Identifying opportunities for collaboration and growth of genetic counseling services in the Asia Region

Mercy Y. Laurino^{1,2} · Darci L. Stern³ · Jennifer K. Thompson⁴ · Kathleen A. Leppig^{5,6}

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Abstract The Genetic Counseling Pre-Conference Workshop (GCPCW) was held on September 16, 2015, in Hanoi, Vietnam. We report the GCPCW outcomes obtained from pre- and post-conference questionnaires, case-review breakout session, and an open discussion of needs for genetic counseling services in the Asia region. The GCPCW participants completed questionnaires with closed- and open-ended questions regarding the status and needs of providing genetic counseling services in Asia. Utilizing thematic content analysis, common themes shared during the case-review breakout session are summarized and survey results are tabulated. Of the 71 participants, pre- and post-conference questionnaires were returned by 57 (80%) and 44 (62%) individuals, respectively. Of the 42 participants who did not identify themselves as students in training, 36 (86%) stated they are currently providing genetic counseling services. Participants cited that the most useful information obtained during the GCPCW related to the status of genetic counseling services in the region, discovery of shared challenges, professional networking, and the need to establish genetic counseling training programs and

its accreditation. The GCPCW provided a collaborative forum to address current challenges and needs of genetic counseling services in the region. Strategies were identified to foster genetic counseling training and clinical service opportunities.

Keywords Genetic counseling · Asia · Genetic counselor · Pre-conference workshop · Education report

Introduction

While advances in medical genetics and genomic technology have increased the global need for genetic counseling services, there is paucity of individuals trained in medical genetics and genetic counseling who can provide such services (Chien et al. 2013; Christianson and Modell 2004; Kingsmore et al. 2012; Laurino and Padilla 2013; Lee and Thong 2013; Maltese et al. 2017). Specifically in Asia, providing genetic counseling services is daunting given that this region represents approximately 35% of the world's land mass and an estimated 60% of the world population with countless ethnic minorities (Kumar 2012). Without question, establishing these services requires a commitment from local resources and global participation from health-care providers with expertise in medical genetics and genetic counseling, along with professional societies with well-developed resources. This partnership opportunity is in alignment with the United Nations' Millennium Project Task Force on Science, Technology and Innovation, which promotes global economic development while enhancing knowledge base in developing countries (UN 2011). In addition, the 2011 evidence-based consultation report from the World Health Organization on community genetics in low- and middle-income countries specifically noted the importance of "training health professionals in basic concepts of genetics and their application to

✉ Mercy Y. Laurino
mlaurino@seattlecca.org

¹ Department of Pediatrics, College of Medicine, University of the Philippines Manila, Manila, Philippines

² Seattle Cancer Care Alliance, 825 Eastlake Ave East K-231, Seattle, WA 98109, USA

³ Seattle Children's Hospital, Seattle, WA, USA

⁴ Hereditary Cancer Program, BC Cancer Agency, Vancouver, British Columbia, Canada

⁵ Group Health Cooperative, Seattle, WA, USA

⁶ University of Washington, Seattle, WA, USA

community genetics services” when determining recommendations for the most efficient interventions (WHO 2011).

Medical genetics professionals in low- and middle-income countries are requesting global partnerships specifically, but not limited to, on how to best establish clinical medical genetics and genetic counseling services, guidance on cost-effective ways in utilizing diagnostic genetic technologies, and how to strategically establish a genetic counseling training program. To address such requests, the Global Health Genetics Partnership Group (GH-GPG) organized the Genetic Counseling Pre-Conference Workshop (GCPCW) in Hanoi, Vietnam, held on September 16, 2015 (APSHG 2006), in partnership with the newly established Professional Society of Genetic Counselors in Asia (www.psgca.org). The GCPCW was organized in conjunction with the biennial Asia-Pacific Conference on Human Genetics (APCHG). The GCPCW’s intention was to bring together genetic counselors, genetic counseling students, clinical geneticists, and allied health professionals interested in the field of genetic counseling in the Asia region. The four main objectives for the GCPCW were the following: (1) evaluate strategies for genetic counselor partnership in the Asia region; (2) determine a feasible infrastructure to support implementation of genetic counseling training programs and enhance training in existing programs; (3) recognize the provision of genetic counseling in the context of the varying cultural, religious, and health systems in Asian countries; and (4) define critical elements for genetic counseling resource tools.

Background

The GH-GPG was formed in 2009 and has consisted of medical geneticists, cytogeneticists, genetic counselors, interested researchers, and members from industry. Core members of the GH-GPG have extensive experience working in Asia as volunteers (D.L.S., J.K.T., and K.A.L.) and as a grant recipient of the Philippine Department of Science and Technology (M.Y.L.). Equipped with our members’ personal experiences, the GH-GPG combined resources to determine what did and did not work to promote the development of medical genetics and genetic counseling services in Southeast Asia. Early collaborations with medical geneticists Dr. Dung Chi Vu at the National Hospital of Pediatrics and Dr. Nguyen Viet Nhan at Hue College of Medicine and Pharmacy included grant writing, medical school teaching, clinic consultation, and laboratory training (personal correspondence). They requested collaboration to support the development and eventual implementation of sustainable genetic counseling training programs, and to enhance existing clinical genetic services. This led to the first Vietnamese-North American Genetics Meeting, held in Hanoi, Vietnam, on March 25–26, 2013 (Leppig et al. 2013). The goals of this first conference were to bring together genetic providers from across Vietnam to discuss the scope of

current genetics care provided in the country, discuss examples of new technologies that may allow more cost-effective genetic testing, highlight genetic counseling as part of health-care delivery services, and facilitate collaboration and mentorship with international colleagues. The partnerships and networking from this meeting, as well as the successful implementation of the genetic counseling training program in the Philippines, led to the culmination of the PSGCA, which was officially launched at the GCPCW.

Materials and methods

Over 90 participants attended the GCPCW, with a total of 71 registered participants representing Australia, Canada, Hong Kong, India, Indonesia, Malaysia, Philippines, Singapore, Taiwan, Thailand, USA, and Vietnam. Participant demographic information was requested in a pre-conference questionnaire and included type and level of training, whether and in what circumstances they were providing genetic counseling in their current position, and what they perceived as the major barriers regarding the development of a genetic counseling program in their country. Participants could choose more than one answer for a specific question, as appropriate. Survey results from the closed-ended questions were calculated, including frequencies, and tabulated. In addition, anonymous feedback was provided by the participants through a post-conference questionnaire regarding the utility of the conference and suggestions for future conferences.

The GCPCW was organized into two sessions. In the morning session (session I), invited speakers reported on the status of genetic counseling in their respective countries (publication *in review*). The afternoon session (session II) was comprised of a case-review breakout session, followed by an open forum to discuss the challenges and future directions for those currently providing genetic counseling in the Asia region. Participants were organized into case-review breakout groups purposely arranged to have members from multiple countries represented in each group. Using a case-based approach, each group outlined country-specific genetic counseling best practices for a particular clinical case scenario and discussed critical elements necessary to have a common genetic counseling resource related to the condition associated with their case scenario. To enumerate, the clinical case scenarios were the following: (1) a 2-year-old child with trisomy 21 (Down syndrome) based on clinical findings, (2) a 25-year-old woman with breast cancer, (3) two brothers with intellectual disability/mental retardation and concern for X-linked intellectual disability, (4) a 32-week gestation pregnancy with hydrops and suspected thalassemia, (5) a newborn child with ambiguous genitalia and an abnormal newborn screening result positive for congenital adrenal hyperplasia, and (6) a newborn child with an open sacral neural tube defect. To conclude

the case-review breakout session, one representative from each group presented a summary of their group's discussion using a template provided to guide the synthesis of key discussion points (i.e., What countries are the people in your discussion group from? What do you think are the most significant barrier(s) to providing genetic counseling in this scenario? What are the strongest similarities between genetics services in the countries in your group? What are the strongest differences between genetics services in the countries in your group? What resources/tools would be of most value to provide genetic counseling?). Using the written summaries presented by the six group representatives, thematic content analysis was conducted to identify common themes.

Results

Of the 71 participants, pre- and post-conference questionnaires were returned by 57 (80%) and 44 (62%) individuals, respectively. Registered participant demographics are presented in Table 1 and show the spectrum of training of individuals providing genetic counseling in Asia with the majority of individuals having post-graduate training or health-care training. Participant opinions about genetic counseling training are summarized in Table 2. Participants were asked to state in the post-conference questionnaire the most valuable information they learned during the GCPCW. The most common responses were as follows: (1) availability and status of genetic counseling in the Asia region, (2) different approaches to providing genetic counseling, (3) shared challenges, (4) professional networking/meeting friends, (5) opportunities for collaboration, and (6) need to promote awareness of the role of genetic counseling and develop accreditation.

The 44 participants who completed the post-conference questionnaire also shared that their expectations for the conference were met and the majority shared they would attend another genetic counseling workshop at a future APCHG meeting. Participants listed the following topics of interest for future conferences: best practices in genetic counseling, east and west genetic counseling differences, effective genetic counseling communication skills, cross-country training opportunities, and research studies pertinent to genetic counseling.

Case-review breakout session summary

Five main themes emerged during the group discussion of the clinical case scenarios, and these were the following:

1. The need for best practice guidelines
2. Unequal access to genetic counseling services
3. Method for obtaining accurate family history
4. Creating culturally relevant resources
5. Availability of patient support groups

Table 1 Participant demographics and pre-conference survey responses

Categories	<i>n</i> (%)
Gender (<i>n</i> = 57)	
Male	11 (19.3)
Female	46 (80.7)
Areas of genetics (<i>n</i> = 57)	
Clinic	25 (43.9) ^a
Laboratory	21 (36.8) ^b
Teaching	22 (38.6) ^c
Research	23 (43.4) ^d
Student/trainee	15 (26.3)
Years of practice (<i>n</i> = 42; students were excluded)	
0–5 years	14 (33.3)
6–10 years	14 (33.3)
11–15 years	9 (21.4)
16+ years	5 (11.9)
Currently provide clinical genetic counseling (<i>n</i> = 42; students were excluded)	
Yes	36 (85.7)
No	6 (14.3)
Education level (<i>n</i> = 42; students were excluded)	
Undergraduate/college degree	1 (2.4)
Masters degree in genetic counseling	10 (23.8)
Masters degree in (other)	6 (14.3) ^e
Nurse	2 (4.8)
PhD	8 (19.1)
MD	6 (14.3) ^f
MD with fellowship in medical genetics	7 (16.7)
Other: _____	2 (4.8) ^g

^a Two noted clinical only

^b Seven noted lab only

^c Four noted teaching only

^d Two noted research only

^e Two genetics; one biology; one med biotech; two undeclared

^f One with PhD

^g Two with graduate diploma in genetic counseling

The need for best practice guidelines There were many differences expressed in terms of the resources available within a country and barriers to providing the best patient care. While there was overall consensus about the ideal clinical care and management for each case scenario, the development and publication of best practice guidelines specific to the Asia region have been addressed as one of the most important action items from the GCPCW. Participants identified that the care of individuals with

Table 2 Participant opinions about genetic counseling training

Categories	<i>n</i> (%)
Training program availability	
Genetic counseling	40 (72.7)
Medical genetics	31 (56.4)
Other: _____	
No training programs	8 (14.5)
I do not know	3 (5.5)
For those who answered No, opinion whether there is importance of having a genetic counseling training program	
Y/N	Yes: 8 (100)
Ideal professionals to receive training in genetic counseling (circle all that apply) (<i>n</i> = 57)	
Undergraduate	14 (24.6)
Nurse	39 (68.4)
Midwife	20 (35.1)
Masters in _____	16 (28.1)
PhD	19 (33.3)
MD	35 (61.4)
Other: _____	7 (12.3)
Major barriers in genetic counseling training program development (circle all that apply) (<i>n</i> = 57)	
Financial limitations	31 (54.4)
Commitment from key leaders within the health-care system	35 (61.4)
Commitment from an education institution	25 (43.9)
Need of trained educators	39 (68.4)
Other: _____	4 (7.0) ^a

^a Government support for job provision; lack of awareness, support, and recognition of genetic counseling

genetic conditions often requires coordination between multiple specialists due to the complex nature of these conditions. However, participants noted that it would be difficult to establish best practice guidelines for the provision of prenatal genetic counseling for the Asia region given the vast differences between countries (and different cultural groups within a given country) in the access to and attitudes towards termination of pregnancy (TOP). Further, when considering students traveling to other countries to receive genetic counseling training, one participant noted: “there will be a need to consider that students in countries where TOP is not an option may experience barriers in developing their counseling skills related to prenatal diagnosis/decision making.” Consensus was that best practice guidelines are therefore needed to include guidance for collaboration and communication by genetic counselors with other health professionals.

Unequal access to genetic counseling services The majority of participants expressed that there is currently unequal access to genetic counseling services across their country, and one group shared “the genetic counselor is usually practicing just by him or herself or there are no GCs.” The common barriers impacting access to genetic counseling services, including the provision of services through both public and private institutions, were identified to be:

- Inconsistent follow-up plans
- Time and funding for travel for patients and/or providers
- Access to an interpreter and language-specific printed resources
- A lack of awareness of genetic counseling consultations as an option for families, which may result in a delay or absence of appropriate referrals

Participants discussed disparities between services that could be offered to individuals in each country given legal and cultural restrictions, particularly around the issue of TOP.

Method for obtaining accurate family history Participants identified that obtaining an accurate family history is essential to provide effective genetic counseling, but that a variety of limitations can restrict this process. This highlighted the opportunity for registries that document family clinical history to serve as a potential resource for more accurate pedigrees when medical record systems are not robust or there are limitations to family communication. Varying cultural beliefs and taboos were identified to play a large role in the elicitation of an accurate family history and these barriers need to be considered and factored into risk assessment.

Creating culturally relevant resources For each case scenario, participants highlighted that genetic counselors rely on culturally relevant resources. These resources include diagnostic and risk assessment tools, such as anthropometric measurements, dysmorphology databases, and population data; counseling aids, such as shared decision-making tools, figures, and flip charts; patient handouts; and increasingly, the use of iPad/iPhone applications. Each of the individual countries represented had identified the need for culturally appropriate resources to serve patients from various ethnic minorities. As one group shared, a pressing need is a “risk assessment tool for Asian specific population data, e.g., VUS [variants of uncertain significance], BRCA risks, etc.”

Availability of patient support groups There was general consensus about the importance of culturally appropriate disease-specific and more broad-reaching peer and parent support groups for rare and undiagnosed genetic conditions, and varying availability and accessibility of these in each country represented. It was apparent during the group discussion that

most of the participants were not fully aware of existing patient support groups and the best way to connect their patients to such resources.

Discussion

The GCPCW provided important insight into the current status and provision of genetic counseling in the Asia region, and the delineation of the resources needed to further develop these services. While the need for trained genetic counselors in Asia has been recognized, developing the needed resources and effectively training the genetic counseling workforce are indeed a challenge. Nonetheless, the GCPCW provided a forum for those who currently provide genetic counseling services in the Asia region to meet in person, as a collective and cooperative group, and give voice to the professional demand and future needs of genetic counseling services.

Opportunities and future directions

Specifically highlighting the GCPCW's four main objectives, the following opportunities and future directions emerged:

1. *Evaluate strategies for genetic counselor partnership in the Asia region:* A major outcome of the group discussion was the decision to form PSGCA subcommittees. With the intention of having a representative from each country in the Asia region, representatives participate in the respective subcommittee to ensure adequate representation and foster ongoing partnership. As one of its goals, PSGCA will outline guiding principles and facilitate the development of best practice guidelines for genetic counselors practicing in the region. There was a request to create a "Find a Genetic Counselor" feature within the PSGCA website in order to increase accessibility of health professionals providing genetic counseling services across the region. Other partnership opportunities include publication of a genetic counseling newsletter for the Asia region to disseminate information and resources and promote research and collaboration and the publication of articles with cross-nation joint authorship.
2. *Determine a feasible infrastructure to support implementation of genetic counseling training programs and enhance training in existing programs:* Specific needs identified for training genetic counselors include cross-nation training programs and the development of academic exchange and mentorship programs. The establishment of the Philippines' genetic counseling program, which involved trained professionals from both the Philippines and the USA (Laurino and Padilla 2013), can serve as a model on how to develop and implement such training in a low- to middle-income country. Other countries in Asia have existing programs and some local universities are collaborating with genetic counseling training programs abroad. Videoconferencing has been successfully utilized for case review and education between the genetic counseling programs at the University of the Philippines and Stanford University in the USA and would be a model to develop other virtual case review and online training programs. Utilizing existing resources and the commitment for collaboration from trained professionals can hasten the progression of genetic counseling training programs in the Asia region. It is important to note that financial resources and commitment from dedicated individuals to establish genetic counseling training programs are necessary. As such, grassroots organizations such as the GH-GPG and programs such as the GCPCW can be part of the process of identifying models to address genetic counseling training and service needs in Asia and in other regions of the world.
3. *Recognize the provision of genetic counseling in the context of the varying cultural, religious, and health systems in Asian countries:* While it is essential to have standardization in curriculum and eventually genetic counseling training program accreditation across the Asia region, it is important to gain cultural competency in acknowledging the various cultural and spiritual perceptions on genetic contribution towards disease development (Abad et al. 2014; Barlow-Stewart et al. 2006; Cura 2015; Yoon et al. 2011). As an example of divergence in cultural acceptance that is reflected in genetic counselors' training is in regard to the availability of legal pregnancy termination. In some countries, a genetic counselor may be faced with individual legal ramification if pregnancy termination is discussed with a patient (unless intended to save the mother's life); while in other countries, termination of pregnancy is legal and can be an included option during genetic counseling consultations (UN 2013). Although it would be ideal to have a global standardization for genetic counseling training resources, national legislation must be respected to ensure the establishment and ongoing support of genetic counseling training and services programs.
4. *Define critical elements for genetic counseling resource tools:* Zayts and colleagues had emphasized the need to incorporate communication-oriented research findings in genetic counselor training because of the diverse official language and local dialects in the Asia region (Zayts et al. 2013). As such, creating an open resource portal, possibly via the PSGCA website, to share resources with open access for members is a great opportunity. This platform provides a format that allows for easy modification of resources and creating of country/culture-specific resources. Examples of resources

include disease-specific information/fact sheets and websites and should include information regarding support groups and clinical trials when available. Leveraging already available resources is essential to accelerate the development of genetic counseling resources, and reducing its associated costs. Specifically, the Greenwood Genetics Center has adapted their widely utilized Genetic Counseling Aids for Spanish- and Mandarin-speaking populations and is an example of a resource that could be further adapted and utilized (Greenwood Genetics Center 2013).

Challenges

It is necessary to take into account the impact of resource allocation when establishing genetic counseling training programs, as well as clinical and research services in the Asia region. Given the variability of available genetic counseling services, the development of sustainable and comprehensive programs to disseminate accurate information in order for patients and their family members make informed choices is essential. This disparity is evident when only the minority of an affected population has access to such resources. Thus, it is important to provide an in-depth understanding of the existing gap between the currently available genetic counseling resources in Asia, its ongoing demands, and future needs.

Conclusion

The GCPCW facilitated a much-needed networking opportunity amongst the participants on many levels. Partnerships were fostered and opportunities were identified to support committed local health professionals to establish and incorporate genetic counseling services into their expanding and developing private and public health-care infrastructure. Plans are in place to carry this work forward in a second GCPCW at the 12th Asia-Pacific Conference on Human Genetics in 2017.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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