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Happenings

Mapping the Human Genome: Psychosocial Impacts and Implications for Nursing

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Introduction and Background

The Human Genome Project is a fast-developing area of health research that is leading to new ways of dealing with health and disease. The information and technology gained as a result of the Human Genome Project are expected to have a profound impact on individuals and society. This paper highlights Canadian research initiatives related to genetics, including the implications of such research for nursing.

Until recent developments in the field, geneticists mainly dealt with rare disorders and prenatal or pediatric populations. To this area of practice has been added counselling and testing for adult-onset hereditary disorders, whereby individuals may be provided with information about their own risk for disease. Hereditary diseases include, among others, cancers (e.g., breast, ovarian, and colorectal), cardiovascular disease, diabetes, and some neurodegenerative diseases such as Alzheimer disease and Huntington disease. Genetic advances are leading to new approaches in the assessment of disease risk and in prevention recommendations for healthy individuals (Burke et al., 1997). Genetic treatments and prevention strategies are already being integrated into clinical medicine; this trend will continue over the coming decades as translation of research findings becomes possible for a wider and wider range of conditions. Biomedical advances are increasing our understanding of genetic differences in pharmacological sensitivity among individuals, leading to the tailoring of medications based on inherited characteristics (Evans & Relling, 1999). Preventive or surveillance interventions will be specifically targeted to those at greatest risk, with the goal of reducing the incidence and mortality of disease. As well, careful assessment of health behaviours (e.g., with regard to smoking, diet, exercise, use of oral contraceptives) and biomedical and genetic factors in those at increased genetic risk will be a greater focus in nursing care.

Advances made by the Human Genome Project will, accordingly, raise questions about how the information will be used and interpreted by individuals, families, and society, and about the impact of such use and interpretation. Additionally, empirically based interventions will be required to assist individuals in adapting to and comprehending genetic information, in making decisions concerning risk-reducing options, and in adopting surveillance regimens.

Recent Research Initiatives

In anticipation of the full impact of the Human Genome Project, a series of special initiatives was launched, initially by the Medical Research Council (MRC) and then (with partners) by the Institute of Genetics of the Canadian Institutes of Health Research (CIHR). The late 1990s witnessed the first competition for grants to conduct pilot or small studies on ethical, legal, and social issues surrounding genetics. The MRC recognized the need for research on the potential social impacts — for example, ways in which people understand inherited risk. Perception of statistical risk is notoriously poor, even among members of high-risk families with single-gene mutations, the least complex of the genetic circumstances (Evans, Burnell, Hopwood, & Howell, 1993; Lerman, Seay, Balshem, & Audrain, 1995), and is subject to a variety of influences that can lead to misunderstandings. How will individuals respond and adapt to genetic knowledge? How will family communication and interaction be altered? The role of genetics in medicine suggests that the ways in which family members communicate with each other about illness and genetic susceptibility will have considerable impact on how genetic and family history information is disseminated and utilized (Glanz, Grove, Lerman, Gotay, & LeMarchand, 1999; Patenaude, 2001). Developmental considerations further complicate the thinking about how to talk with children and teenagers about disease risk.

Our team responded to this initial competition and has collaborated nationally with leaders across Canada, and across disciplines, in committing to the investigation of the psychosocial impact of genetic testing, particularly in the development, standardization, and testing of counselling methods and clinical tools to assist individuals and families in comprehending, coping with, and utilizing genetic information. As a result of the MRC competition, several small descriptive studies were funded, including two projects by our team: a project to develop and test a group-support intervention for women with BRCA1/2 mutations; and a program of research to develop and validate psychological instruments to examine the psychological impacts of genetic testing (e.g., an instru-

ment to measure self-concept among individuals who carry genetic mutations).

The establishment of CIHR and its institutes brought about further opportunities to support research. Several institutes joined together to hold workshops related to the social, ethical, and health-care implications of genetics knowledge. In 2003, CIHR's Institute of Genetics and Institute of Health Services and Policy Research funded two Interdisciplinary Capacity Enhancement Teams to build capacity and promote research and knowledge translation in this area. One of these teams is known as GeneSens (with Drs. Wilson, Caulfield, and Wells as principal investigators). The goals of GeneSens are to support sharing and learning about new perspectives in health services research; to improve the methods and skills of those members of the research team who are established investigators; and to identify, develop, and conduct research projects to address key questions relating to the effectiveness, efficiency, or sustainability of policies or services in genetics health care. Also, two proposals related to knowledge translation of genetic services recently received funding from CIHR. One of these focuses on disseminating new genetic knowledge to primary care practitioners in a timely fashion and is led by Dr. June Carroll, a family physician and researcher in Toronto.

One area where genetics knowledge will have a potential impact is the realm of psychosocial and behavioural response. Any test result that has implications for challenging decisions can pose a psychological burden. Examples include decisions about prevention and treatment options (e.g., increased surveillance, prophylactic surgery, chemoprevention); communication of test results to extended family members, offspring, and insurance companies; and relationship decisions, such as those concerning marriage or childbearing (Esplen et al., 2004). Relationships among siblings, parents, and offspring can be complicated by different test results. For example, it appears that some people found not to carry the mutation for Huntington disease feel rejected by their family when they no longer appear to have one of the key "bonds" (being at risk) that had tied them together (Tibben et al., 1999).

In 2003 a request for proposals entitled Staying Ahead of the Wave was designed to fund a range of reserach projects in genetics-related health services and policy. Our team, with principal investigators (Drs. Mario Cappelli and Mary Jane Esplen) and team members across Canada, was funded to conduct a clinical and training needs assessment as a first step in developing a Canadian genetics health service to meet the overall needs of individuals considering genetic testing and their families. The goal of the needs assessment is to provide baseline information for service providers, decision-makers, and funders. The assessment, which has the potential to influence training programs for genetics health

professionals, focuses on the level of psychosocial need as perceived by clinicians in direct contact with patients undergoing genetic testing for adult-onset disorders. It is intended to identify the types and level of psychosocial services currently available to such patients; determine whether psychosocial services now in place, however delivered, are sufficient to meet current and projected future needs; and identify options for meeting needs. The ultimate goal is to develop a national strategy for addressing research and service gaps.

Preliminary findings from the needs assessment indicate a lack of professionals to provide psychological support. In centres where psychosocial services are available, there is variability within multidisciplinary teams. In urban centres, for example, the team may include a psychiatrist, psychologist, or advanced practice nurse practitioner. Rural centres have limited or no access to specialty services for psychosocial support. This pattern reflects the general lack of psychosocial/psychiatric services across Canada, particularly in non-urban centres. All the centres identified psychosocial care as an important area for further development. The assessment of training programs suggests a lack of formal courses on psychosocial issues; however, lectures on risk communication and counselling are provided in graduate programs in genetic counselling.

In 2004 CIHR developed an additionl request for applications, Addressing Health Care and Health Policy Challenges of New Genetic Opportunities. Again, our team (led by Drs. Esplen and Cappelli) responded, and was funded for a proposal with two components: a review of the literature for evidence of predictors of difficulties in psychological adjustment to genetics information and an effort to develop and validate a psychological screening instrument for use across adult onset heriditary disorders; and the development of evidence-based clinical guidelines for managing distress. These tools are considered critical for the field: genetics counsellors, genetics nurses, and geneticists have cited challenges in identifying and managing distress and in screening for particular areas of adjustment difficulty, including grief issues and psychological issues related to prior experiences with the disease in the family. The emerging descriptive literature on genetic testing provides evidence for predictors of poor adjustment that can be incorporated into a screening tool. Our team comprises a number of disciplines and includes researchers, clinicians, and policy-makers, in an effort to produce a "user-friendly," clinically relevant yet evidence-based tool that will be rigorously validated.

The use of screening mechanisms, drugs, or prophylactic surgery targeted to those at increased hereditary risk will rest in large part on the psychological factors that govern acceptability of and adherence to recommendations. These factors include cultural and socio-economic differences in attitudes towards genetics and affect uptake of target treat-

ments. Behavioural research will be important not only in understanding and addressing these differing views among groups, but also in understanding what accounts for differences in views of genetics and related treatment recommendations among members of the same group or family. Empirically supported decisional tools will be required for frontline clinicians involved in genetic services to assist individuals in making difficult choices concerning risk reduction. Decisional aids are typically designed to assist individuals with regard to known risks and benefits (both physical and psychological) and personal values, as well as to help them work through difficult decisions.

Dr. Joan Bottorff's team has also been instrumental in contributing to the general literature on psychosocial impacts and has developed and conducted testing on counselling tools, such as a decisional aid (developed by Mary McCullum) to enhance decision-making on prophylactic mastectomy among women at high risk for breast cancer. Pilot testing of the aid is complete, and promising preliminary findings have implications for a future randomized controlled trial. Dr. Kelly Metcalfe is currently pilot testing a decisional aid for women with a BRCA1 or BRCA2 mutation who have not had breast cancer. The aid is intended to help women make decisions regarding breast cancer prevention and screening, including the options of prophylactic mastectomy, chemoprevention, prophylactic oophorectomy, and screening surveillance. It is designed to be used in addition to standard genetic counselling and is being pilot-tested for its impact on knowledge, decisional conflict, and cancer-related distress.

Over the past decade several teams and proposals have been funded in Canada to establish genetic and epidemiologic registries for biological and epidemiological data on probands and family members (e.g., Interdisciplinary Health Research Teams funded by CIHR; collaborative cancer registries funded by the National Institutes of Health). Registries allow for a range of hypothesis-testing studies involving the discovery of new genes related to disease and for exploration of gene-gene and gene-environment interactions, providing ample opportunity for behavioural research. CIHR has funded two large Interdisciplinary Health Research Teams in cancer (e.g., breast; colorectal); both include behavioural researchers and separate studies to investigate psychosocial impacts and interventions (M. Dorval in Quebec; Dr. Esplen in Toronto).

In relation to health professional education, Dr. June Carroll leads a research program focused on the education of primary care providers. Her team has developed a number of educational tools to help providers gain knowledge in genetics and to translate new information into care. Dr. Esplen and her team collaborate closely with Dr. Carroll to integrate new knowledge on the psychosocial and behavioural aspects of care.

In relation to nursing roles, Dr. Bottorff has taken a leadership role in Canada in systematically reviewing the literature and studying current and emerging nursing roles in genetics services (funded by CIHR). A recent series of papers, focused on a literature review, describe current roles and important factors in developing future roles for nurses in Canada (Bottorff et al., 2004; Bottorff et al., 2006; Bottorff, McCullum, et al., 2005). This work is significant in that it points directly to the wide gap in nursing training in Canada. In contrast to the situation in the United States and the United Kingdom, Canadian training programs for nursing include little or no formal education in genetics. A few faculties report lectures or integration of genetic information within existing courses; the content, however, is minimal. This absence of training is troublesome: a recent survey (Bottorff, Blaine, et al., 2005) found that most nurses are already encountering issues surrounding genetics and related risks in their practice and believe that they will have a significant role to play in genetics health care in the future. Nurses report virtually no formal training in genetics and a lack of confidence related to these clinical situations. Nurses currently working in the field of genetics health care have been pioneers in carving out innovative roles (Bottorff et al., 2006). They have mostly been trained and mentored by other disciplines and have sought out workshops outside of Canada to enhance their knowledge, often obtaining certification or registration in the United States. The identified roles for both specialty and general nurses vary, but usually include taking a family history, communicating risks, providing psychosocial counselling, and supporting family communication. Training programs have been established, particularly in the United States but also in the United Kingdom, at all levels of nursing education (undergraduate and graduate) and include specialty in-depth training as well as continuing education workshops (Bottorff, McCullum, et al., 2005).

The National Coalition for Health Professional Education in Genetics (NCHPEG), a multidisciplinary organization in the United States, has initiated a national effort to promote health professional education. The organization has outlined core competencies recommended for all health professionals involved in care and recommends a basic level of genetics knowledge, terminology, and skill. While medical schools in Canada have begun to take some steps in this direction, the goals of NCHPEG are yet to be reached among nursing programs and providers in Canada. The goals can be achieved through increased awareness of the Human Genome Project, its health-care implications, and the efforts of current nurse leaders in the field.

In summary, the Human Genome Project is an exciting initiative that will lead to revolutionary new treatments and preventive programs. The

opportunities for nursing care and research are vast. Nurses represent the largest group of health-care providers, and, as more genes are cloned and as more information on disease risk/genomics health care becomes available, there will be a need for professionals to provide new genetics services and to translate new technologies into health care, as there will be an insufficient number of specialists (e.g., genetic counsellors, geneticists) to meet all of Canada's health-care needs. Nurses will have to be equipped to identify individuals at risk for disease and individuals who may stand to benefit from targeted treatments and preventive measures. Nurses in most health areas will be involved in the coordination of care and in communicating with patients and their families concerning new technologies and health promotion. Opportunities exist to develop training programs and courses to address current knowledge gaps and to evaluate educational models. Nurse scientists are in an ideal position to lead research on the health-care and psychosocial aspects of genetic knowledge. CIHR's research initiatives present several opportunities for training graduate-level nurses and for nurse researchers to lead or collaborate with the multidisciplinary teams that are often required to generate new knowledge in this area. Ultimately, generating and applying new knowledge on the psychological and social implications will result in optimal care for Canadians.

References

Bottorff, J. L., Blaine, S., Carroll, J. C., Esplen, M. J., Evans, J., Nicolson Klimek, M. L., Meschino, W., & Ritvo, P. (2005). The educational needs and professional roles of Canadian physicians and nurses regarding genetic testing and adult onset hereditary disease. *Community Genetics, 8,* 80–87.

Bottorff, J., McCullum, M., Balneaves, L. G., Esplen, M. J., Carroll, J., Kelly, M., & Kieffer, S. (2004). Proceedings of the "Nursing in the Genomic Era: A Canadian Planning Forum." *Canadian Nurse, 100*(8), 24–28.

Bottorff, J. L., McCullum, M., Balneaves, L. G., Esplen, M. J., Carroll, J., Kelly, M., & Kieffer, S. (2006). Establishing roles in genetic nursing: Interviews with Canadian nurses. *CJNR, 37*(4), 96–115.

Bottorff, J. L., McCullum, M., Balneaves, L., Kelly, M., Esplen, M. J., Carroll, J., & Kieffer, S. (2005). Canadian nursing in the genomic era: A call for leadership. *Canadian Journal of Nursing Leadership, 18*(2), 56–72.

Burke, W., Daly, M., Garber, J., Botkin, J., Kahn, M., Lynch, P., McTiernan, A., Offit, K., Perlman, J., Petersen, G., Thompson, E., & Varrichio, C. (1997). Recommendations for follow-up care of individuals with an inherited predisposition to cancer: II. *Journal of the American Medical Association, 277,* 997–1003.

Esplen, M. J., Hunter, J., Leszcz, M., Warner, E., Narod, S., Metcalfe, K., Glendon, G., Butler, K., Liede, A., Young, M. A., Kieffer, S., DiProspero, L., & Irwin, E. (2004). A multi-centre phase II study of supportive-expressive group therapy

for women with BRCA1 and BRCA2 mutations. *Cancer, 101*(10), 2327–2340.

Evans, D., Burnell, L., Hopwood, P., & Howell, A. (1993). Perception of risk in women with a family history of breast cancer. *British Journal of Cancer, 67,* 612–614.

Evans, W., & Relling, M.V. (1999). Pharmacogenetics: Translating functional genomics into rational therapeutics. *Science, 286,* 487–491.

Glanz, K., Grove, J., Lerman, C., Gotay, C., & LeMarchand, L. (1999). Correlates of intentions to obtain genetic counseling and colorectal cancer gene testing among at-risk relatives from three ethnic groups. *Cancer Epidemiology Biomarkers and Prevention, 8,* 329–336.

Lerman, C., Seay, J., Balshem, A., & Audrain, J. (1995). Interest in genetic testing among first-degree relatives of breast cancer patients. *American Journal of Medical Genetics, 57,* 385–392.

Patenaude, A. F. (2001). Genetic testing and family relationships. In B. Sarason & S. Duck (Eds.), *Personal relationships: Implications for clinical and community psychology* (pp. 43–59). Chichester: John Wiley.

Tibben, A., Vegter-van der Vlis, M., Niermeijer, M. F., van der Kamp, J. J. P., Roos, R. A. C., Rooijmans, H. G. M., Frets, P. G., & Verhage, F. (1999). Testing for Huntington's disease with support for all parties. [Letter.]. *Lancet, 335*(8688), 553.

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