IMPACT OF TELEGENETICS AND ALTERNATIVE MODELS OF CARE ON HEREDITARY CANCER GENETIC COUNSELING IN BRITISH COLUMBIA*

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Resumo: *British Columbia é a província mais ocidental do Canadá e inclui uma paisagem variada de regiões costeiras do norte do Pacífico, com a população estimada em 4,6 milhões. Os serviços de aconselhamento genético e testes genéticos para populações de alto risco contribui para estratificar o risco de câncer, redução da morbidade e mortalidade relacionada ao câncer, bem como a diminuição dos custos de cuidados de saúde através da detecção e prevenção de doenças malignas mais cedo.*

Palavras-chave: *Hereditário. Aconselhamento Genético. Câncer.*

Hereditary predisposition to cancer has been observed in families for many years (WARTHIN, 1913; THORSON, 1999), however only 5-10% of cancer is due to an inherited major gene mutation (GARBER; OFFIT, 2005). Hereditary cancer syndromes include hereditary breast and ovarian cancer, (HBOC), hereditary colorectal cancer including Lynch syndrome (HNPCC) and the hereditary polyposis syndromes familial adenomatous polyposis (FAP) and MutYH associated polyposis (MAP). Less common syndromes such as Cowden syndrome, Li Fraumeni syndrome, hereditary paraganglioma, familial multiple melanoma (FAMMM) are also well described (PAGON, *et al*., 1993-2014).

The development of the human genome project in the 1990s and the identification of major genes for these syndromes has allowed for clinical genetic testing and identification of familial mutations in many families with strong histories malignancies.

GENETIC COUNSELLING FOR HEREDITARY CANCER

Hereditary cancer risk assessment is provided in a clinical setting to individuals and families with a strong personal or family history of cancer. At most genetics clinics in Canada, patients must meet eligibility criteria to attend genetic counseling and have the option to consent to publicly-funded genetic testing. The ultimate goal is to reduce morbidity and mortality for patients and families affected by hereditary cancer.

A genetic counseling consultation includes a review of patient personal and family history of cancer with confirmation of diagnoses and pathology when possible. Education is provided regarding genes and inheritance. A hereditary cancer risk assessment based on patient and family history is offered and may include both cancer and gene mutation risk prediction model estimates and interpretation. Patients are counseled on their options for genetic testing with the goal of risk stratification and management guidelines for increased cancer risks if a familial gene mutation has already been identified. Index testing involves complete analysis of the gene(s) suspected based on family history. A genetic counseling session would also include psychosocial evaluation and support, detailed facilitation of genetic testing decision making including exploring pros and cons of genetic testing such as impact on medical care, anxiety and cancer worry, third party insurability, and family relationships.

For patients who consent to genetic testing, test results are most often disclosed by a genetics specialist (genetic counselor/medical geneticist). Patients are referred to specialists as indicated for cancer risk management (eg. specialty imaging, risk reducing surgery) and psychological support, and are also invited to participate in research studies when appropriate. Genetic counseling and testing has been shown to be cost effective for hereditary cancer syndromes (BALMAÑA et al., 2004; BREHENY *et al*., 2006). These services facilitate targeted surveillance for those at high risk and secure efficient use of health care resources while reducing cancer-associated morbidity.

HEREDITARY CANCER GENETIC COUNSELING IN BRITISH COLUMBIA

The discovery of the BRCA1 and BRCA2 genes in 1994/1995 (MIKI *et al*., 1994; WOOSTER *et al*., 1995), and previous clinical assessments of families with strong histories of cancer at the British Columbia Provincial Medical Genetics Programme led to the establishment of the Hereditary Cancer Program at the British Columbia Cancer Agency (HCP) in 1999. This was a joint venture of the Provincial Medical Genetics Programme and the BC Cancer Agency. The HCP is a specialized provincial service providing cancer genetics consultations to individuals and families across BC and Yukon, and is part of the BC Cancer Agency, an agency of BC Provincial Health Services Authority (PHSA).

The HCP began with clinical staff including one genetic counselor, a part time geneticist, and a nurse educator. Currently this specialized provincial program includes two permanent sites with staff of 1 medical director, two geneticists, one nurse educator, one clinical coordinator, 8 genetic counselors, and clerical support staff. There is also one research genetic counselor.

Patients are most often referred to the HCP by a family physician, oncologist, or other specialist though 1/3 of patients are self referred. Patients and their physicians provide basic information about family history of cancer and whether a gene mutation is known in the family. Patient referrals are triaged to a genetic counselor or medical geneticist depending on the indication. Referring physicians of patients who did not meet provincial eligibility guidelines are informed by mail and resources for appropriate cancer screening information are provided.

Referrals to the Hereditary Cancer Program are most often for assessment of hereditary breast and ovarian cancer syndrome (77%). Less often referrals are for hereditary colorectal cancer (15%) and other hereditary cancer syndrome (8%). Similar distribution of reason for referral has been seen in other cancer genetics clinics in Canada (C. Handford and L. Currie, personal communication 2014) and described in an American clinic evaluation study (ZLOT *et al*., 2010).

Patients seen at the HCP are provided with cancer risk management guidelines. Patients identified to carry a high risk breast cancer gene (eg. BRCA1, BRCA2, CDH1, TP53) who are not in the care of an oncologist can be referred to a specialized High Risk Surveillance Clinic for coordination of high risk breast screening including breast MRI and referral to surgeons for risk reducing surgery as appropriate. In most other cases, cancer risk management guidelines are provided to the patient's referring physician to coordinate management. Cancer risk management strategies are based on departmental protocols generally developed from evidence based guidelines including BC Ministry of Health guidelines, National Comprehensive Cancer Network (NCCN), and Mallorca Group/Europeans Initiative Gastrointestinal Hereditary Tumours Guidelines (EIGHT).

BRITISH COLUMBIA

Population estimate for BC is 4.6 million people (BC Stats -.bcstats.gov.bc.ca, 2014). The province includes one major urban area, Vancouver (pop. 600,000) as part of Metro Vancouver (pop. 2.5 million). Cities that are of notable population but remotely located from Vancouver include Kelowna in the BC interior (pop. 180,000) and Victoria on Vancouver Island (greater Victoria pop. 345,000). In rural and remote areas, particularly the northern region of the province the population is less dense; the largest city in this region is Prince George, population 88,000. The province is also diverse in topographic and climatic areas with a temperate wet north and south coast along the Pacific Ocean, coastal and interior mountain ranges, dry interior valley regions, north and central plateau and mountains, and northeastern Great Plains. Travel in all areas, particularly the interior and north are significantly impacted by winter snow conditions.

Five health regions have been established to provide health care services to each area of the province (Figure 1). The PHSA provides services to each of the 5 health regions. The HCP and BC Cancer Agency also provides services to the Yukon Territory, located north of BC and bordering the Northwest Territory and Alaska, USA. Population of the Yukon is around 36,000 with about 2/3 of inhabitants in the capital Whitehorse (Yukon Bureau of Statistics, www.eco.gov.yk.ca/stats).

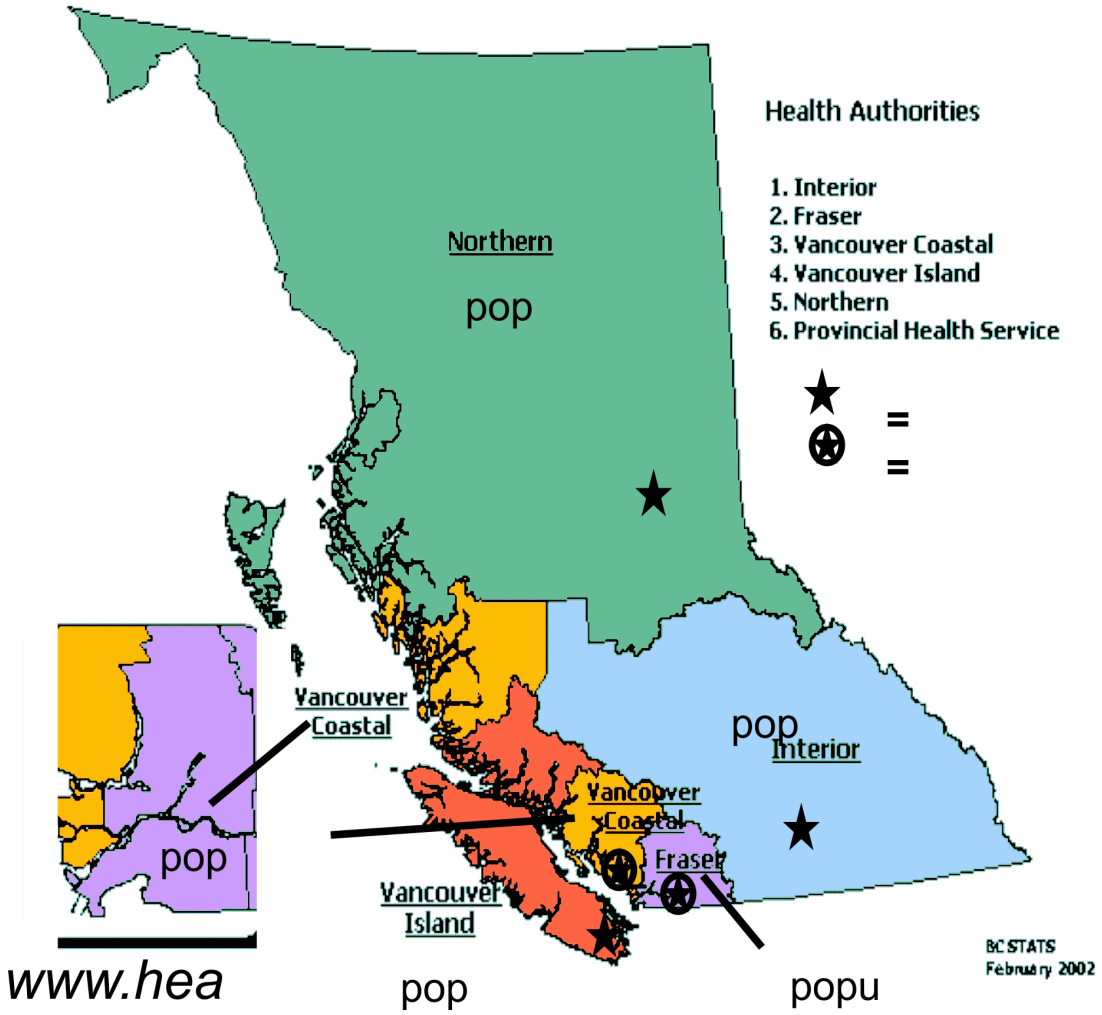

Figure 1: British Columbia Health Authorities

BC has a diverse population with many cultures and languages. In BC, the Ministry of Health Aboriginal Health Directorate 'applies an Aboriginal lens to policy development, collaborates with Aboriginal stakeholders and organizations and works with health authorities to integrate Aboriginal interests into their services' (http://www.health.gov.bc.ca/aboriginal/). The 2006 Canadian Census showed 196,000 persons in BC identifying as Aboriginal peoples (BC Stats -.bcstats.gov.bc.ca), about 5% of the total provincial population. First Nations represent 20% of the Yukon population (Yukon Bureau of Statistics, www.eco.gov.yk.ca/stats).

Over 80% of the population in BC speaks English. Chinese, Panjabi (Punjabi), Korean, Tagalog and Farsi are the most common languages spoken in BC after English. There are many Aboriginal languages spoken in BC (http://www.welcomebc.ca/). Language interpreters are available in over 150 languages for all HCP consultations when required (PHSA Language Access Policy – www.phsa.ca).

ALTERNATIVE MODELS OF CANCER GENETIC COUNSELING IN BC

At the time of the creation of the HCP in 1999, hereditary cancer risk assessment was provided to patients in both urban and remote areas. Patients could attend in-person consultations in Vancouver or Victoria. Outreach clinics also took place in specific locations in the interior and north of BC including Kelowna and Prince George, often as extra consultations added to outreach services provided by the Provincial Medical Genetics Programme for pediatric and general genetics outreach. The topography of British Columbia and the mandate to provide standard of care to patients in both urban and rural/remote locations in the province drove the need for alternative models of genetic counseling delivery.

Alternative models of genetic counseling for hereditary cancer have been a topic of interest in the genetic counseling community. A standardized set of models has been proposed by the US based National Society of Genetic Counselors (NSGC) Services Delivery Model Task Force (COHEN *et al*., 2012). This group outlines models which can be applied in a variety of health care settings, both public and private and can be employed and explored by genetic counselors, medical geneticists, and other cancer health care providers. These models include in-person counseling where follow up and results disclosure may occur by telephone or other means; telephone counseling where the call may be supplemented by written, online, or other resources; group counseling where patients are educated in a group setting with a genetic counselor and sometimes followed by a brief individual consultation; and finally telegenetics, where genetic counseling is provided remotely by videoconference (Cohen *et al*., 2012) (the term telehealth has several definitions, but herein refers to healthcare services provided remotely by videoconference). A survey of NSGC genetic counselors about service delivery models in their practice found that just over half of the responders use the in-person model exclusively and that of the remainder, those using telephone and telegenetics saw patients who lived the furthest away. Trends toward shorter wait times for consultation and shorter length of appointments in the non-traditional model users were also observed (COHEN *et al*., 2013). Models such as telegenetics, telephone counseling, and group counseling have been explored at HCP over the past 10 years.

IMPLEMENTATION OF TELEGENETICS AT HCP

A telehealth network established by the BC government first became operational in 2001 and began with specialized locations in Vancouver and 12 remote sites mainly in the North and Interior regions of BC (SCHAAFSMA *et al*., 2007). To expand cancer genetic counseling services to underserved areas of the province, a pilot study in 2003 assessed feasibility and acceptance of genetic counseling by videoconference to Prince George (northern region) and Williams Lake (interior region). Consultations were arranged by contacting IT personnel at the hospital site and discussing directly with patients who had been referred to the HCP but had not been able to travel to Vancouver for assessment. This study demonstrated that telegenetics was a feasible option for providing hereditary cancer risk assessment.

A larger evaluation pilot study completed in 2005 involved patients in six BC communities in the North and Interior regions. A total of 67 patients were invited to participate in the study and 43 consultations took place with 48 participants in total. About 50% of those who declined indicated they preferred an in-person genetic counseling consultation. Post-appointment satisfaction questionnaires demonstrated high levels of satisfaction from patients (4.68/5) and favourable (3.97/5) from genetic counselors. Major advantages reported by patients included costs-savings, convenience and comfort of familiar setting, and the ability to have family members present. Some patients indicated they would not have followed through on the HCP referral / attended genetic counseling if it wasn't available in their local community (D'AGINCOURT-CANNING, 2008). Genetic counselors found technical problems a concern, and described some difficulty in building rapport and changes in counseling style. Increased preparation time was also noted. Patients described that when possible, they would prefer to be seen in-person. However, given that pre-telegenetics waitlist for some patients from remote communities were a number of years for those not able to travel to Vancouver, the study authors concluded that telehealth was an essential option for access. This study highlighted the value of telegenetics providing a clinical service in remote communities for patients who may not otherwise have access.

In 2008 the Provincial Ministry of Health established a Telehealth Office to implement telehealth services throughout BC. A similar program was established in Yukon. Telehealth facilities supported approximately 18,000 telehealth consults through videoconferencing for fiscal year 2007-2008, with anticipated annual growth of 25% per annum[1].

TELEGENETICS SERVICE EXPANSION

In 2009 the HCP experience with telegenetics was reviewed. At that time, the HCP had been providing video consultations from the Vancouver location to 28 different remote sites around BC and Yukon. A summary of service provided to the northern and interior health regions were compiled. For these areas combined, telegenetics was not used for any of the initial genetic counseling consultations in 2005 (inclusion of the telehealth pilot study consultations for 2005 would create an increase with 5% of consultations to these regions of BC using telegenetics), but over 40% by April 2009. After the implementation of the BC Telehealth office in 2008, an increase in telegenetics service to all health regions considered remote to Vancouver were observed, with over 60% of all telegenetics appointments to the Interior region. However, the expansion of telegenetics service did not result in an observed increase in referrals from either the northern or interior health regions.

An approximate 185% increase in the total number of referrals for genetic counseling from all health regions has been observed over a 10 year period from 2003 to 2013 (Figure 2), and the proportion of telegenetics consultations has also risen. The northern health region has seen an increase in video appointments from 2003 when no appointments were provided using telegenetics, to between 50% and 70% of patients seen by telegenetics from 2011-2013 (Figure 3). The interior health

region also showed an increase to 70% of patients seen using telegenetics in 2013 (Figure 4).

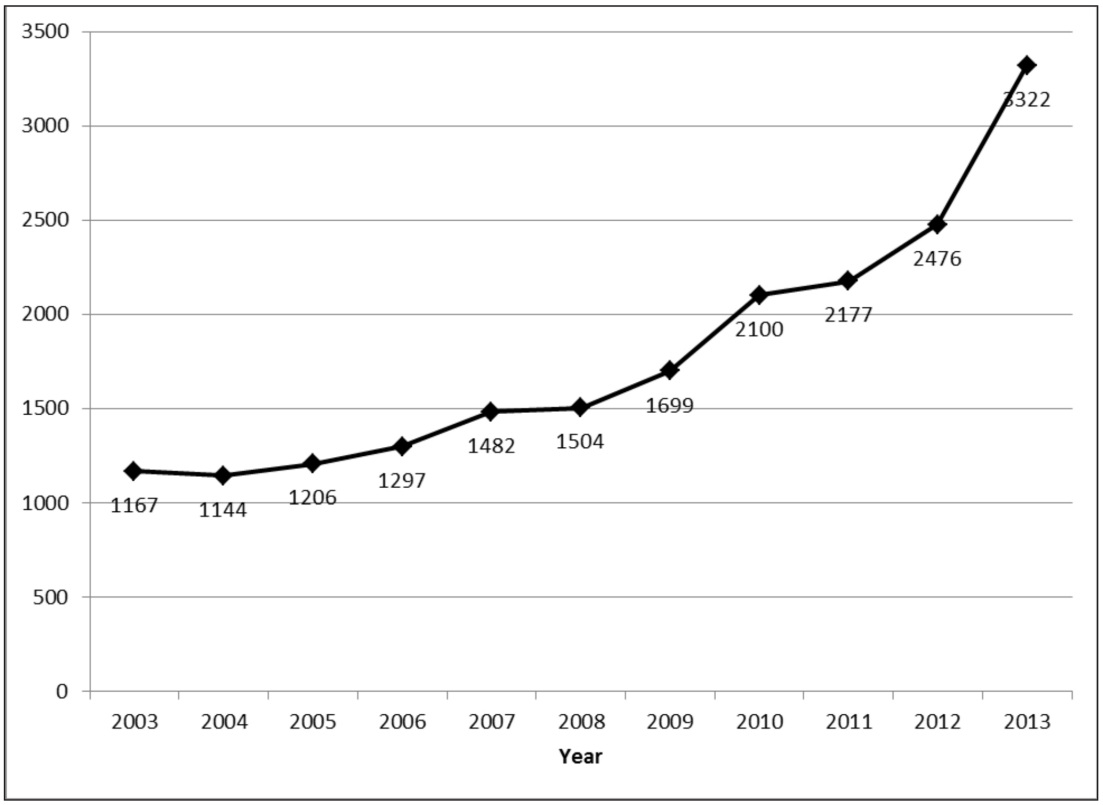

Figure 2: Genetic Counseling Referrals Received at BC Hereditary Cancer Program 2003-2013

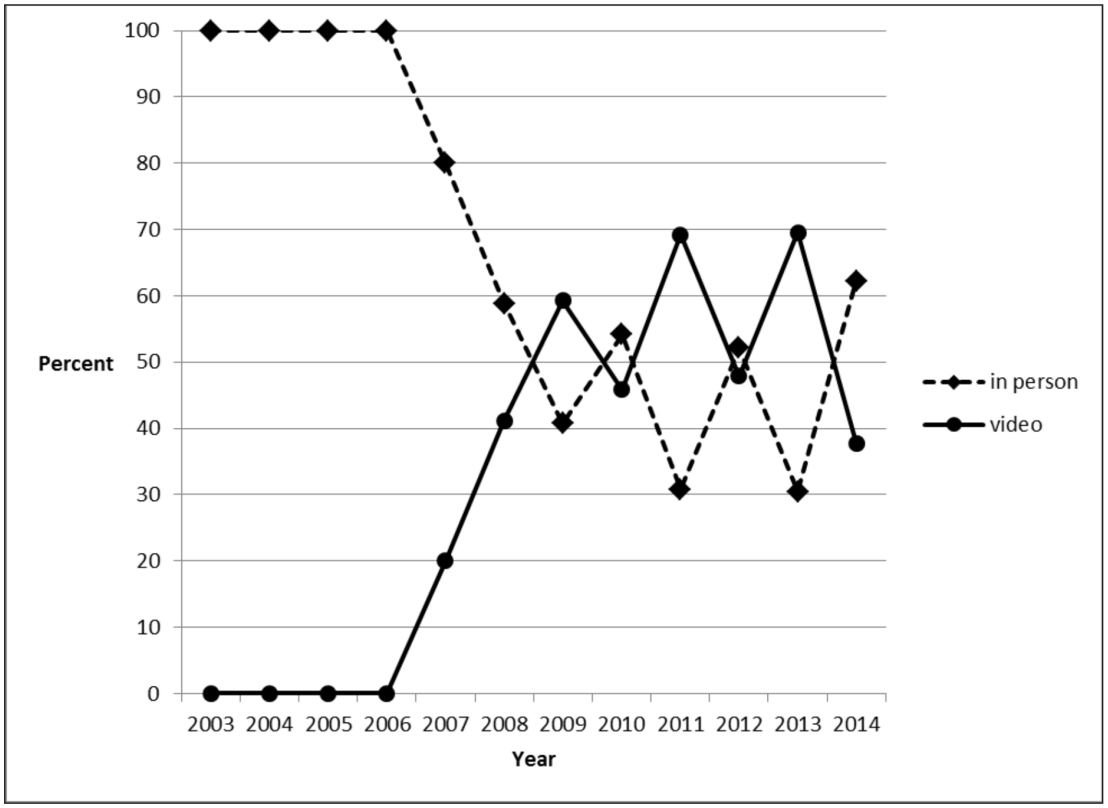

Figure 3: Initial Genetic Counseling Consultation - Northern Health Authority

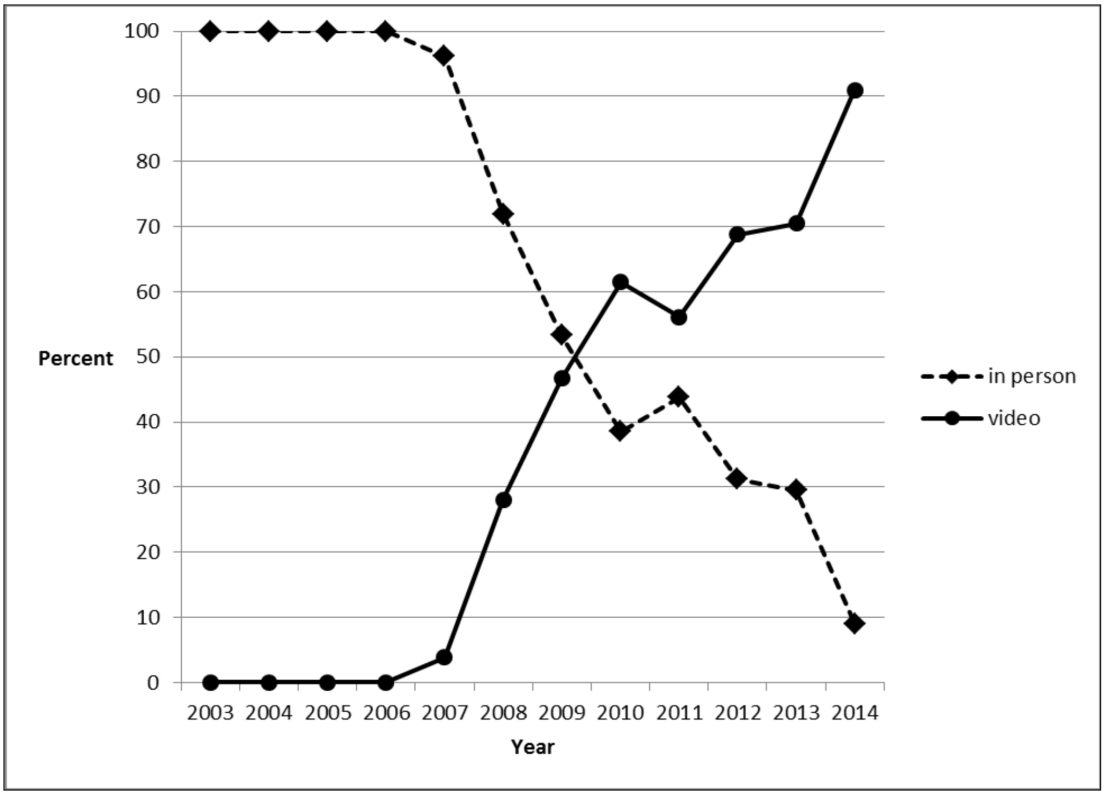

Figure 4: Initial Genetic Counseling Consultation - Interior Health Authority

A similar pattern on Vancouver Island has been demonstrated with on average less than 10% of patients seen by telegenetics in 2004-2006 to about 60% seen by telegenetics in 2013 (Figure 5).

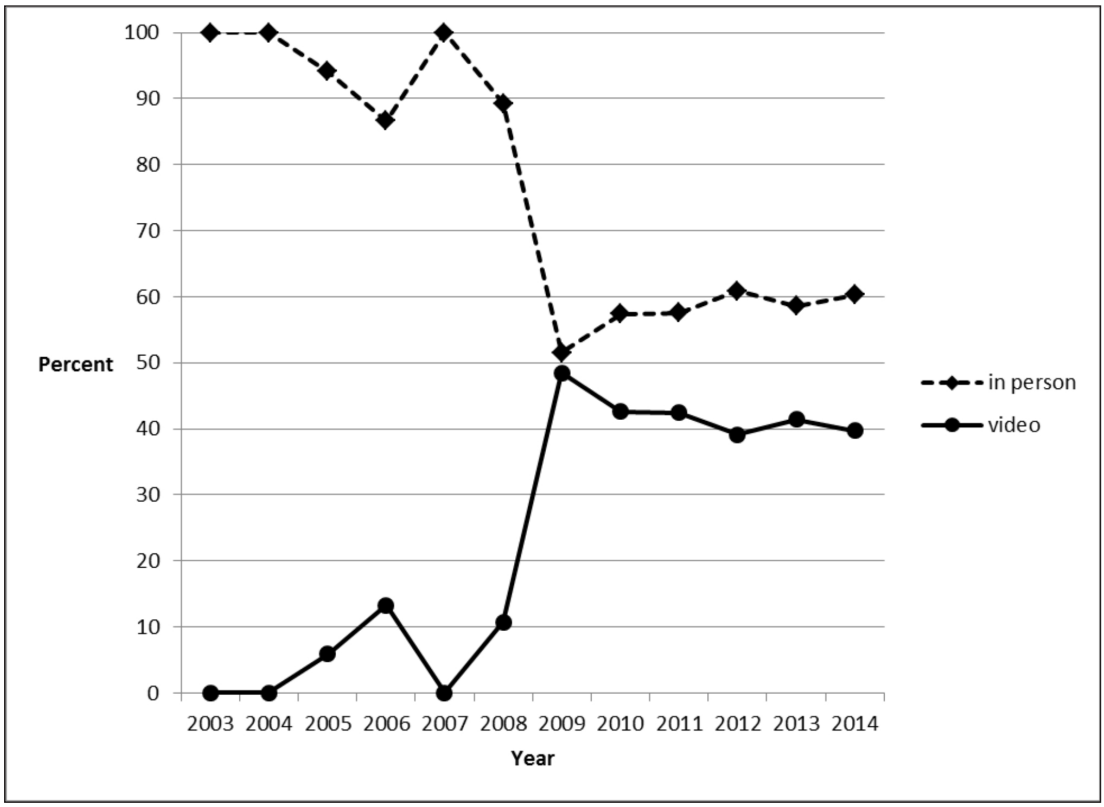

Figure 5: Initial Genetic Counseling Consultation - Vancouver Island Health Authority

This number is expected to increase for this region as the number of outreach clinics decreased dramatically in 2014 from 20 consultations per month with a genetic counselor to 20 consultations per year. Decrease in outreach to the interior and possibly the north is also expected to lead to an increase in telegenetics consultations in both these areas of the province.

In addition to the pilot studies at HCP, telegenetics consultations for hereditary cancer risk assessment have been studied in comparison to in-person genetic counseling. This approach has been demonstrated to be comparable to in-person counseling in areas of patient satisfaction (GRAY *et al*., 2000; GATTAS *et al*., 2001; COELHO *et al*., 2005; ZILLIACUS *et al*., 2010b; ZILLIACUS *et al*., 2011), knowledge gained/ understanding (GRAY *et al*., 2000; COELHO *et al*., 2005; ZILLIACUS *et al*., 2011), and perceived empathy (ZILLIACUS *et al*., 2011), and considered an acceptable and effective method to provide cancer genetic counseling to patients in remote and therefore underserved locations (ZILLIACUS *et al*., 2011; HILGART *et al*., 2012; ELLIOT *et al*., 2012). Although telehealth is comparable to in-person for genetics provider satisfaction, concerns about impact on the role of the genetics specialist in the consultation and decrease in rapport building have been described (GRAY *et al*., 2000; GATTAS *et al*., 2001; D'AGINCOURT-CANNING *et al*., 2008; ZILLIACUS *et al*, 2009; ZILLIACUS *et al*., 2010a).

Telehealth in British Columbia is used in other disciplines and the labour intensiveness of scheduling consultations times has been noted. In the past, limitations included audio/video quality that could compromise consultation accuracy as well as lack of bandwidth required in some rural areas of the province (HO *et al*., 2004). These disadvantages were experienced at HCP and although some of the technological concerns in utilizing telegenetics have improved over time, the laboriousness of booking these consultations has continued. Streamlining the booking process and using technology to simplify the communication steps required with the community hospital could improve the efficiency of employing telegenetics.

Beyond the successful experience in British Columbia, telegenetics has become a common part of clinical practice in hereditary cancer genetic counseling (COHEN *et al*., 2013; ELLIOT, *et al*., 2012) and is now established to be comparable to in-person consultations for both patient satisfaction and understanding (D'AGINCOURT-CANNING *et al*., 2008; COELHO *et al*, 2005; ZILLIACUS *et al*, 2011). The Accreditation Counsel for Genetic Counseling has determined that standards for genetic counseling training programs clinical fieldwork requirements can include telegenetics consultations in 10% of cases (ACGC Standards of Accreditation for Graduate Programs in Genetic Counseling, 2013).

TELEPHONE COUNSELING

Traditional in-person hereditary cancer genetic counseling consultations followed by in-person results disclosure is based on the early Huntington disease genetic counseling model, and had been established as a common practice that met ethical guidelines (WHAM *et al*., 2010; DEWERT, 1998). This model allows for immediate

psychosocial evaluation and support by the health care provider. However, given the population distribution around British Columbia, an alternative model for genetic test results disclosure was explored. Telephone appointments are convenient for patients, and had been requested in patient feedback at HCP (HITCHMAN; RIDGE, 2005). This is also a method of results disclosure for other types of genetic testing as well as medical tests such as karyotype results from amniocentesis and biopsy findings for cancer diagnoses.

A study at the Hereditary Cancer Program in 2004 examined satisfaction of telephone results for women across BC who had consented to BRCA1/2 testing. All patients were satisfied with the results disclosure experience. This is consistent with other studies (BAUMANIS *et al*., 2009; JENKINS *et al*., 2007). Reasons for telephone preference included reduced pre-disclosure anxiety, avoiding need for travel, and convenience. Participants in this study would have had to travel between 0 and 550km to reach the HCP Vancouver site to meet a genetic counselor in-person. Opportunity for follow up in-person or telephone appointments were always offered (HITCHMAN; RIDGE, 2005).

In current practice at HCP, patients are offered in-person or telephone test results consultations and the options are explored with the genetic counselor at the initial consultation. Telegenetics is used in a minority of cases (13 genetic test results consults over 10 year period), but this is not convenient for most patients and can lead to a significant delay in results disclosure due to the current telehealth appointment booking system and limited availability of telehealth rooms in some locations. In 2003, 30% of genetic test results disclosure appointments were in-person. Since 2006, about 10% of appointments are in-person with 90% by telephone (Figure 6).

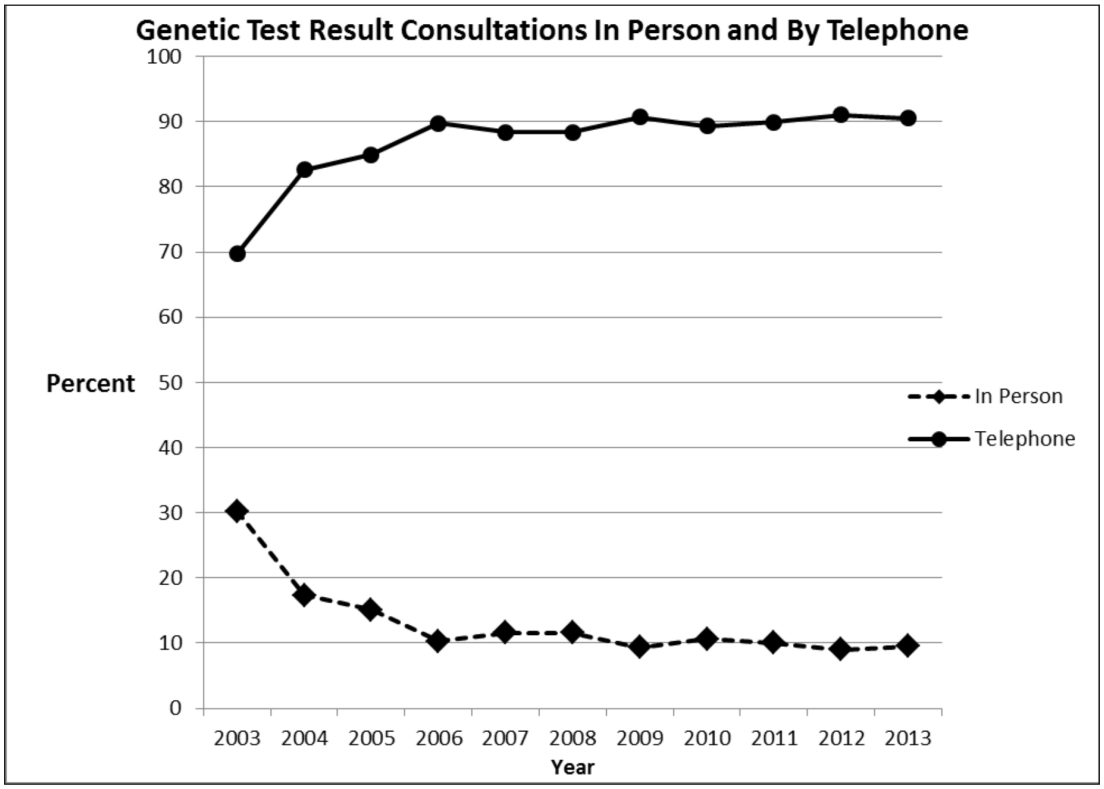

Figure 6: Genetic Test Result Consultations In-person and By Telephone

Telephone counseling is part of HCP current practice, not only in the context of genetic test results disclosure, but also for initial genetic counseling consultations for a select group of patients, including those where mobility is a significant concern, and patients where publicly funded genetic testing has been completed in the family and additional analysis is not available.

Telephone counseling is common practice in that most cancer genetic counselors have provided BRCA1/2 test results by telephone (BAUMANIS *et al*., 2009). Telephone counseling may offer advantages beyond cost savings and convenience. Patient satisfaction has been demonstrated (SCHWARTZ *et al*., 2014; SUTPHEN *et al*, 2010; PLATTEN *et al*., 2012; BAUMANIS *et al*., 2009) without differences in anxiety and general well being (JENKINS *et al*., 2007) in comparison to in-person consultations. Patients may benefit most from the option to choose the setting of their genetic counseling session, as has been shown for patients given choice about test results disclosure (BAUMANIS *et al*., 2009). For those who have strong emotional reactions to medical facilities where they or their family members had cancer care, telephone appointments may be a preferable option (MADLENSKY, 2014). Schwartz *et al*. also demonstrated that telephone counseling with visual aids did not significantly alter BRCA1/2 knowledge using the Breast Cancer Genetic Counseling Knowledge scale (SCHWARTZ *et al*., 2014; BUTRICK *et al*., 2014). This scale does not assess knowledge of index normal or uninformative test results (affected family member tested with no mutation identified) or variants of uncertain clinical significance, however it is comprehensive in other areas of hereditary cancer risk understanding and carrier testing outcomes (ERBLICH *et al*., 2005). More studies may be warranted to assess knowledge of these more complex outcomes on personal and family member cancer risk in the setting of telephone counseling.

Telephone counseling may not suit all patients and populations. A recent random noninferiority trial of telephone counseling for hereditary breast and ovarian cancer found a slightly lower uptake of genetic testing in the cohort who received counseling by telephone, however the reason for this is not clear. The same research group (BUTRICK *et al*., 2014) found that race/ethnicity was significantly associated with lower uptake of testing among participants who were counseled by telephone but did not observe this trend in the in-person cohort.

Billing per patient consult is not in practice at HCP as the BC Cancer Agency is funded by direct provincial government grant. However, this is an issue in many health care systems when considering a model other than a traditional in-person consultation. Schwartz *et al*. recently demonstrated justification for expanded reimbursement of telephone counseling by US based insurers (SCHWARTZ, *et al*., 2014) and described at least one large health insurance provider in the US that reimburses for hereditary cancer telephone genetic counseling.

GROUP COUNSELING

In order to meet the increasing demand for hereditary cancer genetics services, the HCP explored providing genetic counseling to groups of patients to investigate whether this improved efficiency. A pilot study at the HCP in 2005 (RIDGE *et al*., 2009) offered group

genetic counseling to women referred for hereditary breast and ovarian cancer. Patients were receptive to group counseling but 17/42 (40%) actively declined participation. At study completion, 52% had in-person counseling and 36% participated in group counseling. Positive outcomes included participant sharing and support. Negative outcomes included enhanced frustration at not being eligible for publicly funded genetic testing, privacy concerns, group influence on decision-making, and the presence of intrusive co-participants. This study highlighted concerns of adverse group dynamics in a group setting for participants who do not self-select for this option. Group counseling combined with individual sessions has been studied for women at high risk for HBOC (CALZONE *et al*., 2005; ROTHWELL *et al*., 2012) and shown to be both feasible and acceptable. A group only or group followed by individual session model is currently in regular practice in a number of cancer genetics clinics in Ontario and Alberta, Canada, as well as the United States.

FUTURE ROLE OF ALTERNATIVE SERVICE DELIVERY MODELS

The HCP has explored and embraced alternative models of genetic counseling service delivery for most of the program's historical existence. However, there has been a continued increase in referrals including a sudden drastic increase in referrals in 2013 (Figure 7), similar to the increase for the same time period in the UK described by Evans *et al*. in 2014. This pattern is likely related to the opinion-editorial piece by Angelina Jolie describing her BRCA1 gene mutation status and risk reducing surgery (New York Times, May 2013), often described as the Angelina Effect. The HCP as well as cancer genetics centres in Canada and the US (BORZEKOWSKI *et al*., 2014; RAPHAEL *et al*., 2014; DEMARCO *et al*., 2007; M. FERGUSON PERSONAL COMMUNICATION, 2013), are faced with similar challenges and a new paradigm in cancer genetic counseling delivery models may develop (Izzo, 2014).

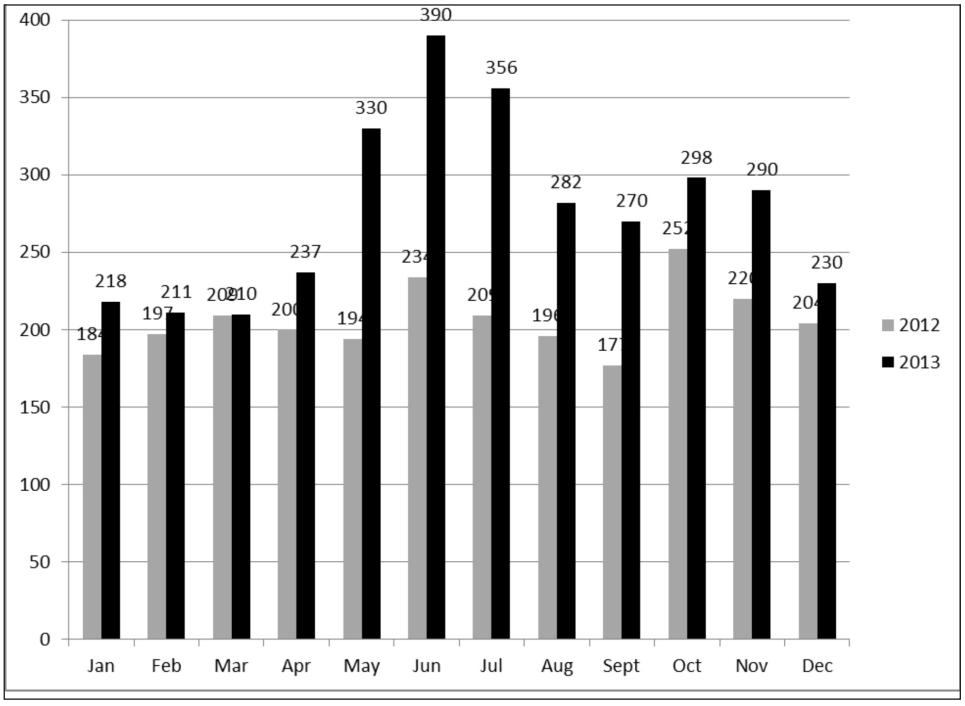

Figure 7: Referrals for Hereditary Cancer Genetic Counseling 2012 vs 2013

In British Columbia, the impact of the Angelina Effect is further exacerbated by the longer term 'historic' HCP patients where almost a generation of time has passed since inception of the program and who require ongoing care; family histories of previous patients are emerging with new cancer diagnoses and require consideration and review of new genetic testing and cancer risk management options. In addition, advances in genetic testing methodology and developments in cancer risk gene identification with related increased awareness of genetic testing may have contributed to an increase in appropriate referrals. There is a pressing need to further explore alternative models of hereditary cancer genetic counseling to improve efficiency. For example, a high risk cancer gene panel has recently been implemented at the BC Cancer Agency as the standard clinical test for most individuals consenting to index genetic testing. It is possible that this new platform will yield incidental pathogenic findings that are not consistent with the family history of malignancies or the current HCP guidelines on genetic testing eligibility. This may increase the number of individuals seeking hereditary cancer counseling and genetic testing. There may in fact be a universal need for new genetic counseling models in response to increasing demand for these services due to expanding applications of genomic technology (INSTITUTE OF MEDICINE, 2009). Healthcare provider position statements and evidence based care guidelines also contribute to the demand for hereditary cancer genetic counseling (TREPANIER AND ALLAIN, 2014). In addition, we expect increase demand for cancer genetic counseling in British Columbia with the expansion of universal Lynch syndrome tumour testing (immunohistochemistry for mismatch repair genes) across the province, as well as a need for genetic counseling for patients having BRCA1/2 testing for chemotherapy treatment planning using PARP inhibitors (FONG *et al*., 2009; DAVAR *et al*., 2012).

Currently the HCP clinic is equipped with a dedicated telehealth unit (camera, large tv screen, document camera) used daily for clinical consultations. A clinician to patient desktop video connection could be considered to expand the number of clinicians providing video consultations. Currently telegenetics appointments in BC and Yukon are only available using the secure provincial telehealth network established in 2008 as part of the Provincial Telehealth Program. However, telehealth utilization is increasingly common across healthcare fields and telegenetics is established as acceptable. The challenges described in earlier studies about technical disadvantages and patient/provider discomfort with technology or the technology as a barrier to establishing rapport and connection with patients may be less prevalent with the increase in video enabled calling platforms such ask Skype and Facetime that are common across cultures and age groups. In fact, home videoconferencing for hereditary cancer risk assessment has shown patients are satisfied with this model (MEROPOL *et al*., 2011). Applying home videoconferencing in BC would increase accessibility for patients y *et al*low genetic counselors to connect from a desktop computer located in the clinical setting. Patient information material could be provided to these patients in advance of the consultation as is current practice for HCP telegenetics appointments. Transmission of live screen shots or video feed could replace a document camera often used in the telegenetics consultation. Alternatively, video platforms designed for physicians to videolink from their home to health care centre in-patients can be adapted for consulta-

tions where patients are at home and genetic counselors/geneticists are at the clinic site (G. Mitchell, personal communication 2014). A limitation of this system may be the availability of the software or specific platform that each patient would need to load or download onto their in-home computer. A web browser based platform that includes scheduled video consults and secure messaging may also be a viable option to expand the HCP telegenetics service.

Telephone counseling services can also be expanded. Currently telephone counseling is mostly used for genetic test results disclosure at HCP. Triage of groups of patients most suited to this mode of service delivery can be implemented using guidelines such as those with less complex indications and current health status as well as referral indication to differentiate between simple and complex counseling scenarios. The possibility that patients needing urgent testing would benefit from telephone counseling because of shorter wait time has been suggested (MADLENSKY, 2014) and wait time for consultation is a relevant factor in triage. Visual aids to demonstrate relevant concepts are a common tool for genetic counselors in all disciplines of medical genetics and their use is ubiquitous for in-person consultations. These tools can also be effective for alternative modes of delivery (SCHWARTZ *et al*., 2014). A strategy is in development to expand telephone counseling at HCP to a more complete genetic counseling service with use of visual aids. These documents will be sent by mail and/or available electronically for use during the consultation, in order to improve congruency with the in-person genetic counseling experience, and assist patients to understand complex concepts in the absence of visual cues (MADLENSKY, 2014). Careful consideration will be required to determine the contributing factors in clarifying the most appropriate patient population for this model of service delivery. Periodic review of these consultations may help to optimize triage to telephone or in-person/telegenetics consultations.

Although group counseling is not part of current practice at the HCP, this could be explored again in the future for specific populations of patients such as patients meeting simple common referral criteria (for example women diagnosed with breast cancer at very young ages with no family history of breast or ovarian cancer).

TELEHEALTH AND CANCER GENETICS IN BRAZIL - THE BC EXPERIENCE AS EXAMPLE

There are a number of notable differences between the healthcare settings in Canada and Brazil. Although both countries have a large land area of 8-9 million km squared, the population of Brazil is over 5 fold that in Canada. The mean annual per capital income in Canada is 3 fold that seen in Brazil and health spending per capita in Canada is at least 3-5 fold that in Brazil (www.statcan.gc.ca; http://www.who.int/countries/bra/en/). Both countries have a socialized health insurance system. Canada uses a publicly funded and administered health care system with some allowance for private pay services. Brazil employs universal public health care (SUS) funded on taxation and social contributions with variable access among regions for different types of services and private funding is also an essential component of national health spending (HOROVITZ *et al*., 2013).

In Canada, medical genetics is a 5 year clinical residency program and clinical medical geneticists are affiliated with the Canadian College of Medical Geneticists (CCMG). The CCMG also includes laboratory scientists admitted after completing certification in areas molecular genetics and cytogenetics. There are 58 clinical geneticists serving all provinces and territories and 10 laboratory geneticists affiliated with CCMG. Genetic counseling is provided by clinical geneticists and genetic counselors. Genetic testing is performed in local CLIA approved laboratories or sent to out of country CLIA approved laboratories for certain tests.

A review of genetics services in Brazil by Horovitz *et al*. in 2013 describes The National Familial Cancer Network which includes multiple genetics centres across the country and is coordinated by the NCI Division of Genetics. These centres provide both genetic counseling and laboratory support for genetic testing. However, access to publicly funded cancer genetics services may not be available to a significant number of individuals at high risk possibly due to insufficient training of health care professionals, difficult access to genetic testing, and a resistance to seeking these services.

Horovitz *et al*. also describes that few medical schools in Brazil include practical training in genetics and that most physicians do not recognize the genetic basis of some diseases, have limited knowledge on how to refer to genetics services, or do not value the process of genetic counseling.

However, graduate training programs in genetics in Brazil produce a diverse range of medical genetics research and the Brazilian Society of Medical Genetics (SBGM) offers board certification in human molecular and cytogenetics as well as areas of clinical medical genetics. There are 11 residency training programs in Brazil, all established in the South and South-East region as well as one in Brasilia-Federal District. Specialist laboratory services are also located in the Centre-South. Professional accreditation in medical genetics is awarded to specialists in other areas of medical practice with approval from the SBGM and the Brazilian Medical Association. Over 200 medical genetics board certifications have been awarded in Brazil since 1981 via one of these pathways (HOROVITZ *et al*., 2013).

Genetic counseling and genetic testing services including cancer genetics services have been slow to be integrated into the SUS. Most hereditary cancer risk assessment is provided in academic centres and genetic testing performed in academic research laboratories with grant funding (PASSMAN, INCA ASCO, 2012).

British Columbia serves as a comparable model for cancer genetics services provided in regions with centralized cancer care, serving a population over a large area with both urban and rural/remote areas. BC has a well established telehealth network with locations 200 locations across the province and 470 'videoconference endpoints'.

Brazil has a national Telehealth Primary Care Program established in 2006 coordinated by the Ministry of Health in support of primary care with a focus on family health teams. A report in 2009 showed teleconsultations in 9 states and 900 municipalities. In Goias, 84 municipalities participated including one telehealth centre that completed a tele-ophthalmology project using retinography for appropriate ophthalmology triage (CAMPOS *et al*., 2009). A second phase of the National Telehealth Project in 2009 saw further expansion to all states (www.telessaudebrasil.org.br). The Telemedicine

University Network (RUTE) connects many university hospitals across all states and aims to address indigenous health care services. RUTE had two locations in Goias (Hospital Universitario da UnB and Hospital das Clinicas da UFG). RUTE had 33 special interest groups in 2010, including oncology and gynecology, and has a mandate to implement telehealth procedures including education, second opinion, and teleconsultations into rural and underserved regions (www.rute.rnp.br).

Geographic barriers to hereditary cancer genetic counseling in some regions of Brazil could be addressed with alternative models of delivery. Telegenetics and telephone counseling with the use of visual aids could be implemented if access through the established telehealth networks is available. Multidisciplinary collaboration with programs such as oncology and gynecology already engaged with telehealth could increase awareness of the value of cancer genetics services and access to telehealth for professionals providing these services. This collaboration may also increase the acceptability of telehealth for the genetics service provider, as it has been shown that training and experience using telehealth reduces healthcare providers' resistance and reluctance to use telehealth technology (HILGART *et al*., 2012). Access to existing telehealth networks for hereditary cancer risk assessment may be a financial benefit to service providers and patients; in BC the existence of a telehealth network with necessary equipment to provide hereditary cancer services costs the healthcare system less than providing outreach clinics to rural areas (D'AGINCOURT-CANNING *et al*., 2008). In the scenario that telehealth networks are not available to the genetics service providers in Brazil, exploring the use of connectivity tools available may be successful. For example, personal electronic devices and person to person video calling technology may be a platform that both health care professionals and patients are comfortable and familiar with. Visual aids provided by mail, email, or a web based resource could support the video call to approximate an in-person consultation.

Submission of samples for genetic testing from remote locations could be arranged by courier or mail using peripheral blood samples, cheek swab collection kits, or dried blood spot collection cards where samples are not temperature or time sensitive compared to other collection methods.

The paucity of genetics specialists in Brazil is a limiting factor in providing services to those at increased risk for hereditary cancer as access to medical genetics services is mainly at tertiary centres in the south and southeast regions (ACOSTA *et al*., 2013). Non-accredited genetic counseling training programs in Brazil are available for health care professionals including nurses and psychologists (ACOSTA *et al*., 2013). Increasing the presence of, or access to cancer genetic counselors, particularly in underserved areas of the country could expand access to genetic counseling and facilitate genetic testing. Currently patients may meet with a health professional with background in nursing or psychology before a consultation with a medical geneticist, to provide information on investigations that may be part of the genetics assessment (ACOSTA *et al*., 2013). These are roles that could be provided by a genetic counselor or health professional with training in specific aspects of genetic counseling. Certified genetic counselors in North America, Europe, and Australia provide hereditary cancer risk assessment to patients as part of a team with a medical geneticist, or in

many settings, cases are reviewed in advance or after the consultation with a physician if necessary. Establishing a formally recognized genetic counseling profession in Brazil with training and certification in the necessary core competencies and drafting guidelines for necessary elements for hereditary cancer genetic counseling and testing such as those determined by the NSGC (RILEY *et al*., 2012) could increase the cancer genetics services being provided across the country and improve access for this area of growing demand at the established tertiary centres as well as underserved areas of the country. The increase in patients receiving genetic counseling with the development of an established genetic counseling profession may be a more financially sustainable model for medical geneticists than current practices.

In addition to incorporating genetic counselors into the service delivery plan, collaborative models with nurses, family health professionals and community health agents could be employed as part of a multidisciplinary team. These allied health professionals could complete training in specific aspects of genetic counseling such as psychosocial support and facilitating informed consent to allow medical geneticists to consult an increased number of patients across a larger geographic area. The medical and co-facilitation models used in Australia described by Zilliacus *et al*. in 2009 with a genetic counselor and patient in the patient's community connecting via telehealth to medical geneticist at a tertiary centre could be adapted to the Brazilian healthcare setting to increase access to cancer genetics services.

Expanding access to genetic counseling and hereditary cancer risk assessment warrants consideration of additional essential elements in complete patient care, including access to appropriate genetic testing and cancer risk management options. This requires broad availability of screening such as mammography, breast MRI, colonoscopy, and other imaging depending on the hereditary cancer syndrome, as well as access to risk reducing surgery when indicated. The value of cancer risk stratification when economic resources are limited reinforces the merit of genetic counseling in both publicly and privately funded healthcare systems.

In summary, the BC HCP has evolved over more than 10 years to provide a comprehensive specialized cancer genetics service to patients and families in BC and Yukon. Alternative models of delivery including telegenetics, telephone counseling, and group counseling have been explored and in some cases implemented to become a routine part of the cancer genetic counseling practice. There are increasing demands for hereditary cancer risk assessment and the alternative models currently in use in BC require both expansion and enhancement in order to meet the ongoing growing genetics services needs.

BC's experience in providing genetic counseling using telehealth and telephone counseling and the medical literature on alternative models of cancer genetics service delivery can provide a framework to explore expansion of hereditary cancer genetic counseling in Brazil.

Abstract: *British Columbia is the western-most province of Canada and includes a varied landscape of Pacific coastal and northern regions with population estimate of 4.6 million. Provision of genetic counseling and genetic testing for high risk populations contributes to stratifying cancer risk, reducing cancer-related morbidity and mortality, as well as decreasing health care costs through earlier detection and prevention of malignancies.*

Keywords: *Hereditary. Genetic Counselling. Cancer*

Nota

1 <http://www.health.gov.bc.ca/ehealth/Telehealth_project.html>.

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