Main Submission

Genetic screening: A cautionary tale for the public and a need for greater public education

Anita Acai and Naythrah Thevathasan

McMaster University & Queen's University

In a 2013 article written for The New York Times, American actress Angelina Jolie announced that she had chosen to undergo a double mastectomy after learning that she was a carrier of the BRCA1 mutation[1]. Her story led to unprecedented media coverage and an increased public awareness of genetic screening globally. However, according to a survey of the American general public, while 75 percent of respondents were aware of Angelina's story, fewer than 10 percent had an appropriate understanding of how to interpret her screening results and her relative risk of cancer[2]. Recent advances in our knowledge of genetics and increased media coverage of stories like Angelina's have increased public awareness of genetic screening. Unfortunately, this awareness has not necessarily translated into an improved understanding of its purpose and implications.

A direct result of the recent publicity of genetic screening has been an increased consumer demand for this health service. Research on the "Angelina Jolie effect" in the UK has shown that referrals for genetic screening more than doubled in the months after Angelina's announcement, and remained at that level for nearly five additional months[2]. While it has historically been physicians and genetic counselors ordering tests and explaining results to patients, genetic information is now readily available at an individual's fingertips. With the introduction of self-screening kits into the Canadian market, individuals can now order a kit from 23andMe Inc. for only $199. With the provision of a saliva sample, they receive information on genealogical and health information based on more than 200 genetic markers[3].

The problem with these screening kits – and genetic screening in general – is that they have limited clinical utility[4]. Simply taking a test and getting the results does not guarantee improved health outcomes. Therefore, in deciding whether or not to undergo genetic screening, one must carefully evaluate whether the information obtained from the test is likely to be useful in directing clinical care and if the value gained from the information outweighs the costs of obtaining it. This is also true in policy decisions where it is necessary to evaluate the full clinical utility of genetic tests when making decisions related to subsidizing costs in a public healthcare system.

Another problem with the widespread availability of genetic testing is that the general public may not have an adequate level of knowledge to interpret their screening results. For example, a study in 2004 found that while most respondents had conversational familiarity with genetic terminology, they became increasingly frustrated and hesitant when they were asked to specifically define these terms or to discuss the location of genes in the human body[5]. Study responses showed a poor understanding of basic scientific concepts, a result that has considerable implications for public health. Another study, which assessed individual responses to genomic risk information for Type 2 diabetes mellitus, showed that respondents were less informed about the social consequences of genetic testing (e.g., genetic discrimination by health insurers and employers) than about its medical uses[6]. Understanding of genetic concepts appears to be influenced by certain demographic variables such as race, education level, and age[6-8]. These variables have been shown to affect both an individual's understanding of genetic screens and the level of determinism with which they interpret their results.

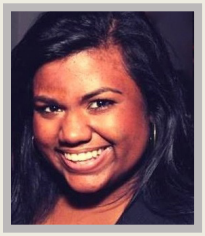
A poor understanding of genetic concepts coupled with an increased public interest in genetic screening means that consumers may be opting for genetic screens without understanding the full emotional, ethical, financial, and physical implications of doing so. An issue of primary concern is the confidentiality of results. How should the information obtained during screening be communicated, and whom should this information be shared with? For

example, the introduction of self-screening kits in Canada have led to questions about the legislation governing the privacy of results[3]. Unlike the United States, there are no similar genetic privacy or discrimination laws in Canada [9]. Thus, there is little keeping insurance companies or employers from asking about screening results and then using these results to the disadvantage of the consumer.

Given major scientific advances in genetics, there has been a significant push toward incorporating genetics into our healthcare practices. Media attention has also piqued public interest in how genetics could be used to reduce the burden of disease in society. While public awareness has translated into greater consumer demand for genetic screening, this has not been accompanied by an adequate public understanding of screening and its implications. Therefore, it is imperative that health care providers and policymakers consider the implications of mainstream genetic screening and invest in education efforts surrounding this topic. Although understanding the genetic determinants of disease is a promising field of study, its social implications deserve much greater attention than they have been given so far. ■

References

1. Jolie A. My medical choice [Internet]. The New York Times; 2013 May 14 [cited 2014 Dec 17]. Available from: http://www.nytimes.com/2013/05/14/opinion/my-medical-choice.html
2. Evans DG, Barwell J, Eccles DM, Collins A, Izatt L, Jacobs C, et al. The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services. Breast Cancer Res. 2014 Sep 19;16(5):442.
3. 23andMe - Genetic testing for ancestry [Internet]. 2015 [cited 2015 Mar 10]. Available from: https://www.23andme.com/en-ca/
4. Burgess MM. Beyond consent: ethical and social issues in genetic testing. Nat Rev Genet. 2001 Feb;2(2):147–51.
5. Lanie AD, Jayaratne TE, Sheldon JP, Kardia SLR, Anderson ES, Feldbaum M, et al. Exploring the public understanding of basic genetic concepts. J Genet Couns. 2004 Aug;13(4):305–20.
6. Haga SB, Barry WT, Mills R, Ginsburg GS, Svetkey L, Sullivan J, et al. Public knowledge of and attitudes toward genetics and genetic testing. Genet Test Mol Biomark. 2013 Apr;17(4):327–35.
7. Kaphingst KA, McBride CM, Wade C, Alford SH, Reid R, Larson E, et al. Patient understanding of and responses to multiplex genetic susceptibility test results. Genet Med Off J Am Coll Med Genet. 2012 Jul;14(7):681–7.
8. Saukko PM, Ellard S, Richards SH, Shepherd MH, Campbell JL. Patients' understanding of genetic susceptibility testing in mainstream medicine: qualitative study on thrombophilia. BMC Health Serv Res. 2007 Jun 12;7(1):82.
9. Lippman A. Why genetic self-test kits should not be allowed into Canada [Internet]. The Globe and Mail; 2014 Oct 16 [updated 2014 Oct 17; cited 2014 Dec 16]. Available from: http://www.theglobeandmail.com/globe-debate/why-genetic-self-test-kits-should-not-be-allowed-into-canada/article21124570/



Naythrah Thevathasan

Naythrah is currently pursuing her MPH (Master of Public Health) at Queen's University. Her passion for public health stems from her interest in health and education, and how the two can come together to keep the population healthy. After completing an internship with the World Health Organization in Sri Lanka, she has further developed a particular interest in health equity issues among marginalized populations. Naythrah holds a BSc in Bio-medical Sciences from the University of Guelph. She is a 3M National Student Fellow and a two-time Young Woman of Distinction Nominee.

Anita Acai

Anita is currently pursuing her MSc (Medical Education) at McMaster University. Anita's thesis research investigates how postgraduate medical programs can more effectively deliver curriculum to ensure positive learning outcomes for students. She is also involved in research projects related to competency-based surgical skills training, measures of admission (including personal and professional characteristics), and the professional development of health care workers. Anita holds a BSc (Biochemistry) from the University of Guelph where she was recognized as a President's Scholar and a