Access to genetic testing and genetic counseling in vulnerable populations: the d/Deaf and hard of hearing population

Sandra Cooke-Hubley, M Sc., MPH ·
Victor Maddalena, BN, MHSA, PhD

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Abstract Genetic testing holds great potential for preventing morbidities and mortalities for a number of diseases through early detection and effective intervention. As the number of genetic tests expand, so will public demand for these services. Therefore, it is essential to evaluate access to genetic testing and genetic services to ensure that all Canadians, including vulnerable groups, have equitable access to all forms of health care, in keeping with the mandate of the Canadian Health Act. The purpose of this paper is to examine the literature to determine if and how the Deaf community, as a vulnerable group, is at an increased risk of inequitable access to genetic services in Canada and to discuss how those who are deaf and hard of hearing are subject to the same risks. First, we define vulnerability and describe why the Deaf community, as a social group, can be considered a vulnerable group, followed by a description of the benefits of genetic testing. Second, we describe the barriers to accessing genetic testing, and how the d/Deaf and hard of hearing population experience additional barriers. Third, we examine the difficulties incorporating genetic testing into medical practice, and how this creates additional barriers to those already at risk. Finally, we discuss the steps necessary to promote equitable access to genetic testing among the d/Deaf and hard of hearing populations within Canada, and provide recommendations for further research in

this topic area. Lastly, we comment on how barriers to genetic testing vary among the d/Deaf and hard of hearing is dependent upon the type of health care system available (whether public or private).

Keywords Genetic testing · Vulnerable populations · Deaf community · Hard of hearing population · Equitable access to medicine

Introduction

The sequencing of the human genome promised new developments in medicine, novel treatments for disease, and improved understanding of man's genetic and biological composition (Brunger et al., 2000). This biotechnological feat was such an accomplishment that scientists, politicians, and researchers alike claimed it would revolutionize not only medicine, but society itself. President Bill Clinton stated in June 2000, at the official unveiling of the genetic map,

“With this profound new knowledge, human-kind is on the verge of gaining immense new power to heal. Genomic science will have a real impact on all our lives and even more on the lives of our children. It will revolutionize the diagnosis, prevention and treatment of most, if not all, human diseases” (Collins 2010a, b, p. 674).

Mapping the human genome captured the public's imagination who viewed the undertaking as a great expedition; a revolution that would change society forever, similar to Neil Armstrong walking on the moon (Fink and Collins 2000). The potential unleashed by mapping the genome rests in our technological ability to use this code to

S. Cooke-Hubley (✉) · V. Maddalena
Division of Community Health and Humanities, Memorial
University of Newfoundland, Health Science Centre,
St. John's, NL A1B 3V6, Canada
e-mail: s.cooke@mun.ca

V. Maddalena
e-mail: victor.maddalena@med.mun.ca

predict the onset of inherited diseases. The current method for detecting disease relies on the presence of somatic symptoms, which in most instances indicates the disease has already started its course. For a set of specific but ever growing list of conditions, genetic testing—also known as DNA testing—has the ability to predict the probability of an individual developing a disease, even while the person is asymptomatic. If effective intervention is taken to prevent or curb the onset of the disease, genetic testing has the potential to drastically reduce the associated morbidities and mortalities for these diseases, and in some cases, totally prevent the disease. In addition to alleviating pain and suffering, further benefits include the reduction of hospitalizations and health care costs (Collins 2010a, b). As the use of genetic testing becomes more widespread and accepted in mainstream medicine, it is important to consider how all groups in society are able to access this novel form of medicine, especially vulnerable populations, to ensure equitable access (and hence, benefits) accruing from these services.

A well-documented body of literature provides evidence that vulnerable populations, defined as an increased relative risk of an adverse outcome (Flaskerud and Winslow 1999) experience poorer health status created in part by differential access to health services (deChesnay and Anderson 2008; Fang et al. 2009; Statistics Canada 2008; Booth and Hux 2003). However, little research exists on the potential for genetic tests and genomic medicine to create health disparities among vulnerable populations. The purpose of this article is to examine the literature to determine whether the d/Deaf and hard of hearing population is at an increased risk for diminished access to genetic testing and genetic counseling services in Canada. We will first define vulnerable populations and describe the grounds upon which the Deaf community can be considered a vulnerable population, and how the deaf and hard of hearing populations experience similar challenges. Second, we examine the benefits of genetic testing as a preventive medicine approach and cite examples of Lynch syndrome and genetic deafness. Third, we will review the difficulties associated with incorporating preventive genomic medicine into medical practice, and describe populations most likely to access genetic testing. Finally, we will describe why vulnerable populations, such as the d/Deaf and hard of hearing population may not be availing of genetic testing and the potential for health disparities that may result from this differential access. We conclude with recommendations how to increase access to genetic testing for vulnerable populations, specifically focusing on the d/Deaf and hard of hearing population and comment on how these barriers to genetic testing vary depending on the type of health care system implemented within a country.

Vulnerable populations and health disparities

In general terms, individuals or social groups who experience poor physical, psychological or social health due to their social, economic or personal characteristics or group membership are, as noted above, at an “...increased relative risk of an adverse health outcome,” i.e., vulnerable (Flaskerud and Winslow 1999, p. 69). In this context, we are using the definition of a social group as proposed by Iris Marian Young (1990). She defines a social group as “...a collective of people who have affinity with one another because of a set of practices or a way of life; they differentiate themselves from or are differentiated by at least one other group according to these cultural forms” (p. 186). Social groups can be defined in terms of gender, race, culture, ethnicity, age, religion, disability, or sexual orientation (among others), and group membership often serves to afford privilege and advantage for some, and disadvantage and vulnerability for others. Examples of vulnerable groups include the homeless, indigenous people, refugees, immigrants, the unemployed, radicalized minority populations, the mentally ill, people and families living in poverty, among others. Each group is vulnerable for different reasons, and therefore should be examined individually to determine the factors that contribute to their vulnerability.

Research has shown individuals who live at the lower end of the socioeconomic hierarchy are at a high risk of developing chronic diseases and shortened life expectancy (Fang et al. 2009). The Canadian Census Mortality Follow-up Study which tracked the mortality rates of 2.7 million Canadians found the highest mortality rates among those who had less than secondary education, were unemployed or not in the labor force, worked unskilled jobs, and those who were within the lowest income brackets. Life expectancy was higher in each successively higher income groups for both sexes: for example, in the lowest fifth percentile of the income distribution, 51% were expected to live to the age of 75 compared with 72% of those in the highest fifth percentile of the income distribution (Statistics Canada 2008).

Although Canada has a public health care insurance program, statistics suggest that health disparities exist between low and high socioeconomic status (SES). Low income and lack of health benefits play a major role in these health discrepancies, but reasons for the poorer health of those in the lower end of SES status are largely situational or practical, as many are constrained by daily limitations. For example, those on a fixed income may have difficulty keeping scheduled medical appointments due to transportation costs, inability to take time off from work or child-care concerns, or limited resources to obtain adequate housing and food security (Booth and Hux 2003). Individuals who have limited income and are struggling in their

daily existence may not have the time, energy, or money for preventive health measures, even those that are considered “free”. Furthermore, publicly funded health care in Canada does not cover many preventive or dental services and medications. Typically, it is those in secure, high wage-earning jobs who are eligible for additional health care employee benefits. Unemployed and part-time employees, casual, and minimum wage earners (the “working poor”) generally do not have access to additional health care coverage, and without individual coverage, will end up paying the full cost of either of these medical services (Rogers 1997). Low levels of education and poor literacy skills can also act as a barrier to accessing health care, as those with a higher education level are better positioned to autonomously negotiate the health care system, better able to communicate their needs and more empowered to ask for a referral or treatment, and may have less fear about navigating the health system. Inadequate knowledge about preventive treatment or different ideas about its perceived value may also explain why persons of lower SES do not access preventive screening and other health services (Katz and Hofer 1994).

The deaf as a vulnerable population for poor health status

The word deaf has two distinct meanings; uppercase D “Deaf” refers to the community and culture of Deaf people, while lowercase d “deaf” (or deafness) is a medical term, which is defined as the audiologic lack of hearing ranging from hard of hearing to profoundly deaf (Canadian Association of the Deaf (CAD) 2010). Although these terms may be used interchangeably, not all persons who are deaf participate in the Deaf community, and are therefore not considered Deaf (Barnett 1993). The Deaf community is a distinct cultural and linguistic community. One of the defining characteristics of membership in the Deaf community is use of Sign Language (for example, American Sign Language, British Sign Language, etc.) as the principal means of communication. Sign Language is a nonverbal, gestural language that uses hand and body movements and facial expressions, fully exploiting the capacities of vision, spatial relations, and visual-kinetic memory (Allen et al. 2002). Within Deaf culture, deafness is not considered a disease or disability and status within the community is based on signing ability, not on hearing status (Stebnicki and Coeling 1999; Steinberg et al. 2006). The Deaf community empowers Deaf people, and encourages the view that it is society that disables and not that people who are Deaf or deaf are ‘disabled’ (Canadian Association for the Deaf 2010).

For the purposes of this article, and as noted above, we consider the Deaf community, as a social group, to be

vulnerable, i.e., at an increased relative risk of an adverse health outcome (Flaskerud and Winslow 1999). We acknowledge the inherent difficulties associated with characterizing an entire social group as vulnerable. The Deaf community is diverse and made up of individuals, many of whom would not consider themselves vulnerable. In this article, we use the term “vulnerable” in a very specific manner, and we deliberately apply this term to the Deaf community as a social group and not to individuals. Despite the ability for words to categorize and stereotype a social group, the use of terminology as descriptors can help to advance our understanding of the general features and experiences of a social group.

The Deaf population reports poorer health status than the hearing population (Steinberg et al. 2006; Zazove et al. 1993; Barnett 1993). This may be partly explained by difficulties in communication with health care providers and lack of adequate accommodations in health care facilities (e.g. access to interpreters) (Stebnicki and Coeling 1999; CAD 2010). A study of Deaf people’s experiences with the health care system in the United States determined a number of challenges, including poor understanding of clinicians’ instructions, difficulty using the TTY (telephone base communication), voice mail systems and lack of availability of medically experienced certified interpreters (Stebnicki and Coeling 1999). A US qualitative study of 91 deaf adults who communicate with American Sign Language found that many of the participants, which included Deaf, deaf, hard of hearing, and late-deafened cultural identities, expressed emotions such as fear, mistrust and frustration due to poor communication with health care providers and felt that some health care workers disliked working with deaf people (Steinberg et al. 2006). The Canadian Association for the Deaf reports that only a handful of Canadian hospitals have visual alarm systems or visual communication safety systems, and reports there is only one deaf doctor and one deaf psychologist in all of Canada (CAD 2010). Due to the reported communication barriers between deaf persons and professionals in the Canadian and US health care systems, as well as associated difficulties of access to health care due to low socioeconomic status, Deaf people, as a social group, are at higher risk than the hearing population for poorer health status, and are therefore considered vulnerable.

In terms of vulnerability, the Deaf population is often compared with members of other linguistic and cultural minority groups (Barnett 1993). Since most Deaf individuals do not speak fluent English, they face language and communication barriers that may result in discrimination. Although English literacy levels tend to be lower than in mainstream populations, most Deaf individuals have an average IQ (Pollard and Barnett 2009). Most Deaf

individuals read English at a third or fourth grade level, reflecting the difficulty of translating the hundreds of concepts and ideas from English into sign language (Steinberg et al. 2006; Newfoundland and Labrador Association for the Deaf (NLAD) 2007; Pollard and Barnett 2009; Zazove et al. 1993). In a survey of 162 Deaf adults living in Newfoundland and Labrador, 65.2% self-rated their reading skills using English language as poor or fair (NLAD 2007).

For the purposes of this paper, we are concerned with those individuals who align themselves with Deaf culture. Having said that, we also recognize persons with hearing loss who are not associated with the Deaf community, and are considered deaf or hard of hearing (a person whose hearing loss ranges from mild to profound and whose usual means of communication is speech) (CAD 2010). Many of the studies in this literature review have participants from the Deaf community as well as those who are deaf and hard of hearing populations. Therefore, the conclusions from these studies apply not only to the Deaf community, but also to the deaf and hard of hearing, and we assume that all these groups face similar communication challenges when accessing health services, including genetic counseling and genetic testing. For this reason, the issues identified in this literature review refer to the d/Deaf and the hard of hearing populations to encompass all peoples with hearing loss.

Although many d/Deaf and hard of hearing individuals have a variety of capacities and skills to offer society, many may experience unfair discrimination due to a communication barrier. As a result, educational and employment prospects for many d/Deaf and hard of hearing individuals are limited, which may contribute to the low socioeconomic status common among these populations (Pollard and Barnett 2009).

Benefits of genetic testing

Genetic testing is useful for a select number of diseases and conditions wherein the disease is caused by an identifiable number of gene(s) and has only minimal environmental influence. For these conditions, the result of a genetic test coupled with proactive screening and treatment, can be used to alter the course of a disease, or prevent the disease from occurring. Colorectal cancer (CRC) is one of the most predominant cancers affecting Canadians, with 12.4% of Canadian men and 10.1% of Canadian women estimated to be diagnosed with the disease in 2010 (Canadian Cancer Society 2010). Lynch syndrome (LS) is the most common form of hereditary predisposing colorectal cancer syndrome, comprising 3% of all cases of CRC. Ninety percent of Lynch syndrome cases are caused by mutations in

several mismatch repair genes (*MMR*), including *MLH2*, *MLH1*, *MLH6*, and *PMS2* (Lynch et al. 2009). DNA testing for mutations in *MLH1/MLH2* in individuals with a family history of LS followed by periodic colonoscopy and subsequent removal of adenomas can drastically reduce the mortality and morbidity associated with the disease (Stuckless et al. 2007). Several studies have shown that surveillance of Lynch syndrome families identified to have one of the genetic mutations, followed by proactive and early treatment can reduce the development of CRC by 60% and decreases mortality significantly (Vasen et al. 2010). In addition, women who have a germline (inborn) mutation for LS have a 20–60% risk for endometrial cancer and can benefit from periodic endometrial screening (Stuckless et al. 2007). Individuals susceptible to hereditary breast cancer and stomach cancer and who undergo genetic testing to determine their probability of developing an inherited cancer can also benefit from early detection or identification of associated risk factors (Collins 2010a, b). As with Lynch syndrome, prophylactic surgery and surveillance in those identified through a genetic test has the potential to reduce morbidity and mortality associated with these diseases.

Benefits of genetic testing for deaf and hard of hearing individuals

In addition to reducing morbidity and mortality, genetic testing may also be beneficial for their diagnostic value. Some individuals within the d/Deaf and hard of hearing populations have a genetic form of hearing loss. Mutations in the Connexin26 (*GJB2*) gene has been found to be responsible for 30–50% of all cases of autosomal recessive cases of hearing loss, and up to 30% of all types of etiogenic pre-lingual (before speech) deafness (Arnos 2003; Brunger et al. 2000; Geelhoed et al. 2009). Furthermore, genetic research on Newfoundland's founder population has identified five deafness genes (*PCDH15*, two in *TMPRSS3*, *WFS1*, and *GJB6*) which have been found to be the cause of deafness in a number of families. Furthermore, since many Newfoundlanders have a common ancestry, it is likely that nonenvironmental hearing loss in other Newfoundlanders may be caused by one or more of these genes (Doucette et al. 2009). For Deaf or hard of hearing individuals, undergoing genetic testing to determine whether their deafness is genetic may be used for themselves or family members to assist with their health care or with family planning issues (Arnos 2003). According to a study by Burton et al. (2006), Deaf adults expressed a positive motivation to seek genetic testing to clarify their own self-identity and to prepare for future children. As well, parents of deaf children may avail of the results of a positive genetic

test for deafness as it provides accurate information about the cause of deafness and about other medical implications, as well as providing an estimate of the chance of reoccurrence in other children (Palmer et al. 2009). A confirmed genetic etiology of deafness can benefit siblings of deaf individuals as well, as they may be interested to know whether they carry a gene for deafness, and this may influence their family planning options (Smith and Hone 2003).

A secondary benefit to genetic testing for deafness is the potential to identify or diagnose other health conditions that may seem unrelated to deafness, but may accompany hereditary deafness. Approximately 30% of all types of deafness are syndromic, meaning deafness presents with additional health complications such as ophthalmic disorders (Usher's syndrome), kidney dysfunctions (Alport syndrome), or heart conditions (Jervell and Lange-Nielson syndrome). Knowledge that additional physical problems may coexist with deafness can enable parents as well as educators, audiologists, and other professionals who have a responsibility for deaf children to avoid medical and educational consequences that may occur if the additional conditions are overlooked (Schein and Miller 2008).

Attitudes of d/Deaf and hard of hearing individuals towards genetic testing for deafness

A number of studies conducted in various countries have assessed attitudes towards genetic testing for deafness within Deaf communities, among deaf and hard of hearing individuals and their family members. The results of these studies should not be broadly generalized to report that the Deaf community has a negative perception towards genetic testing for deafness, as study results vary, and are dependent on factors such as affiliation with the Deaf or hearing community, previous education on genetic testing, and the actual questions being asked in the study. For example, Guillemin and Gillam (2006) in their assessment of attitudes of 19 Australians who were either Deaf or hard of hearing had a deaf family member or found that the majority were in support of genetic testing for deafness but only when fully informed about the advantages and disadvantages of genetic testing. Boudreault et al. (2010) examined the impact of genetic counseling and genetic testing on 256 Deaf and hard of hearing adults in the USA who had no prior knowledge why they were deaf and found that respondents were strongly interested in using genetic testing only to determine why they were deaf, if the cause of their deafness was genetic or to help with scientific research. However, there was little interest in using genetic information for partner selection, making decisions whether or not to have children, with the exception of those

affiliated with the Deaf communities who wanted to learn if they could have deaf children. Burton et al. (2006) has also published a study that found positive attitudes towards genetic testing within the Deaf community.

On the other hand, a number of studies have been published that convey less favorable views on attitudes towards genetic testing. Middleton et al. (1998) surveyed 87 delegates at an International Conference on the Deaf Nation held in the UK, of which 96% were culturally Deaf or identified equally with the Deaf and hearing communities. The study found that 55% of respondents thought genetic testing for deafness would do more harm than good and 46% believed that this practice devalued deaf people. A US study published by Stern et al. (2002) distributed a questionnaire to 337 d/Deaf and hard of hearing students at Gallaudet University, and found that culturally Deaf respondents were approximately four times more likely to feel that genetic testing would have a negative effect on the Deaf community than those who identified with the hearing community (OR=5.84; 95% CI, 2.76–12.38). However, the majority of adjectives checked to describe their sentiments towards genetic testing were positive feelings. A number of studies have focused primarily on d/Deaf and hard of hearing attitudes pertaining to the use of prenatal diagnosis of genetic testing for deafness, the majority of which demonstrate a strong negative attitude towards this particular use of technology (Taneja et al. 2004; Middleton et al. 2001; Martinez et al. 2003). Attitudes towards genetics and genetic counseling are explored in a recent study by Middleton et al. (2010), which recruited 573 d/Deaf or hard of hearing individuals from UK through magazine readership. Participants filled out a questionnaire, which included information on genetic counseling, and questioned them on their knowledge of and future benefits of genetics. In addition, 30 participants were selected to undergo a qualitative interview. The results showed that almost half (45%) did not understand the correct meaning of genetic counseling and >75% did not know how to access genetic counseling services. Opinions were mixed when asked if the aim of genetic counseling was to reduce deafness in society or if genetic counseling for deafness should exist. The authors concluded that some of the participants did have knowledge about genetic counseling and did not assume that its aim is to reduce deafness in society. The largest issue identified was not that d/Deaf or hard of hearing persons did not know about genetic counseling or that there exists a gross misconception about its aims, but was rather the issue of how to access a genetic counselor. Indeed, the authors of this paper have identified access to genetic services by members of the d/Deaf or the hard of hearing population as an important issue that requires further examination in order to identify the barriers to access.

Current problems with incorporating genetic testing into medical practice

Although medical genomics has the potential for early diagnosis and early treatment for individuals with inherited diseases, the practical application of incorporating genetics into routine medical practice has been met with many challenges, many of which are not easily surmountable. The variability of criteria outlining who and when to refer for genetic testing and counseling services is overwhelming, and it is difficult to uniformly apply standard procedures to identify who is eligible for, or, who may benefit from a genetic test. A common problem identified in the literature is physician confidence in making appropriate referrals for genetic testing. For example, McCahon et al. (2009) randomly distributed a questionnaire to 797 general practitioners (GPs) in England to determine attitudes towards genetic testing. Although 50% stated that they could recognize when testing was appropriate, 43% said they felt unprepared to collect family history or assess familial risk. Lack of training was a significant predictor of feeling unprepared to undertake these activities. In addition, only 51% of GPs indicated they could recognize a pattern of disease that suggested inherited familial risk and only 12% felt they had enough knowledge to assess whether family history reflected a high, moderate, or low risk (for the individual to express the disease).

Miller et al. (2009) surveyed 329 cancer physicians in Ontario to determine respondents' perceptions of the importance and accessibility of molecular oncology testing (MOT). Less than half (45%) agreed that patients in their region received MOT as a standard of care, and 70% of physicians stated there was a lack of clear guidelines regarding clinical indications. Only a minority of physicians expressed confidence in assessing whether, or which, MOT was indicated. As well, Brandt et al. (2008) designed a ten-page survey completed by 82 American physicians regarding cancer genetics referral processes. Of these, only 59% reported an awareness of the hospital's cancer genetics program. Reasons cited why patients were not referred included patients' disinterest (54%) and physicians' concern about insurance coverage or discrimination (44% and 31%, respectively).

Other barriers to genetic testing include limited knowledge on patients' eligibility and, lack of evidence of the direct benefits to patients, local guidelines, and specialist services (McCahon et al. 2009; Brant et al. 2008; Collins 2001). In addition, the general perception that genetic testing is an expensive or merely an academic endeavor may limit the integration of genomic medicine into medical practice (Taylor et al. 2006). Furthermore, there is no clear funding policy for genetic tests as resource allocation, for genetic testing varies from province to province, and may

be included under a hospital budget in some jurisdictions or directly under the provincial Ministry of Health in others in Canada (Adair et al. 2009).

Studies have shown that access to genetic services is strongly correlated with patients who act as their own health care advocates. Independent of need, it is individuals who are aware and ask for genetic services that are most likely to receive access (Hall and Olopade 2006; Hall and Olopade 2005). Brandt et al. (2008) reported patients' eligibility was based on a family history (96%), history of cancer (83%), and patient request (73%). Wideroff et al. (2003) reported that the factor most strongly associated with physician referral to genetic services is patients inquiry and testing request (OR=5.52; 95% CI), and concluded that even if a physician fails to appropriately recommend genetic counseling, informed individuals may act as their own health care advocates. From this research, we can assume that those with lower level of education, limited knowledge of the health system, and those who have existing barriers to health care because of language, income, or socioeconomic status will be most vulnerable to unequal access to genetic services as they are least able to act as their own health advocates request genetic services.

Potential for health disparities caused by differential access to genetic testing

Relatively few studies have examined differential access to, and utilization of, genetic counseling and genetic testing services among under-served and vulnerable populations. Although the research is lacking in this area, preliminary studies have found inequalities in terms of genetic testing in minority groups. For example, Hall and Olopade (2006) reviewed the population of *BRCA1* testing subjects (breast cancer gene) from Myriad Genetic laboratories in the USA and found that less than 10% of the 10,000 individuals tested who identified their race were from under-represented racial/ethnic subgroups. Suther and Kiros (2009) surveyed three ethnic/racial in the US groups to determine their knowledge of genetic testing. After adjusting for all variables, they found that both African Americans and Latinos had significantly lower knowledge compared with non-Hispanic whites. In these studies, education played a role: less than high school education was negatively associated with knowledge (OR=0.70, 95% CI), compared with high school education, having some college education (OR=1.39, 95% CI), and college or more than college education (OR=2.13, 95% CI). Limited or lack of knowledge about genetic testing within the African American and Latino populations is likely due to lower education levels associated with these groups, in addition to

a lack of information provided by their physicians about genetic testing.

Furthermore, an analysis performed by Wideroff et al. (2003) from the 27,405 respondents of the US National Health Survey determined that awareness of genetic testing was significantly associated with higher education, white race, age <60 years, female gender, private health insurance, personal or parent's history of certain cancers, and level of physical activity (among other variables). A total of 44.5% of respondents reported they had heard about genetic tests, including 49.9% of whites, 32.9% of African Americans, 32.3% of American Indians/Alaskan Natives, 28.0% of Asian/Pacific Islanders, and 20.6% of Hispanics. A large differential in test awareness (35%) was found between those with the highest level of education and those with the lowest level. This suggests that those with the least education are less informed about genetic testing.

Consequently, persons with limited education and those with language barriers will likely be among those who have limited knowledge of genetic testing and are at decreased likelihood of accessing genetic testing and genetic counseling services. This differential access to genetic testing and genetic counseling services could result in (and confluence) health disparities between less-educated vulnerable groups and well-educated nonvulnerable populations, which puts vulnerable populations such as the d/Deaf and hard of hearing populations at higher risk for not accessing genetic testing and counseling services. Although these ideas are constructed from investigating the current literature, the study by Middleton et al. (2010) previously referenced provides evidence that d/Deaf and hard of hearing individuals do indeed experience barriers to accessing genetic counseling.

Health literacy of d/Deaf and hard of hearing adults: acting as their own advocates

Lower levels of literacy have been documented among the Deaf population generally reflecting the difficulty of translating the hundreds of concepts and ideas from English into sign language (NLAD 2007; Pollard and Barnett, 2009). Since medical literature is written for individuals with higher levels of literacy, the majority of d/Deaf and hard of hearing adults may miss relevant health information, contributing to a lower level of “genetics” health literacy. Low health literacy is defined as a knowledge deficit in terms of illnesses, symptoms and causes, illness prevention, treatment options and risks, medicines, and related matters (Pollard and Barnett, 2009) Further confounding the low health literacy of many d/Deaf and hard of hearing adults is the lack of access of ambient information such as radio, television and movie soundtracks as well as everyday conversations provide hearing individ-

uals with a source of information that is inaccessible to many hearing impaired individuals. As Barnett (1993) explains, language differences and health knowledge limitations are often barriers to appropriate health care for Deaf individuals, which may contribute to the poorer health status of members in this community.

As individuals who are d/Deaf or hard of hearing are at risk for low health literacy, it is less likely they would be knowledgeable about the availability of genetic testing both for deafness and for other hereditary conditions, and therefore unlikely to request a genetic test from their physician. For the d/Deaf or hard of hearing individual who is knowledgeable about genetic testing, he/she must deal with additional barriers to communicate this request and navigate through the health system. First, they may need to work through an interpreter (if one is available) or family member who may be unfamiliar with medical terminology in general or genetic testing specifically. Second, it is unclear whether American Sign Language (or other signed languages) includes appropriate signs to explain genetic testing and related concepts (this will be explored in further research). For these reasons, as well as the lower health care utilization by the d/Deaf and hard of hearing generally, the potential to create inequitable access to genetic testing between those in the hearing and d/Deaf or hard of hearing populations exists.

Conclusions and Ideas for Future Research

The Canadian Health Act promotes the health of Canadians through principles promoting equitable access to health care. equitable access to health care. Therefore, it is imperative to address concerns and challenges that create health disparities, including access to genetic technologies to ensure equitable access to health services for all Canadians. Other international health care systems may face similar challenges as they seek to promote equitable access to genetic technologies. To the extent that genomic medicine appreciably improves quality of care and clinical outcomes, equitable access to emerging genetic-based treatments should be an essential component of any comprehensive strategy to eliminate health disparities (Peinkofer 1994). Countries that do not offer public health care will experience additional barriers to access health care services and create a greater difference in access between those who can afford health care and those who cannot. However, this is outside the scope of this article, which examines the health care system in Canada.

Genetic testing has many potential benefits that may be realized as preventive genomic medicine becomes increasingly incorporated into routine clinical practice. A number of steps can be taken to further this goal. First, physicians need to be better educated on genetic services available and have an ability to determine who can benefit from various

types of genetic testing. This would require on-going education of medical practitioners, as genomic research moves at a rapid pace and new developments occur frequently. Secondly, positive informational campaigns about genetics and the benefits of genetic testing should be developed and focused on the unique needs and characteristics of vulnerable populations and especially populations with low education and literacy levels and communication difficulties. Educating individuals about the potential benefits of genetic testing enables and empowers these individuals to act in their own self-interest (Suther et al. 2009). Campaigns should consider the unique needs of the target audience to ensure meaningful communication of concepts or ideas. For example, many d/Deaf and hard of hearing people depend on visual elements to learn new information including sign language, pictures, slides, videos, and live presentations in addition to textual information to communicate ideas more effectively (Pollard and Barnett 2009). Information on the potential benefits of genetic testing illustrated with visual elements should be available at locations where the Deaf community socializes and interact as well as health care facilities, family physician offices, or clinics. Third, health practitioners who interact with persons from these vulnerable populations need to be aware of the issues and barriers experienced in terms of health care access to create positive meaningful health care interactions and experiences. Fourth, it is important to conduct research and interact with people from vulnerable populations to incorporate their needs and concerns into social and health policies.

It is unclear whether family physicians routinely provide information to d/Deaf or hard of hearing people about genetic testing for deafness or for disease-related conditions such as hereditary cancers and cardiomyopathies. Further research is required to assess the roles of physicians and their interactions to understand how to assist d/Deaf and hard of hearing individuals make informed choices about their own health, in particular with genetic testing.

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