and economic benefits to patients and institutions if integrated fully into medical practice. While this attempt has met with resistance from some of the physicians who would be using personal genomics, this resistance could be lessened and eventually surmounted with effective education (in medical schools and through continuing medical education) on the use of personal genomics in clinical practice and a combination of regulation and education to keep patients that use personal genomics safe and well-guided. Of course, for personal genomics to function as a part of medical practice, it must also operate smoothly within major medical institutions via regulation by the FDA, integration with hospitals, and

cooperation with health insurance companies. While the power of personal genomics to revolutionize health care for those who can afford it may blind us to those who can not, we must nevertheless keep in mind the barriers that cost, access and education pose against those in most dire need of personalized medical care. But despite the many obstacles, the clear benefits offered by the integration of personal genomics into medical practice give personalized health care a very bright future.

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