

Commentary

## Genomic medicine: considerations for health professionals and the public

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### Abstract

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Advances in human genomics are ushering in a new era of predictive, preventative and personalized approaches to medicine. However, as the integration of genomic medicine progresses, the health community has a responsibility to communicate to the public the risks and challenges of genetic information. A possible knowledge transfer framework is outlined as a means to bridge the practical uses of genetics within various ethical, social and economic contexts. Tools and resources are needed to help clinicians understand genetic risks and help them inform the public appropriately and effectively.

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### Introduction

After decades of identifying genes involved in single gene diseases, large-scale genotyping technologies with one million single nucleotide polymorphisms per sample now enable the association and identification of genes involved in complex diseases. Genome-wide association studies identify hundreds of commonly occurring gene variants that are thought to result in an increased risk for some common complex conditions such as type I/II diabetes, Crohn's disease, coronary artery disease and several forms of cancers. Furthermore, developments such as expression arrays allow the identification of genes that are active in normal and diseased cells. These advances in human genomics are ushering us into a new era of predictive, preventive and personalized approaches to medicine. Hopefully, this will allow individuals to use their genetic information to avoid or minimize the risk of serious disease, to adopt preventive strategies to cope with common chronic conditions, and to have their medical care targeted to their genetic profile and tailored to their needs [1]. It is thus expected that health professionals of various specialties will face increasing demands to integrate genomic medicine into their practices,

and challenges that include recognizing patients who should be referred for genetic testing, ordering and interpreting tests, communicating risk information, promoting prevention strategies, providing advice to patients about the meaning of genetic variations, prescribing drugs and responding to patients seeking information after receiving direct-to-consumer (DTC) test results [2].

This raises questions about whether physicians and other health professionals are prepared to respond to these challenges and whether they have adequate knowledge about modern genetics and genomics [3]. Most physicians have no formal training in genetics, and currently little research has focused on their understanding of the recent developments in genomics [4,5]. It is unclear how physicians will handle genetic test results, address uncertainty associated with the lack of therapeutic intervention, evaluate and communicate positive or negative results (especially when the test has a limited ability to predict whether the gene variant will result in disease), translate population screening statistics into individual information for a patient, and/or react to possible

biases in the popular press or from an overenthusiastic scientific community [6]. In addition, how will the physician address concerns about the possible impact of positive genetic results within families, or the possible disclosure of the genetic information to third parties? Also, while public demand for gene testing appears to be increasing, there are concerns about a lack of public understanding of the complex genomic information and the potential social harms associated with the disclosure of genetic information [7].

## Discussion

### Integrating genomic medicine into the clinic

A recent review of the literature has concluded that, currently, there are few models for integrating genomic medicine for common diseases into primary care. Moreover, front-line health professionals are not confident of their genomics/genetic knowledge. Patients are ill-informed and seem to have inflated expectations about the value of genetic testing. Finally, there is little scientific evidence regarding the clinical utility of genomic interventions [8].

While reliable scientific evidence is needed, the knowledge generated in a research laboratory is almost never ready for transfer to the clinic, partly because of the lack of integrative approaches. As McBride *et al.* point out [1], it is important to incorporate various perspectives into research projects studying the integration of genetics/genomics into clinical practice. A knowledge transfer framework developed by Landry [9] presents a useful outline for understanding the values and beliefs expressed by patients and health professionals to help guide the process of knowledge transfer so that, for example, systematic reviews and guidelines match the reality and expectations of the end users. The framework seeks to identify, create, transform and transfer knowledge while paying particular attention to various legal, ethical, social and economic contexts. The four phases of the framework address: (1) identification of knowledge-based opportunities, (2) transformation of knowledge-based opportunities into new or improved products, services and practices, (3) communication of the developed knowledge within an organization or with other organizations and (4) application of the value of the communicated knowledge. Value is created when the quality of products or services has been improved, for example at the level of health and safety, profitability, ease of use and so on.

### Health professionals' understanding of genomic information

While geneticists play a key role in the delivery of genetic and genomic medicine of single gene disorders, the integration of genomics for common diseases will give more responsibilities to other healthcare professionals. This is especially so within the primary healthcare field where family doctors are expected to play a key role. Yet, providing risk information on complex diseases remains a major challenge. An exercise carried out during the 2006 National

Coalition for Health Professional Education in Genetics (NCHPEG) meeting in Bethesda illustrates the difficulty with interpreting genetic risk information. Each attendee received a name tag with a risk estimate of obtaining a disease and was labelled as either having a 96% chance of being unaffected with a disorder, a 1 in 25 chance of being affected, or a 4% chance of being affected. The attendees were asked to choose whether they were in a high-, moderate- or low-risk group. Three distinct groups formed in the lecture hall, indicating that most genetic professionals failed to recognize that they were all in the same low-risk group [10]. This shows that development of new tools and educational approaches is needed to help clinicians understand the concepts of genetic risk more effectively.

Additionally, no more than 3% of published clinical research in genomics moves beyond the initial phase of basic genome-based discovery into a health application (for example, risk-assessment strategies, decision or patient management) [11]. This number is disappointingly low, but this gap is being redressed by the Evaluation of Genomics Applications in Practice and Prevention (EGAPP) Working Group, set up by the Centers for Diseases Control and Prevention (CDC). EGAPP aims to evaluate evidence, and to assess genetic tests and other genomic applications as they are being introduced into the clinical or public health environment. EGAPP reviews will provide guidance on the appropriate use, for example, of genetic tests in the clinic [12]. There will be a need for large clinical studies to identify risk factors involved in health and illness of large populations, which, needless to say, will require additional funding and a close collaboration between all health professionals to accelerate the translation of these discoveries and to help narrow the gap between bench and bedside.

### Public understanding about genomic information

The public's knowledge of genomics is low but their attitude toward genomic medicine is generally positive [13]. DTC genomic testing further intensifies the need for enhanced educational initiatives, as the public will require additional knowledge and resources in order to make informed choices in relation to their genetic "profile" being seemingly predictive of future illnesses [14]. For example, some patients overestimate their own risk or have difficulty understanding that a breast cancer gene mutation can be carried by a male. For some, it is not inconceivable that genetics may push patients to adopt a deterministic attitude or "genetic fatalism" following the disclosure of their risk status. Increasingly, family doctors are reporting that their patients are likely to be interested in the genetic causes of the disease and the potential benefits of genetic testing.

### Education to improve health professionals' and public's knowledge about genetics

Given the rapid pace of genomics research, health professionals will require access to continuing medical education

resources that keep abreast of scientific advances. A good example of such an initiative is provided by the NCHPEG, an interdisciplinary group of diverse health professional organizations, consumer groups, and private and governmental organizations that provides resources to integrate genetic information into all levels of professional education [15]. Educational resources for the public have been developed by governmental organizations such as the CDC [16], and via policy databases by non-governmental groups, including HumGen International [17].

Many international organizations also support community engagement to facilitate the educational process for the public. The public expects to know more about what is going on, and wants to be active in matters of scientific policy making and research [18]. However, enabling public engagement is complex, costly, and raises a number of practical questions, including just who the public is (for example, individuals or groups, patients, stakeholders?), what approaches are the most effective (for example, consultation, partnership, public deliberations?), and how the impact and contributions of different models of public engagement can be evaluated and compared. The field of genetics is changing the way health care is practiced. In addition, as the public hears more and more about genetics, health professionals will need to know how to answer the questions asked by their patients. However, health professionals have not always kept up to date with the genetic advances, let alone with the ethical and professional challenges. For example, patients have concerns about the usefulness of the risk information, how genetic information can affect insurance and how it will impact on their family and on their children [19]. To meet these challenges, there has to be a corresponding increase in efforts to improve medical education, in particular by placing genetics, genomics and risk assessment into the core medical curriculum.

## Conclusions

Recent genomics-related discoveries and biotechnological progress are impressive and hold much clinical promise. There is, however, a relative paucity of evidence on how health professionals are or ought to be incorporating genomics into the delivery of care. There is thus also a pressing need for greater attention to the design and conduct of integrated evaluative research, particularly with regard to risk communication. There is a similar need for an expansion of professional training programs and public education and engagement initiatives in which the social, ethical, scientific and policy implications of advances in genomics are discussed in a transparent and forthright manner. In the spirit of the White Papers that have been launched by the National Human Genome Research Institute [5,7,20], we hope by this commentary to stimulate further interest in these pressing issues, and invite both health professionals and the public to contribute their views.

## Abbreviations

CDC, Centers for Diseases Control and Prevention; DTC, direct-to-consumer; EGAPP, Evaluation of Genomics Applications in Practice and Prevention; NCHPEG, National Coalition for Health Professional Education in Genetics.

## Competing interests

The authors declare that they have no competing interests.

## Authors' contributions

DA carried out the conception and drafted the manuscript. BMK participated in the drafting of the document.

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