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Ethics watch

OUT OF SEQUENCE: HOW CONSUMER GENOMICS COULD DISPLACE CLINICAL GENETICS

The era of personalized medicine has arrived, but not in the way that many predicted[1]. Rather than emerging as clinical tools derived from years of biomedical research that characterized the accuracy and clinical usefulness of genetic markers of increased disease risk, the first publicly available applications of whole-genome technologies are being developed primarily in the private sphere, by companies that market testing services directly to consumers through the Internet[2]. Consumer genomics companies such as 23andMe, deCODE genetics, Navigenics, and Knome now offer a range of personal genotyping and sequencing services to clients who are interested in probing their genomes. With the declining costs of whole-genome analyses, ever larger segments of the population will soon be able to access their personal genomes, long before the usefulness of clinical applications of prospective genomic information has been established in medical settings.

Ethicists and others have complained that bringing genomic services to the market quickly is out of sequence with normal translational practice and should be a cause for concern[3]. The availability of consumer genomics services does raise important ethical questions, including those about the accuracy and predictive value of the information reported, how best to handle the many ambiguous and unanticipated findings that result from whole-genome analyses, how to protect individual privacy and manage potential implications for biological relatives, and how to ensure appropriate regulatory oversight of whole-genome diagnostics[4]. Amid these many ethical concerns, however, there are other potential outcomes of consumer genomics that might be cause for some degree of cautious optimism.

First, expanded access to their personal sequences might prompt some individuals to assume more responsibility for health-promoting behaviours. If commercial genomic testing services are successful, we might expect that over time they will come to displace clinicians as the primary providers of genetic information related to health promotion (although clinicians no doubt will continue to retain primacy with respect to medical diagnosis and treatment). Empowered with that information, clients can take greater responsibility for their health and identify preventive steps that are consistent with their individual conceptions of healthy living. Faced with the initial task of understanding their own genetic data, many individuals will come to a better understanding of both its promises and its limitations for predicting future health and disease, including becoming more aware of the importance of behavioural and environmental contributors, many of which are within their personal control to change.

Second, clients who understand the possible implications of the information encoded in their genomes might assume more responsibility for maintaining the privacy of that information and its potential implications for the health of biological relatives. Although the locus of control of clinically relevant genetic information always will tilt toward clinicians, with the advent of consumer genomics the locus of the information generated lies squarely in the hands of individual clients rather than in the institutional formulation and interpretation of often complex regulations and criteria for disclosing personal health information. Consumer genomics can motivate individuals to have a more active role in the creation, storage and protection of their personal genetic data.

Third, consumer genomics might play a major part in developing the next generation of strategies for patient education and clinical genetic counselling. The speed at which whole-genome technologies are maturing suggests that equally rapid innovations in their application are needed so that the potential benefits of these tools are realized sooner and are disseminated as widely as possible. Although the initial whole-genome online offerings are marketed primarily to affluent clients, the Internet has the longer-term promise of democratizing access to personal genomic information. For example, initial experiences with self-selecting clients could be used to pilot more effective and direct ways of delivering genetic education and counselling services to much larger numbers of people accessing their personal genomic information than are currently seen by genetic counsellors or clinical geneticists.

Whereas less responsible commercial testing services inevitably will overstate the health implications of genomic information, the question is whether a sufficient number of commercial services will establish voluntary standards that are sufficient to outweigh the likely transgressions of others and thus secure a more permanent role for non-medical providers of personal genomic information. Such standards are vital if commercially generated data are to become reliably used by individuals to fashion their own preventive health strategies in ways that are complementary to diagnostic information generated in more traditional medical settings.

For the purposes of health promotion and disease prevention, consumer genomic information could be conceptualized as having more limited relevance for health behaviours (mainly through raising individual awareness of potential susceptibilities) but as not rising to the level of being considered 'medical information'. This distinction would draw an appropriate line between the quality of genetic data and their interpretation in non-medical and medical settings, but also would be the basis for developing somewhat different ethical and regulatory standards for each setting.

Done well, examining one's genome can be an extra medical surveillance practice that has some preventive value and is complementary to traditional forms of health surveillance and patient care. As such, commercial genomic services can advance the aims of personalized medicine by providing a truly individualized approach to defining health-promoting behaviours. Rather than choose to ignore or over-regulate consumer genomics, we should work constructively with commercial providers to develop standards and practices suitable to their role as the front end of what could be a continuum of personalized health awareness and care.

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