Main Submission

Communicating genomic risk in primary health care: Challenges and opportunities for providers

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Introduction

Using genomic data to guide patient health management is a major advancement in health care delivery. Genomics is the study of: a) a person's genetic material; b) how the components of that genetic material interact; c) how that material interacts with the environment; and d) the resulting phenotypic changes.[1] Genomic information can help guide treatment and dosing and identify those at risk for developing certain conditions (e.g., genetically influenced cancers, such as BCRA1 breast cancer).[2] Traditionally, discussing genomic information with patients has been the responsibility of genetic counsellors. However, genomics will be increasingly integrated into primary health care as patients seek genomic information,[3] including direct-to-consumer testing. This is particularly important considering the increased emphasis on patient engagement and patient-centred care. Although there are many opportunities for genomics, this rapidly advancing area presents many social, ethical and legal challenges. In the primary health care (PHC) context, one of the most significant difficulties is the communication of genomic data and genomic risk to patients. Genomic risk communication includes discussing the physical, psychological and social risks and benefits of accessing and interpreting genomic information. This review will address the context and challenges of genomic risk communication and highlight implications of successful risk communication for PHC.

Risk Communication and Genomics

Communication of genetic data with patients highlights the need for thoughtful education, risk communication, and emotional support strategies.[4] Genetic information dramatically increases the complexity of risk normally associated with disease. For example, gene variation, detected in genetic testing, increases the likelihood of, but does not ensure certainty in disease expression.[2] Alternatively, a certain gene variation might be expressed only through interaction with the environment.[1] Additionally, the implications of risk communication differ depending on whether they pertain to developing a disease or transmitting genetic conditions.[5]. These are uncertainties and implications that need to be discussed between patients and providers.

Challenges of Communicating Genomic Risks in PHC

Communicating meanings of uncertainty, risk, and statistics in genetic information will be difficult.[6] Increasingly, decision aids are being used for risk communication and patient education in informed decision-making.[4] Decision aids are "tools that help people involved in decision making by making explicit the decision that needs to be made, providing information about the options and outcomes, and clarifying personal values."[7] A recent review of risk communication interventions found that decision aids improved knowledge, but did not necessarily decrease anxiety.[4] However, access to decision aids prior to clinician interactions increased time for discussion and consideration of personal genetic risks.[8]

In genomic data risk communication, challenges include communicating uncertainty of risk (i.e., likelihood of developing genetic conditions), decision aid biases, and human biases in decision-making in general.[9] For example, patients are often optimistic towards their health, uncomfortable with their health protection/management choices, and struggle with presentation of statistical risk.[9] These issues increase our need to understand the role of genetic data use and communication of risk management options available to those with genetic variations in the PHC context. ▶

Implications and Opportunities for Communicating Genomic Risks in PHC

Considering the objectives of PHC, successful genomic risk communication has the potential to play a significant role in achieving these goals. Especially relevant objectives include expanding team-based approaches, focusing on prevention and management of chronic and complex illnesses, and encouraging patient engagement.[10]

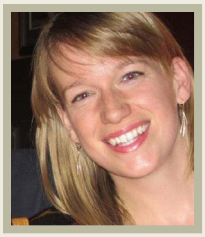
a) As previously stated, genetic counsellors have traditionally provided risk information. As genomic data becomes increasingly prevalent in PHC practice, there may be opportunities to increase the presence of genetic counsellors to collaborate in a multidisciplinary team environment to provide patient-centered care and effective risk communication. Given the reported discomfort of some providers in communicating this information, this could lead to better outcomes in primary care.

b) One of the uses of genomic information is to identify individuals at risk of developing disease, which is particularly relevant in the primary care setting. If providers are comfortable using the data and communicating the risks to patients, there is substantial potential to prevent chronic illnesses. This data will also prove useful in guiding appropriate treatment and management strategies, and avoiding ineffective and potentially unsafe interventions for patients with complex illnesses.

c) Perhaps the most important implication for communicating genomic risk in PHC is related to patient engagement. One of the fundamental goals of risk communication in health is to foster informed decision-making. This goal requires health professionals to disclose a full and honest account of all the information necessary to make an informed health decision.[11] This principle aligns with PHC practices of shared decision-making and patient empowerment.

Conclusion

If providers can successfully communicate the risks related to genetic information, they have the potential to achieve the objectives of PHC and improve patient/health system outcomes. Genomic information is playing a larger role in individuals' health care[3] and PHC is an optimal venue, logistically and economically, for discussions about genomic risk. Regardless of their preparedness, PHC providers will increasingly face the challenges of genomic risk communication as patients seek and demand genomic testing.[3] Ensuring PHC providers are equipped with the knowledge, resources and skills to communicate these risks should be a priority for health systems going forward. ■



Stephanie Kowal

Stephanie is currently a graduate student in the School of Public Health at the University of Alberta. Her thesis work comprises a community-based research project aimed at understanding how new immigrant mothers in Edmonton, Alberta make immunization decisions for themselves and their children. With the findings, she and her research partners will create information content and delivery strategies that better suit the cultural needs and day-to-day realities of different immigrant communities in Edmonton.

Derek Clark

Derek Clark is completing a Masters of Public Health in health policy and management at the University of Alberta's School of Public Health. Derek earned a Bachelor of Nursing from Mount Royal University in 2011. As a Registered Nurse, he has a clinical background in blood and marrow transplantation, hematology and oncology. Now, he is interested in conducting policy relevant research to improve the health of populations, particularly in the area of health technologies. Currently, Derek is conducting projects aimed to develop practical tools that decision-makers in Canada can use to guide their processes for orphan drugs and personalized medicines to support equitable, efficient and sustainable health care systems.

References

1. Saul RA. Genetic and genomic literacy in pediatric primary care. Pediatrics. 2013;132(S3):S198-4.
2. Prathikanti S, Weinberger DR. Psychiatric genetics-the new era: genetic research and some clinical implications. Br Med Bull. 2005;73-74:107-15.
3. Vassy JL, Green RC, Lehmann LS. Genomic medicine in primary care: barriers and assets. Postgrad Med J. 2013;89(1057):615-1.
4. Edwards A, Elwyn G. Shared decision-making in health care: achieving evidence-based patient choice. 1st ed. New York, NY: Oxford University Press; 2009.
5. Huiart L, Eisinger F, Stoppa-Lyonnet D. Effects of genetic consultation on perception of a family risk of breast/ovaria cancer and determinants of inaccurate perception after the consultation. J Clin Epidemiol. 2002;55:665-2.
6. Schwartz LM, Woloshin S, Welch HG. Using a drug facts box to communicate drug benefits and harms: two randomized trials. Ann Intern Med. 2009;150:516-11.
7. Ottawa Hospital Research Institute. Patient decision aids [Internet]. Place of publication unknown. 2013 [last update unknown; cited 2014 Mar 7]. Available from: https://decisionaid.ohri.ca/.
8. Green MJ, Peterson SK, Baker MW. Use of an educational computer program before genetic counseling for breast cancer susceptibility: effects of duration and content of counseling sessions. Genet Med. 2005;7:222-7.
9. Chapman GB, Weber BJ. Decision biases in intertemporal choice and choice under uncertainty: testing a common account. Mem Cognit. 2006;34(589):13.
10. Hutchison B, Levesque JF, Strumpf E, Coyle N. Primary health care in Canada: systems in motion. Millbank Q. 2011;89(2):256-32.
11. Jardine CG, Hrudey SE, Shortreed JH, Krewski D, Furgal C, Craig L, et al. Risk management frameworks in human health and environmental risks. J Toxicol Env Health. 2003;6(569):72.