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ORIGINAL RESEARCH

Genetic Counseling/Consultation in South-East Asia: A Report from the Workshop at the 10th Asia Pacific Conference on Human Genetics

Olga Zayts · Srikant Sarangi · Meow-Keong Thong · Brian Hon-yin Chung · Ivan Fao-man Lo · Anita Sik-yau Kan · Juliana Mei-Har Lee · Carmencita David Padilla · Eva Maria Cutiongco-de la Paz · Sultana M. H. Faradz · Pornswan Wasant

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Abstract This paper reports on the workshop 'Genetic Counseling/Consultations in South-East Asia' at the 10th Asia Pacific Conference on Human Genetics in Kuala Lumpur, Malaysia, in December 2012. The workshop brought together professionals and language/communication scholars from South-East Asia, and the UK. The workshop aimed at addressing culture- and context-specific genetic counseling/consultation practices in South-East Asia. As a way of contextualizing genetic counseling/consultation in South-East Asia, we first offer an overview of communication-oriented research generally, drawing attention to consultation and counseling as part of a communicative continuum with distinctive interactional features. We then provide examples of genetic counseling/consultation research in Hong Kong. As other countries in South-East Asia have not yet embarked on communication-oriented empirical research, we report on the current practices of genetic counseling/consultation in these countries in order to identify similarities and differences as well as key obstacles that could be addressed through future research. Three issues emerged as 'problematic': language, religion and culture. We suggest that communication-oriented research can provide a starting point for evidence-based reflections on how to incorporate a counseling mentality in genetic consultation. To conclude, we discuss the need for creating a platform for targeted training of genetic counselors based on communication-oriented research findings.

O. Zayts (✉)
School of English, The University of Hong Kong, Faculty of Arts, Centennial Campus, the University of Hong Kong, Room 738, 7/F, Run Run Shaw Tower (Building B, Arts) Pokfulam Road, Hong Kong SAR, China
e-mail: zayts@hkucc.hku.hk

S. Sarangi
Health Communication Research Centre, School of English, Communication and Philosophy, Cardiff University, Cardiff, UK

M.-K. Thong
Department of Pediatrics, Faculty of Medicine, University of Malaya, 50603 Kuala Lumpur, Malaysia

B. H.-y. Chung
Department of Pediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR, China

I. F.-m. Lo
Clinical Genetics Services, Department of Health, Hong Kong SAR, China

A. S.-y. Kan
Department of Obstetrics and Gynecology, Queen Mary Hostpial, The University of Hong Kong, Hong Kong SAR, China

J. M.-H. Lee
Department of Medical Social Work, University of Malaya Medical Centre, Kuala Lumpur, Malaysia

C. D. Padilla
Department of Pediatrics, National Institutes of Health, University of the Philippines Manila; University of the Philippines College of Medicine, Manila, Philippines

E. M. Cutiongco-de la Paz
Institute of Human Genetics, National Institutes of Health, University of the Philippines Manila, Manila, Philippines

S. M. H. Faradz
Faculty of Medicine, Diponegoro University, Semarang, Indonesia

P. Wasant
Department of Pediatrics, Faculty of Medicine, Siriraj Hospital, Mahidol University, Bangkok, Thailand

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Introduction

This paper originates from the invited workshop 'Genetic Counselling/Consultations in South-East Asia' as part of the 10th Asia Pacific Conference on Human Genetics in Kuala Lumpur, Malaysia, in December 2012. The workshop brought together leading geneticists, genetic counselors and language/communication scholars from five countries in the South-East Asia (hereafter, SEA) – Hong Kong, Malaysia, the Philippines, Indonesia and Thailand – as well as the UK. While in many Western countries[1] genetic counseling has been established as a profession in the last thirty years or so, in SEA this has been a rather recent undertaking. With the development of the profession in SEA, the issue of which theories and practices have been established in the Western countries are applicable to counseling in other sociocultural contexts has become particularly prominent.

The main aim of the workshop was to reflect on existing culture- and context-specific practices surrounding genetic counseling/consultation in SEA vis-à-vis what is known from communication-oriented genetic counseling research in the Western countries. More specifically, the participants of the workshop were asked to address the following questions in their presentations:

1. What is the current practice of genetic counseling in your country?

   a. Who provides genetic counseling services (i.e. trained genetic counselors; medical professionals, etc.)?
   b. What types of clients/genetic conditions receive genetic counseling services?

2. How do cultural and other contextual factors (including language) influence genetic counseling practice?

3. What are the main challenges facing genetic counselors in SEA that should be addressed via future collaborative research?

In what follows, we first outline the communicative dimensions of genetic counseling in general terms, drawing particular attention to consultation and counseling as part of a continuum. We then report on genetic counseling/consultation research currently being undertaken in Hong Kong. We proceed with the summary of genetic counseling practices/services in five countries in SEA – Hong Kong, Malaysia, the Philippines, Indonesia and Thailand – and discuss the key obstacles to genetic counseling in SEA that were identified by the participants of the workshop. In conclusion, we suggest how communication-oriented research can play a crucial role in addressing these obstacles and in intensive targeted training of genetic counseling professionals.

Background: Communicative Practices in Genetic Counseling

Building on previous work (see in particular Sarangi, 2000; Sarangi et al., 2004, Sarangi et al., 2005), in the opening presentation Srikant Sarangi acknowledged that for a long time the genetic counseling process constituted a 'black box', especially from a communication perspective: while there had been an abundance of studies that looked at the *outcomes* of genetic counseling (e.g. effectiveness of decision-making, client satisfaction), our knowledge of what happened in the actual counseling clinic remained limited (Pilnick and Dingwall, 2001; Biesecker and Peters, 2001). In addition, over the years there have been many interpretations of what constitutes genetic counseling in different parts of the world: who delivers genetic counseling; what professional training is in place; and the nature of clients' access to genetic services. Recently there has been an increase in *process-oriented* studies of genetic counseling that take a discourse analytic perspective,[2] predominantly in the Western countries (e.g. the USA, the UK and Europe more generally), with not many studies in the Asia-Pacific region with the exception of Australia and more recently Hong Kong.

While definitions of genetic counseling adopted by professionals in SEA vary slightly, all of them draw on definitions used in the Western countries, for example Harper's definition of genetic counseling as "the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and of the ways in which this can be prevented or ameliorated" (Harper, 2004). The adopted definitions emphasize information-giving aspect of genetic counseling that concerns a number of specific topics, including the natural history of a genetic disorder; patterns of inheritance, onset and penetrance; the (un)treatability of specific conditions; potential advantages and disadvantages of genetic testing; level of genetic awareness; reproduction choices. This may be referred to as the 'consultation end' of a genetic counseling continuum. As far as genetic counselors are concerned, genetic

[1] We acknowledge the reductionist nature of the labels 'Western' and 'Asian', and that both of them encompass a huge diversity of cultures and languages. We nevertheless use the terms in a general sense to refer to two distinct traditions of genetic counseling research and practice.

[2] Discourse analysis focuses on context-specific nature of language use, both in written texts and spoken interactions. Spoken interactions are audio- or video-recorded and transcribed for analysis. Context is broadly understood as "physical, sequential, extra-linguistic and ideological dimensions of communication" (Sarangi, 2006:1).

testing scenarios are *always* inclusive of counseling (Arribas-Ayllon et al., 2011) that comprises another end of a genetic counseling continuum. The counseling involves facilitating client's autonomous informed decision making which takes into account complex psychosocial issues and risks associated with knowing and disclosing one's genetic status.

Taking a discourse perspective allows a more nuanced interpretation of communication in genetic counseling. Consider, for instance, how risk can be talked about in a genetic clinic. Risk talk can be mainly *information-oriented* consisting of explanations about population risk vs. individual risk as well as patterns of inheritance. Risk talk can be *counseling-oriented* as professionals set out to explore clients' psychosocial anxieties and concerns, and invite clients to reflect upon current and future risks for themselves and their family members as well as the implications of their decisions to test and/or disclose test results. Finally, risk talk can also be *communication-oriented* (communication as information transfer process), whereby professionals and clients actively elicit and display their understanding and assessment of risk information. According to a study by Sarangi et al. (2003), within the framework of risk counseling, risks of knowing and risks of disclosure are likely to assume more significance than risks of occurrence.

Extract 1 taken from a prenatal screening consultation in Hong Kong exemplifies the point about different dimensions of risk talk in genetic counseling. In the extract the nurse delivers information about the woman's risk of having a baby with Down's syndrome.

Extract 1

Both participants of this consultation are non-native speakers of English: the nurse (N) is Hong Kong Chinese, and the woman (W) is a Filipino.

1. N: Alright. So, this figure suggest that your risk of having Down's syndrome baby is very small.
2. W: Ok. ((nods))
3. N: Alright. Mm, you've got one more choice of having amniocentesis. So see whether you want to have amniocentesis. It's up to you because with this report, it do not suggest you to go for amniocentesis.
4. W: ((nods))
5. N: Mm, but (.) do you know the detection rate? (.) [of] the screening test?
6. W: [No.]
7. N: Is around eighty to ninety percent. It's a::round ninety percent.

In Extract 1 risk talk is *information-oriented* as the nurse informs the woman about her individual risk of having a child with Down syndrome, a choice of further testing, and the detection rate of the screening test. Risk talk is also *communication-oriented* as the woman displays her understanding of provided information (verbally, through minimal responses (*OK*, turn 2) and non-verbally (by nodding, turns 2 and 4)).

As an activity-type (Levinson, 1979; Sarangi, 2000) genetic counseling is a hybrid activity in that it resembles both mainstream medical encounters and other counselling/therapy settings. Genetic counseling differs from medical encounters in terms of *purpose*, *content* and *structure* of interactions. By a similar token, genetic counseling resembles other counseling/therapy settings such as psychotherapy, HIV/AIDS counseling, social work encounters and family mediation. Genetic counseling can be characterized along a number of interactional features such as process (or, interaction) orientation being more significant than outcome (or topic) orientation; professionals' use of reflective and hypothetical questions in order to elicit clients' perspectives on past, present and future scenarios, etc.

Communicative differences in genetic counseling encounters are likely to show across different conditions as well as across clients with different ethnic and socioeconomic backgrounds (Rapp, 1988; 1999).

Communication-Oriented Research on Genetic Counseling/Consultation in Hong Kong

In what follows, we provide an overview of communication-oriented research undertaken in Hong Kong that involves clients of diverse backgrounds. In the last six years, a team of language/communication scholars and healthcare professionals in Hong Kong has been collaborating on several projects including prenatal screening for Down's syndrome; counseling/consultation for G6PD deficiency and Sudden Arrhythmia Death Syndrome (SADS); and more recently pre-implantation genetic diagnosis and counseling. Genetic counseling/consultation practices in Hong Kong are characterized by a wide sociocultural and socioeconomic diversity of clients due to the multicultural and multilingual nature of the city, and its geopolitical and economic environment.

The communication-oriented research has begun to outline some striking differences in genetic counseling/consultation practices in Hong Kong in comparison to genetic counseling in other countries as reported in the literature. The research on prenatal screening, for example, has highlighted that professionals appear quite directive in these consultations, both explicitly by assuming that testing will take place and more implicitly through what information they offer or decide to withhold in particular circumstances (Pilnick and Zayts, 2012; Zayts et al., 2012). The clients, on the other hand, often occupy a minimalist participant status with regard to reception of information and decision-making. This trend has been attributed to a range of factors, including institutional regulations of these kinds of encounters, the medical (rather than

counseling) background of the professionals, socioeconomic and familial circumstances of the clients. From an interactional perspective, this pattern of decision-making may also be attributed to assumptions and judgments that professionals make about a client's ability to carry the 'burden' of decision-making. These judgments are made, at least partly, on the basis of clients' participant status in the interactions signaled through minimal responses which encourage professionals to be more directive; whereas clients' more active engagement is indicative that they are able to engage in autonomous decision-making (Zayts and Pilnick, in press).

The research project on prenatal screening has also focused on the impact of clients' diverse socioeconomic backgrounds on their decision-making. Specifically, it has shown that once clients' socioeconomic circumstances become visible in an interaction, the extent to which these are 'allowed' by doctors to impact on decision-making is subject to interpretation (Pilnick and Zayts, 2012). This observation supports previously published findings that doctors actively interpret patients' social characteristics and formulate stances towards lifestyle issues that patients raise (e.g. Silverman 1987; Lutfey and McKinlay 2009; Sorjonen et al. 2006). In the context of prenatal screening, the moral and ethical ramifications of the healthcare professionals' communicative practices may bear more profound effects.

Our ongoing research concentrates on counseling/consultation on G6PD deficiency with a focus on consultation vs. counseling as part of the communicative continuum (Zayts and Sarangi, 2013). More specifically, the research investigates the provision of genetic counseling services via telephone – the aim is to explore whether a distant mode of communication offers a feasible solution to catering for a dense client population in SEA, combined with a shortage of skilled workforce (Hospital Authority Strategic Service Plan 2009–2012). It is expected that the findings will be of relevance to other countries in SEA where the professional workforce is also limited.

As communication-based studies have begun to highlight, in terms of information provision, the encounters in Hong Kong are similar to their counterparts in other countries. Clients are given explanations about genetic disorders; available genetic testing and its advantages and disadvantages and other issues. The 'consultation end' of the genetic clinic encounter appears to take priority for reasons discussed earlier. By contrast, the counseling agenda such as discussion of various psychosocial issues, while present, appears to be marginalized. At a more nuanced interactional level, the research has highlighted that some key interactional features of counseling (e.g. the use of reflective or hypothetical questions) do not constitute the active communicative repertoire of professionals. These observations point to an array of issues related to the context-specific consultation/counseling practices in SEA.

The Current Practices of Genetic Counseling/Consultation in South East Asia

While other countries in SEA are yet to embark on communication-oriented research in the genetic clinic setting, an overview of their current practices is useful in identifying potential challenges that can be addressed through future research.

Table 1 (see Appendix) presents the current provision of genetic counseling services in South East Asia. In what follows we briefly introduce the main discussion points.

Who provides genetic counseling services and what types of genetic disorders are covered?

All presenters noted the absence or shortage of trained genetic counselors in their countries. Genetic counseling positions are available in Thailand (in thalassemia only) and Indonesia. In Hong Kong and Malaysia there are currently no genetic counselor positions supported by the Department of Health (Hong Kong) and the Ministry of Health (Malaysia) respectively. In Hong Kong the services are provided by a range of professionals including clinical geneticists, obstetricians specializing in maternal-fetal medicine, registered nurses, oncologists, surgeons and pathologists. In Malaysia, in four centers in Peninsular Malaysia, there are nine medical geneticists and two associate genetic counselors providing the genetic counseling service, which means that the ratio of medical geneticists to the total population is 1:3 million (the service is also provided by medical doctors and nurses). These statistics signal, first, an urgent need to establish genetic counseling as a profession in SEA; and second, the need to develop certification and training possibilities for genetic counselors. The training possibilities are in place in a number of countries in the form of 'on-the-job' (non-accredited) training, workshops and designated degree courses at local universities (e.g., Hong Kong,[3] Indonesia,[4] the Philippines,[5] and Malaysia).

Currently clients with a wide range of genetic conditions are managed in SEA that includes prenatal/reproductive genetics, pediatric genetics and adult-onset conditions (see Table 1). The presenters repeatedly stressed the multidisciplinary collaborations in managing clients. For example, pediatricians, pediatric cardiologists, clinical geneticists, and parent support groups worked together in managing pediatric genetic cases.

[3] http://www.obsgyn.hku.hk/whatsNew/MMSciGeneticCounselling/MMSci%20Genetic%20Counselling%20-1.htm
[4] http://www.cebior.fk.undip.ac.id/
[5] See Laurino et al., 2011; and http://ngohs.upm.edu.ph/grad_programs.php

The Impact of Cultural and Other Contextual Factors (Including Language) on Genetic Counseling Practice

Three main factors emerged as having an impact on genetic counseling in SEA: language, religion and culture. These concerns echo previously published research in multicultural and multilingual contexts in medical sociology, medical anthropology as well as medical education (Zayts and Pilnick, in press). With regard to genetic counseling involving clients from diverse cultural backgrounds, general counseling skills are not sufficient (Zayts and Pilnick, in press). Various frameworks of multicultural competence, also referred to as culturally appropriate/culturally sensitive genetic counseling (e.g. Weil and Mittman, 1993; Steinberg Warren 2011), have been developed to address the challenges of counseling in these contexts. The common components of various multicultural competence frameworks include, first, knowledge of ethnocultural groups and second, ethnocultural self-awareness, that is awareness by professionals about their own cultural beliefs and attitudes, and their impact on the counseling process (Ota Wang 1998).

With regard to SEA, the presenters noted the co-existence of official languages and local dialects, especially the influx of migrant groups with their own languages and dialects. The professionals reported particular challenges in using genetics terminology with speakers of different languages and dialects. For example, in Hong Kong, while the majority of the population comprises speakers of Cantonese (89.5 %), the remaining 10.5 % include speakers of Mandarin (1.4 %), other Chinese dialects (4 %) and other languages (5.1 %).[6] This leads to different constellations of language use in genetic consultations in Hong Kong: professionals and clients may either use English or Chinese as their first language or as a lingua franca (that is, language used by participants who do not share a common language), or they may use professional or lay interpreters in these encounters. Similar observations regarding genetic counseling in a multicultural and multilingual society have been reported in Malaysia in the context of counseling for hereditary breast and ovarian cancer (Yoon et al., 2011).

As regards the impact of religion and culture, the discussion at the workshop focused on the issues of consanguineous marriage, termination of affected pregnancy, the beliefs surrounding the causes of birth defects and abnormalities, and what consequences abnormalities are believed to bring on a family. For example, in Thailand 95 % of the population follows Buddhism. Among the general population, disabilities are widely understood in terms of one's 'karma', or the choices that a spirit makes before reincarnating. It has been noted that in some cultural contexts there are better mechanisms of accepting and coping with genetic disorders, whereas in other cultures such disorders are seen as a taboo. The issue of alternative traditional treatments that are believed to offer solutions for genetic abnormalities was mentioned.

The presenters also noted limited access to genetic counseling services in some remote areas and the financial considerations surrounding genetic testing. Promotion of genetic education among the general public came across in all presentations as the channel through which misunderstandings arising from cultural and religious diversity could be addressed in the delivery of genetic counseling services in SEA.

As a commentary to the discussion at the workshop, we would like to stress that the issues of language, religion and culture in genetic counseling are far more complex than the mere issue of the differences in participants' backgrounds: for example, language use, and in particular risk communication, encompasses informative, communicative, and counseling dimensions, all of which should be given due attention in the communicative process independent of the cultural, linguistic and religious backgrounds of the participants. In relation to religious and cultural diversity, while it may be tempting to make a link between cultural and religious backgrounds of the participants and what is happening in the genetic clinic, such an approach may not offer an unequivocal explanation of the differences and similarities in genetic counseling practices in SEA and other countries. Other important contextual factors may include, for example, specific institutional regulations, time constraints, and/or the specialization of the professionals involved in the provision of the genetic services. In other words, genetic clinic encounters in multicultural and multilingual contexts should be approached through the lenses of intertwined contextual factors, including but not limited to language, culture and religion. An empirically robust communication research agenda will no doubt offer much needed evidence for improving genetic counseling practice in SEA.

Conclusion

In this report we have outlined the rich tradition of communication-oriented research in genetic counseling settings outside of Asia, and have reviewed the studies currently ongoing in Hong Kong. We have indicated the importance of reflecting on the culture- and context-specific nature of genetic counseling/consultation encounters in SEA from the viewpoint of the counseling/consultation communicative continuum in terms of distinctive interactional features. A counseling mentality must be incorporated in to the genetic clinic encounter and for this to happen we need to peer into the 'black box' of current communicative practices in genetic clinic encounters in SEA. The research-based findings will then inform not only the curricula

[6] Census and Statistics Department, The Government of the Hong Kong Special Administrative Region. Population by ethnicity, 2001 and 2006. Retrieved from http://www.census.gov.hk/hong_kong_statistics/statistical_tables on 24 July 2012.

targeted at future genetic counselors but also the agenda for in-service professional training. A sustainable collaborative link between genetic professionals and language/communication scholars holds the key to future intervention based on reflections and constructive feedback in the form of workshops, training sessions, curricular input as well as on-line resources. The workshop at the Asia Pacific Conference on Human Genetics (APCHG 2012) which occasioned the sharing of genetic consultation/counseling practices in SEA and triggered this report was salutary. The workshop, we believe, served as an important 'stepping stone' in the process of developing the genetic counseling profession in SEA. In future years, we hope that communication in genetic counseling in SEA becomes a durable strand of the conference program and attracts practitioners to engage in emergent topics in genetic counseling.

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Appendix I

Table 1 Genetic counseling services in South East Asia (as of 2013)

Country	Who provides genetic counseling services	Types of genetic disorders/clients that receive genetic counseling services	Available training programs/courses/ 'in-house' training
Hong Kong	No genetic counselor position supported by the Department of Health, Hospital Authority or local universities. Genetic counseling is provided by: 1. clinical geneticists (<5); 2. obstetricians specializing in maternal fetal medicine and reproductive medicine; 3. pediatricians; 4. registered nurses (cases ascertained through prenatal and neonatal screening); 5. oncologists; 6. surgeons; 7. pathologists.	Prenatal: Down Syndrome; Thalassaemia; fetal anomalies detected on ultrasound; abnormal karyotype, chromosomal microarray or genetic findings from invasive procedures; family history of genetic disorders. Postnatal: pediatric; whole spectrum of birth defects due to chromosomal abnormalities, single gene defects, multifactorial inheritance and teratogens that affect growth and development or other body systems. Adult: reproductive problem; premarital or preconception counseling; family history of genetic disease; adult-onset genetic diseases; familial cancer syndrome(s); others.	For clinical geneticists: 'in-house' training with no local accreditation system; overseas training. For genetic counselors: Master of Medical Science (MMedSc) degree program in Genetic Counseling (the University of Hong Kong) For clinicians/laboratory technicians with an interest in genetics: short courses organized by various organizations. For oversees genetic counselors: an elective practicum.
Thailand	1. medical professionals; 2. trained genetic counselors (only in Thalassemia)	all (chromosomal, single gene, multifactorial etc.)	
Malaysia	No genetic counselor position available (proposed position is under consideration by the Ministry of Health Malaysia). Clinical genetics is recognized as a sub-specialty by the National Specialist Register in Malaysia. Genetic counseling is formally provided by: 1. clinical geneticists; 2. associate genetic counselors; 3. medical doctors and nurses.	Prenatal/reproductive genetics, teratogen counseling. Pediatric genetics: birth defects; inherited metabolic disorders; hemoglobinopathies; skeletal dysplasia; global developmental delay; neurogenetics. Family history: learning difficulties; consanguinity; other inherited conditions. Adult-onset genetics: familial cancer; neurogenetics.	Sub-specialty training in medical genetics. Thalassaemia counseling workshop for nurses and allied healthcare professionals. Workshop on genetic counseling for medical practitioners and nurses.

Table 1 (continued)

Country	Who provides genetic counseling services	Types of genetic disorders/clients that receive genetic counseling services	Available training programs/courses/ 'in-house' training
Indonesia	Genetic counseling is provided by: 1. genetic counselors (mostly with physician background); 2. uncertified medical geneticists who were trained overseas (>1 year)	Prenatal (for non-invasive only): family history of genetic disorders: chromosomal abnormalities (mainly Down syndrome and translocation), repeated pregnancy lost; single gene disorders, such as Fragile X syndrome, Congenital Adrenal Hyperplasia (CAH), Androgen Insensivity Syndrome (AIS) and Thalassemia. Postnatal: birth defects/congenital anomalies due to chromosomal abnormalities and single gene defects; intellectual disability or early-onset cognitive disease (Fragile X Syndrome, autism); disorders of sex development (mainly CAH, AIS and severe hyospadia), neuromuscular (DMD, Limb Girdle MD). Adult: reproductive problems (repeated pregnancy loss); infertility; untreated (late diagnosis) of disorders of sex development; family history of genetic diseases; premarital counseling; neurodegenerative late onset (e.g. SCA).	Genetic counselors are available. The Master of Biomedical Science program with a major in Genetic Counseling has been established at the University of Diponegoro (Semarang, Indonesia) since 2006
Philippines	1. Clinical geneticists (Philippine General Hospital and private clinics of Clinical Geneticists); 2. Genetic Fellows (Philippine General Hospital); 3. Pediatric Residents rotating at the Section of Genetics at the Philippine General Hospital 4. Genetic counseling students under supervision.	Pre-natal: congenital anomalies detected through ultrasound; previous child with a chromosomal abnormality; history of anomalies in a previous pregnancy; history of recurrent miscarriages; history of infertility. Post-natal: infants and children with birth defects/dysmorphism; neuro developmental disorders; growth disturbances. Adult: hereditary cancer; history of familial type of mental retardation; a relative with a diagnosed or an undiagnosed syndrome; adult-onset genetic disorders	MS in Genetic Counseling is offered by the Department of Pediatrics, College of Medicine, University of the Philippines, Manila, since 2011.

Appendix II

Table 2 Notation of transcription conventions

Symbol	Meaning
(.)	A tiny gap between the utterances
[	The beginning of overlapping speech
]	The end of overlapping
::	Prolongation of sound
(())	Transcriber's comments additional to transcription

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