DISCUSSION AND DEBATE

The Helix in the Labyrinth: Do We Need Genetic Health Services and Policy Research?

Le labyrinthe de la spirale : la recherche sur les politiques et les services de santé touchant à la génétique est-elle nécessaire?

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Abstract

In Canada and elsewhere, targeted health services and policy research (HSPR) has been suggested as a means to clarify the health system implications of developments in genetics and genomics. But is such research really needed? We argue that substantial investments in basic genetic and genomic research, coupled with persistent uncertainty about the health system implications of advances in these fields, justify the development of specialized HSPR in genetics and the sustained involvement of the wider HSPR community. Genetic health services and policy research will play a crucial role in informing decision-makers at all levels of the health system about whether and how to integrate developments in genetics, genomics and other complex new technologies.

Résumé

Au Canada comme ailleurs, la recherche sur les politiques et les services de santé (RPSS) a été proposée comme moyen de clarifier les répercussions, sur le système de santé, des avancements en génétique et en génomique. Mais une telle recherche est-elle vraiment nécessaire? Nous soutenons que les investissements substantiels de recherche en génétique et en génomique, jumelés à l'incertitude persistante quant aux répercussions, sur le système de santé, de l'avancement de ces disciplines, justifie l'élaboration de RPSS spécialisée touchant à la génétique ainsi qu'un engagement soutenu envers

le milieu de la recherche sur les politiques et les services de santé. La RPSS touchant à la génétique jouera un rôle important pour éclairer les décideurs de tous les niveaux du système de santé dans leurs choix concernant les avancements de la génétique, de la génomique ou d'autres technologies complexes.

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As the Canadian Institutes of Health Research Institute of Genetics and Institute of Health Services and Policy Research embark on another in a series of requests for applications targeted at genetic health services and policy research, it is worth considering what is to be gained by this strategic investment. Is this a case of the squeaky wheel getting the grease, or a measured response to a pressing need? We argue that investment in genetic health services and policy research (HSPR) is needed to clarify the nature and extent of the healthcare and health system implications of developments in genetics and genomics, and to provide the evidence base to support decision-making by clinicians, healthcare administrators and system planners. At the same time, HSPR that takes genetic technologies and associated services as its focus can contribute broader insights about the healthcare and health system implications of other complex interventions. Genetic HSPR offers opportunities for all interested researchers and policy makers, and benefits from engagement by the wider HSPR community.

The Challenge

There is widespread agreement that developments in genetics pose challenges for healthcare and health systems, but consensus is lacking about which challenges are most important.

Many commentators expect the role of genetics in medicine to expand in the coming years as increasing amounts of genetic information are used to identify health risks and manage disease (Khoury 2003). For some, these developments promise a revolution in medicine and healthcare. Francis Collins and Allan Guttmacher, Director and Deputy Director, respectively, of the US National Human Genome Research Institute, epitomize this enthusiastic view. As they put it, "With this achievement [the completion of the human genome project], humankind has crossed into new territory … there is a high likelihood that medical care will undergo a transformation as a consequence" (Collins and Guttmacher 2001). They add, "the practice of medicine has now entered an era in which the individual patient's genome will help determine the optimal approach to care, whether it is preventive, diagnostic or therapeutic. Genomics

… is poised to take center stage in clinical medicine …" (Guttmacher and Collins 2002: 1519–20).

Yet others question the revolutionary potential of genetics, suggesting inherent limitations in our ability to predict or manage disease with the fruits of genomic science. Holtzman and Marteau (2000) argue that genetic information will not be sufficiently predictive to guide medical practice for most common and complex diseases. Even in rarer cases, where inherited susceptibilities are clearly defined by genetic tests, the gap between the ability to diagnose and the ability to treat (the "therapeutic gap") remains. Meanwhile, Carlsten and Burke (2006) question the added value of genetics in some public health efforts, suggesting that genetic information about increased susceptibility to lung cancer might lead to greater fatalism among the more susceptible, more smoking among the less susceptible and less emphasis on highly successful societal efforts, such as smoking bans and tobacco taxes. Attention to these high-profile areas can detract from more pressing concerns – issues such as home care or the social determinants of health – that may prove more relevant to health improvement and system sustainability. For those who express such doubts, the challenge is less the science itself and more the excess scientific and public enthusiasm – termed "genohype" (Holtzman 1999) – that surrounds this area of science and technology.

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For others, the challenge of genetics for healthcare arises from the marketing practices of those who control intellectual property rights (IPR). In Canada and around the world, policy attention was galvanized by the Myriad case, in which a US biotechnology company sought to assert its intellectual property rights over genes in ways that fostered debate about the appropriate scope of IPR in genetics (Heller and Eisenberg 1998) and compromised the planning, cost control and service delivery efforts of individual jurisdictions (Gold et al. 2002). In response, policy makers struck committees and ordered commissions and reports to consider the implications of such practices (Australian Law Reform Commission 2004; OECD 2000; Ontario Ministry of Health and Long-Term Care 2002; Cornish et al. 2003). While the Myriad dispute has quieted, commentators continue to disagree over whether these policy initiatives were justified and the extent of the ongoing challenge for health systems (Caulfield et al. 2006; Gold et al. 2007).

There is little doubt that developments in genetics and genomics are improving our understanding of disease. It is also increasingly clear that these developments will lead to health improvements for specific inherited conditions. But the scope and depth of additional health improvements, and the full implications of marketing practices in genetics, remain unknown. A host of complex questions need to be addressed: How can clinicians and consumers be supported to use genetic tests appropriately? What outcomes should be measured to judge the benefits of genetic information? And how should associated changes in practice be financed, funded, regulated and delivered? As Calnan et al. (2006) have argued, the real challenge for health system planners and policy makers may in fact be persistent uncertainty about what genetic and genomic science will yield for health and health systems. Policy makers must make decisions in the face of this uncertainty, alongside both high hopes and persistent "hype" about the genomic enterprise, increasing the need for high-quality health services and policy research.

The State of the Art

The path from research discoveries in genetics and genomics to improved health services is not a straight one, fostering an expansive mandate for genetic HSPR.

The Human Genome Project and associated efforts have encouraged major public and private investments in genetic and genomic science. These investments generate important discoveries, but do not lead automatically to clinically useful products and improved service arrangements. One challenge is well known: the growing quantity of basic research discoveries is not always matched by reliable epidemiological information about genetic risks across multiple populations (Ioannidis et al. 2001). But health services and policy challenges are concurrent with, rather than subsequent to, the challenges of genetic epidemiology: technology validation occurs alongside service implementation, service organization addresses both realistic and unrealistic expectations, and stakeholders debate the ends to be pursued as they contemplate current and future possibilities. Genetic technologies are increasingly relevant for large sections of the population – notable examples include prenatal diagnosis, newborn screening and genetic testing for adult-onset conditions (e.g., hereditary cancer syndromes). Service design and implementation challenges are growing, even as the gap between what is hoped for and what is possible persists.

In this context, public policy makers have been motivated to call for HSPR in genetics with an expansive mandate. In Canada, the Institute of Genetics and the Institute of Health Services and Policy Research of the Canadian Institutes of Health Research jointly sponsor a Health Services for Genetic Diseases Initiative that, since 2003, has invested approximately $5.2 million in such research. Like all HSPR, spe-

cialized HSPR in genetics seeks to identify optimal models of primary, secondary and tertiary care for the delivery of validated genetic technologies and services, support service organization and policy development to address public health needs through genomic technologies, and explore systems for the regulation, financing and funding of genetic health services. In addition, HSPR in genetics seeks to develop novel strategies to inform public policy and engage stakeholders (including lay and provider populations) in deliberations regarding the goals of genetic healthcare and ways to manage often-inflated expectations. To this end, the genetic HSPR community has a sustained history of engagement with ethical, legal and social questions (known as ELSI, or GE3LS). Further, the community has developed and used international networks and engaged policy makers at all levels (macro, meso and micro) (Burke et al. 2006). In recent years in Canada, several large research teams have emerged that address this broad mandate, including Apogee-Net, a knowledge network dedicated to supporting policy making in genetics (www.aetmis.gouv.qc.ca/site/en_apogee.phtml), GeneSens, evaluating health service and knowledge translation interventions in genetics (www.genesens.net) and CanGèneTest, examining the organization of genetic laboratory services, including technology assessment, regulation and public health programs (www.cangenetest.org).

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The Proposal

With its broad mandate and analytic approach, insights from genetic HSPR are relevant to HSPR as a whole. Yet continued engagement with the broader HSPR community is essential if these benefits are to be realized.

Genetic HSPR has evolved with a clear focus and an expansive mandate. But does this specialized domain have anything to offer its parent discipline? And what should be the relationship between genetic HSPR and HSPR? We argue that the genetic HSPR community can ill afford to become detached from the broader HSPR community. At the same time, lessons learned from the exploration of genetic health services and health policy can inform the HSPR enterprise as a whole.

It is not necessary to suggest that genetic technologies pose unique social and

health system challenges to argue that distinctive insights can be generated from specialized HSPR in this area. Genetic HSPR is characterized by its engagement with ethical, legal and social issues; by the determination of many of its practitioners to explore genetic health technologies as complex interventions; by the need to consider the impacts of genetic technologies across all domains of healthcare; and by the vexing questions posed where inflated expectations exist alongside limited evidence. None of these characteristics of genetic HSPR is unique to this domain, but the convergence of these characteristics renders this specialization a rich source of methodological, theoretical and empirical insight.

The longstanding engagement of genetic HSPR with ethical, legal and social issues increases the sensitivity of the wider HSPR community to the broad implications of technological innovation and alerts practitioners to the influence of such factors in service and system change. In addition, like other healthcare innovations in which practices and understandings, not only outcomes, may be altered, the use of genetic science and technology in healthcare warrants attention as a complex intervention (Campbell et al. 2007). The "complex intervention" framework enhances the need for the full spectrum of research methods and fosters a reflexive and theoretically sophisticated approach to design and evaluation. Further, HSPR for genetics encourages exploration across all levels of the healthcare system. Genetics is relevant to most of healthcare, including primary care (e.g., family history, risk assessment and referral), secondary and tertiary care (e.g., risk assessment and management) and public health (e.g., newborn screening), with complex interactions across these many arenas. Finally, the expansion of genetic medicine raises fundamental evaluative, organizational and policy questions that have relevance beyond this particular instance, including coverage decisions (how can decisions about public subsidy be made quickly in the face of limited evidence?), service organization challenges (how can inter-professional learning and collaboration be fostered?) and system design dilemmas (what are the implications of private access for the utilization of public services?).

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The appeal of genetics for those who practise genetic HSPR has encouraged many to become highly specialized, with substantive knowledge of genetics alongside expertise in the organization and delivery of health services and relevant evaluative and analytic methodologies. We have suggested that the gains from this specialization

may be considerable. But are there also hazards? Might the genetic HSPR community become a closed clique, with insufficient external peer review and critique to sustain rigorous knowledge production? In our view, this risk is real. For HSPR in genetics to sustain excellence, it requires ongoing cross-fertilization by experienced researchers with interests in associated areas: health system domains that might be affected by genetics (primary care reform, screening, telemedicine) or cognate specializations (e.g., imaging research). These interactions promise to enhance the nascent specialization of genetic HSPR and enrich research in the wider HSPR community.

Conclusion

It has been more than 50 years since Watson and Crick identified the structure of DNA. During that time, there have been successive waves of anticipation and dread about the implications of genetic science for medicine and society. As the double helix approaches middle age, we remain uncertain about the ultimate effects of this new knowledge. Yet, clarification will not come from examination of genetic science and technology alone. Rather, it relies on research that takes the labyrinth of healthcare and health systems as its focus. Genetic health services and policy research will play a crucial role in informing decision-makers at all levels of the health system about whether and how to integrate developments in genetics, genomics and other complex new technologies. To be of the best quality, this research must engage the wider health services and policy research community.

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REFERENCES

Australian Law Reform Commission. 2004. *Genes and Ingenuity: Gene Patenting and Human Health*. Melbourne: Commonwealth of Australia.

Baird, P.A. 2001. "Will Genetics Be Used Wisely?" *Isuma Canadian Journal of Policy Research* 2(1): 94–101.

Burke, W., M.J. Khoury, A. Stewart and R.L. Zimmern. 2006. "The Path from Genome-Based Research to Population Health: Development of an International Public Health Genomics Network." *Genetics in Medicine* 8(7): 451–58.

Calnan, M., D. Wainwright, P. Glasner, R. Newbury-Ecob and E.B. Ferlie. 2006. "'Medicine's Next Goldmine?' The Implications of New Genetic Health Technologies for Health Service." *Medicine, Health Care and Philosophy* 9: 33–41.

Campbell, N.C., E. Murray, J. Darbyshire, J. Emery, A. Farmer, F. Griffiths, B. Guthrie, H. Lester, P. Wilson and A.L. Kinmonth. 2007. "Designing and Evaluating Complex Interventions to Improve Health Care." *British Medical Journal* 334: 455–59.

Carlsten, C. and W. Burke. 2006. "Potential for Genetics to Promote Public Health: Genetic Research on Smoking Suggests Caution about Expectations." *Journal of the American Medical Association* 296(20): 2480–82.

Caulfield, T., R. Cook-Deegan, F.S. Kieff and J.P. Walsh. 2006. "Evidence and Anecdotes: An Analysis of Human Gene Patenting Controversies." *Nature Biotechnology* 2(49): 1091–94.

Collins, F.S. and A.E. Guttmacher. 2001. "Genetics Moves into the Medical Mainstream." *Journal of the American Medical Association* 286(18): 2322–24.

Cornish, W.R, M. Llewelyn and M. Adcock. 2003. *Intellectual Property Rights (IPRs) and Genetics: A Study into the Impact and Management of Intellectual Property Rights within the Healthcare Sector*. London, UK: Department of Health, Public Health Genetics Unit.

Gold, E.R., T. Bubela, F.A. Miller, D. Nicol and T. Piper. 2007. "Gene Patents – More Evidence Needed, But Policy Makers Must Act." *Nature Biotechnology* 25(4): 388–89.

Gold, E.R., T.A. Caulfield and P.N. Ray. 2002. "Gene Patents and the Standard of Care." *Canadian Medical Association Journal* 167(3): 256–57.

Guttmacher, A.E. and F.S. Collins. 2002. "Genomic Medicine: A Primer." *New England Journal of Medicine* 347: 1512–20.

Heller, M.A. and R.S. Eisenberg. 1998. "Can Patents Deter Innovation? The Anticommons in Biomedical Research." *Science* 280(5): 698–701.

Holtzman, N.A. 1999. "Are Genetic Tests Adequately Regulated?" *Science* 286(5439): 409.

Holtzman, N.A. and T.M. Marteau. 2000. "Will Genetics Revolutionize Medicine?" *New England Journal of Medicine* 343(2): 141–44.

Ioannidis, J.P.A., E.E. Ntzani, T.A. Trikalinos and D. Contopoulos-Ioannidis. 2001. "Replication Validity of Genetic Association Studies." *Nature Genetics* 29(3): 306–9.

Khoury, M.J. 2003. "Genetics and Genomics in Practice: The Continuum from Genetic Disease to Genetic Information in Health and Disease." *Genetics in Medicine* 5(4): 261–68.

Ontario Ministry of Health and Long-Term Care. 2002. *Genetics, Testing and Gene Patenting. Charting New Territory in Healthcare*. Report to the Provinces and Territories. Toronto: Author. Retrieved June 19, 2008. <http://www.health.gov.on.ca/english/public/pub/ministry_reports/geneticsrep02/genetics.html>.

Organisation for Economic Co-operation and Development (OECD). 2000. *Genetic Testing. Policy Issues for the New Millennium. OECD Proceedings.* Paris: Author.

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