Genetic counseling in an indigenous Filipino community with a high prevalence of *A2ML1*-related otitis media

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Abstract

In this report, we describe the knowledge and beliefs on causes and management of otitis media of an indigenous Filipino community with a high prevalence of otitis media that is associated with an *A2ML1* variant. Community lectures and individual genetic counseling were provided as intervention. Knowledge, beliefs, and health care-seeking behavior pertaining to otitis media were assessed pre- and post-genetic counseling. Twenty-five heads of households were interviewed. Beliefs regarding etiology of ear discharge varied widely, with swimming in the sea as the most commonly cited cause of ear discharge. During the post-counseling session, poor personal hygiene, dirty environment, and familial inheritance were mentioned as risk factors for otitis media or ear discharge. Knowledge about the genotypes for the *A2ML1* variant and otitis media diagnoses within the household influenced beliefs on the role of hygiene and genetic susceptibility to otitis media and attitudes towards health care-seeking behavior. Genetic counseling was associated with variable improvement in knowledge on otitis media and in their understanding of genetic susceptibility to otitis media due to the *A2ML1* variant. Language barriers, literacy level, cultural factors, and the complex nature of genetic information made genetic counseling in the particular population a challenge. Insights drawn from this experience recommend follow-up visits in the community.

Keywords *A2ML1* · Filipino · Genetic counseling · Indigenous community · Otitis media

Introduction

Otitis media (OM) is a condition characterized by inflammation of the middle ear and can be acute or chronic in nature (Qureishi et al. 2014). Acute OM (AOM) is often characterized by acute ear pain, irritability, and fever. Bulging eardrums due to pus or fluid buildup in the middle ear are commonly present, and perforation with ear discharge can occur (Subcommittee on Management of Acute Otitis Media et al. 2004). Common culprits are bacteria and viruses that are associated with upper respiratory tract infection, colonize the nasopharynx, travel through a compromised eustachian tube, and infect the middle ear mucosa (Kalu et al. 2011). On the other hand, OM with effusion (OME) is a condition characterized by the accumulation of glue-like fluid behind intact eardrums without the signs of acute inflammation (Venekamp et al. 2016). The most commonly reported symptom of OME is hearing loss, which can result in speech delay and learning difficulty among young children (Shriberg et al. 2000). If OM persists after ≥ 3 months, OM is considered

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chronic (Rosenfeld et al. 2016). When indicated, treatment such as antibiotics and surgical insertion of ventilation tubes are important in preventing permanent hearing loss (Qureishi et al. 2014; To et al. 2014). Globally, the incidence of AOM is reported at 10.85% (Monasta et al. 2012). In the Philippines, the overall prevalence of clinically diagnosed AOM is 9.6% among children aged 0–12 years old, with AOM being most prevalent among children aged 0–2 years old (Caro et al. 2014).

While acquiring infection is multifactorial, the role of genetics in infectious diseases is anchored in the concept that inherent genetic makeup contributes to the susceptibility of an individual to an infection. At times, specific genetic mutations and syndromes predispose an individual to infectious diseases (Burgner et al. 2006; Casanova and Abel 2007). There is growing evidence that susceptibility to OM has a strong genetic component. In twins and triplets, the heritability of OM susceptibility was estimated to be > 70% (Casselbrant et al. 1999). Several candidate genes (e.g., *TLRs*, *CD14*, *IL-1*, *TNF- α*) related to innate or adaptive human immunity were also associated with recurrent OM (Rye et al. 2012; MacArthur et al. 2014).

In the Philippines, a duplication variant c.2478_2485dupGGCTAAAT (p.Ser829Trpfs*9) within the *A2ML1* gene was found to be associated with OM in an indigenous Negrito community (Santos-Cortez et al. 2015); this variant confers an increased risk for all types of OM (Santos-Cortez et al. 2016b). In this indigenous Filipino population of around 200 individuals from 30 household units, the prevalence of OM was ~ 50% (Santos-Cortez et al. 2016b). Historically, this indigenous community has inhabited this region for centuries but they were racially segregated due to their physical features (e.g., dark-colored skin, short stature, flat nose, curly hair). This resulted in intermarriage within the community, and over centuries, the proportions of specific genotypes became different from the general population (Santos-Cortez et al. 2015).

Within the community, the *A2ML1* variant was identified to be the primary factor for OM and its progression (Santos-Cortez et al. 2016b). Generally, individuals with AOM are diagnosed within 0–4 years, then have either recurrent AOM or chronic OM (COM) by age 2–5 years, with peak of COM occurrence at 10 years of age. From 6 to 12 years of age, they either have persistent COM or heal. Typically, the incidence of OM tends to decrease as they enter adult life. However, for individuals with the *A2ML1* variant, OM can recur up to about 30 years of age, which suggests that the *A2ML1* variant influences OM onset and recovery (Santos-Cortez et al. 2016b). Microbiome studies performed in the community also indicate that the *A2ML1* variant induces changes in the middle ear microbiome (Santos-Cortez et al. 2016a, b). The discovery of the *A2ML1* duplication variant in the indigenous population and its implications to OM susceptibility has a significant

impact not only to the health of individual variant carriers, but also to the whole indigenous community. Communication of these findings to the affected population is critical.

Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. The process includes interpretation of family and medical history to assess chance of occurrence or recurrence, provision of education on inheritance, genetic testing, and disease management and prevention, as well as promotion of informed choices and adaptation to the condition (Resta et al. 2006). This is a relevant intervention to the indigenous Negrito community with a high proportion of *A2ML1* variant carriers, who are more susceptible to develop OM compared to the general population. In this study, we evaluated the knowledge and beliefs about OM before and after genetic counseling among members of the indigenous Filipino community.

Methods

The study was conducted according to the principles of the Declaration of Helsinki. Prior to initiation, it was approved by the University of the Philippines Manila Research Ethics Board, the National Commission on Indigenous Peoples, and the Colorado Multiple Institutional Review Board. Aside from community consent, informed consent was also obtained from individual participants.

Across the indigenous Filipino Negrito community where the *A2ML1* variant was identified, heads of all 30 household units in the community were invited to participate in the study. Household visits and survey interviews were conducted by a team of three researchers familiar with the community to verify pedigree relations and household data and to administer a 10-item survey questionnaire assessing participants' knowledge and beliefs on OM. This assessment was conducted using a combination of the national language and the local dialect.

After the initial knowledge assessment, all members of the community were invited to a community lecture on OM provided by an otorhinolaryngologist-genetic epidemiologist and a clinical geneticist. The lecture subtopics included the pathophysiology of OM and its prevalence in the general population, basic genetics education, an overview of the *A2ML1* variant, its pattern of inheritance, its implication to OM disease risk, and preventive measures against OM. This community lecture was followed by individual genetic counseling sessions.

A day after the community lecture, the geneticist and the genetic counselors conducted post-genetic test counseling among the participants found to have an *A2ML1* variant and

pre-genetic test counseling for heads of households and/or individuals interested to undergo DNA testing. After each counseling session, participants were asked the same 10-item survey questions. Answers to this 10-item survey before and after genetic counseling were compared. Variables for analyses included participants' age and educational level; number of household members; number and proportion of *A2ML1* variant carriers; new DNA samples provided; OM diagnosis (based on examination findings of otorhinolaryngologists); and pre-/post-counseling responses to the ten questions. Standard statistical analyses (Fisher's exact test for dichotomous independent variables, logistic regression for continuous independent variables) were performed using R software.

Results

Characteristics of participating households

Out of thirty (30) households in the indigenous community, twenty-five (83.3%) had at least one representative (i.e., the head of the household) interviewed regarding their personal beliefs on OM during the pre- and/or post-genetic counseling sessions. Twenty-seven representatives from 25 households

participated, with two households sending both heads of their units (i.e., father and mother) for counseling and survey interview. For these two households, only the data of the representative who answered the pre-genetic counseling survey was included in analysis. Thirteen (52%) heads of households answered the ten questions on OM pre- and post-genetic counseling. Eleven (44%) participated in the post-genetic counseling survey only, while one participant answered the pre-counseling survey only.

Table 1 provides a summary of the characteristics of participating households, including demographic data, presence of *A2ML1* variant and/or OM, and percentage of DNA samples collected and/or tested. Household representatives who participated in the survey were mostly female (96%), aged 30–40 years old (60%) with elementary education as their highest educational attainment (52%), and held minimum-wage jobs (52%). Twenty-one households (84%) have members who have previously submitted DNA samples sequenced for the *A2ML1* duplication variant prior to the interview. Half of those sequenced were found to carry the *A2ML1* variant. Similar to the reported prevalence at the population level (Santos-Cortez et al. 2015, 2016b), for most households, about half of the members per household were diagnosed with OM, and of those with OM, approximately 30% had chronic OM.

Table 1 Summary of characteristics by household ($n = 25$)

Variable per household	Counts	
Number of household representatives (i.e., head of household) interviewed pre- and/or post-genetic counseling	Both pre-/post-counseling = 13; pre-counseling only = 1; post-counseling only = 11; counseled as a couple = 2	
Educational level, head of household interviewee	None = 1; elementary = 13; high school = 6; vocational = 1; college = 1; NA = 3	
Job, head of household interviewee	Housekeeper = 8; community volunteer = 5; basketweaver = 2; laundrywoman = 2; nanny = 1; housewife = 1; none = 1; NA = 3	
	Median	Min-max
Age in years, head of household interviewee	36	23–60
n household members	6	3–11
% household members with DNA samples	75	33.3–100
n household members tested for <i>A2ML1</i> variant	2	0–6
% tested for <i>A2ML1</i> variant	50.0	0–83.3
n <i>A2ML1</i> variant carriers	1	0–5
% <i>A2ML1</i> variant carriers per tested by household	50.0	0–100
n new DNA samples	1.5	0–4
n otitis media, all types	3	1–7
% with otitis media, all types	42.9	11.1–100
n chronic otitis media	1	0–3
% chronic otitis media	16.7	0–50

NA not available

Knowledge and beliefs on otitis media, pre-genetic counseling

In the indigenous community, the term “buog” was used to refer to ear discharge and was used as a surrogate term for OM. It should be noted that none of the individuals examined were diagnosed with otitis externa or outer ear infection with discharge; thus, in this population, the finding of ear discharge is almost always due to OM. Prior to counseling, “buog” was identified to have been mainly caused by swimming in the sea or bathing. One explanation provided by a few respondents was that ear discharge develops when a person is unable to remove the sea or bath water which entered the outer ear. Individuals whose mothers tend to look at and/or touch fish traps during their pregnancy (i.e., the Filipino cultural concept of “lihi” also known as maternal impressions) were believed to be inclined to “swim in the sea all the time” and consequently are more likely to develop ear discharge. “Buog” was also described to result from having colds or a damaged eardrum. Participants answered that both children and adults may develop “buog.” However, among infants and toddlers, breast- and bottle-feeding were identified as possible causes of ear discharge. Ear discharge was believed to develop when milk gets trapped in the infant’s ear. One participant mentioned that sometimes “milk comes out of the ears during feeding” and believed it to cause ear discharge among the younger age groups. “Buog” was also believed to manifest when a person takes in food he/she was not allowed to eat.

Eleven of 14 respondents noted the relationship among personal hygiene, environmental cleanliness, and ear discharge. Poor personal hygiene and unclean environment were recognized as factors increasing the risk to develop OM (Table 2). Five respondents identified OM as heritable while three of the fourteen household representatives considered OM as infectious (Table 2).

Members of the indigenous community shared that they go to a medical doctor for checkup or otherwise clean their ears with cotton swabs to remove the discharge. One respondent mentioned using chicken bones to clean his ear of discharge and another stated that he waited for a medical mission to come to the community for free consultation and management, including medications. Majority of the household representatives also agreed that immunization may help prevent development of ear discharge.

Knowledge and beliefs on otitis media, post-genetic counseling

After genetic counseling, swimming and bathing remained the most perceived cause of ear discharge, although there was some decrease in proportion of individuals who believed so (Table 2). Colds, damaged eardrum, and breast- or bottle-

feeding were still mentioned as potential causes of ear discharge.

After the community lectures and genetic counseling, other perceived causes of otitis media emerged (Table 2). Among the post-genetic counseling group, alternative causes of ear discharge were identified, including poor personal hygiene, familial inheritance, dirty environment, and allergies. Compared to the pre-counseling group, a smaller proportion of respondents answered that it is normal for children to have ear discharge and for children to have hearing loss.

The proportion of respondents who would self-clean the ears with cotton swabs in order to manage ear discharge remained similar pre- and post-counseling. Compared to the pre-counseling group (92.9%), a smaller proportion of respondents post-counseling (41.7%) mentioned seeking physician consultation for management. Some participants answered self-medication using oral antibiotics ($n = 3$) or putting herbal medicine in the ear ($n = 1$) as ways to manage ear discharge. Post-counseling, the belief that immunization will help prevent ear discharge increased from 71 to 100%.

Bivariate analysis of post-counseling responses and identified variables (Table 3) showed that the number of household members previously tested for the *A2ML1* variant, the number of known *A2ML1* variant carriers, the number of members per household who provided new DNA samples, the demographics of respondents (i.e., age and educational attainment), and history of pre-counseling interview influenced their post-counseling knowledge and beliefs on ear discharge ($p < 0.05$). For instance, during the post-counseling survey interview, respondents from households with more members found to have the *A2ML1* variant tend to believe that OM is heritable and that poor personal hygiene does not contribute to its development. Those with more household members who were previously tested for the *A2ML1* variant (regardless of actual genetic test results) also did not consider poor personal hygiene as a possible cause of OM. Younger respondents tend to believe that OM only affects children. Respondents who attended both pre- and post-genetic counseling survey interviews were also more likely to seek consultation with a physician to manage ear discharge.

Discussion

Otitis media (OM) knowledge improved after genetic counseling, specifically regarding OM etiology. Environmental factors (i.e., poor personal hygiene and dirty environment) and familial predisposition, which were not mentioned prior to genetic counseling, were recognized as causes of OM post-genetic counseling. Increased recognition of familial inheritance is also most apparent among representatives of households found to have the *A2ML1* variant. These results

Table 2 Answers to ten questions on knowledge and beliefs regarding otitis media

Question	Pre-counseling, <i>n</i> = 14 (%)	Post-counseling, <i>n</i> = 24 (%)
(1) Do you believe it is normal for children to have ear discharge?	No = 11 (78.6)	No = 13 (54.2)
(2) Do you believe it is normal for children to have hearing loss?	No = 12 (85.7)	No = 19 (79.2)
(3) What do you think is the cause of ear discharge? ^a		
(3.a) sea swimming/bathing	10 (71.4)	15 (62.5)
(3.b) colds	5 (35.7)	7 (29.2)
(3.c) damage to eardrum	2 (14.3)	3 (12.5)
(3.d) breast- or bottle-feeding	2 (14.3)	3 (12.5)
(3.e) poor personal hygiene	0	8 (33.3)
(3.f) familial inheritance	0	6 (25.0)
(3.g) environment (dirty)	0	3 (12.5)
(3.h) I don't know	0	3 (12.5)
(3.i) not knowing how to remove water from ear	0	2 (8.3)
(3.j) allergy	0	1 (4.2)
(3.k) eating specific foods	1 (7.1)	0
(4) What do you do when there is ear discharge or ear problem? ^a		
(4.a) self-clean, usually with cotton swabs ^b	11 (78.6)	19 (79.2)
(4.b) see doctor/check-up	13 (92.9)	10 (41.7)
(4.c) self-medicate, usually oral antibiotics	0	3 (12.5)
(4.d) put herbal meds in ear	0	1 (4.2)
(4.e) wait for medical mission	1 (7.1)	0
(5) Do you believe that having ear discharge (due to otitis media) is heritable?	Yes = 5 (35.7) Maybe = 5 (35.7) Don't know = 1 (7.1)	Yes = 15 (62.5) Maybe = 3 (12.5)
(6) Do you think only children can have ear discharge, or also adults?	Both = 12 (85.7) Children = 2 (14.3)	Both = 18 (75.0) Children = 5 (20.8) Don't know = 1 (4.2)
(7) Do you think personal hygiene has a relation to ear discharge?	Yes = 11 (78.6)	Yes = 23 (95.8)
(8) Do you think environmental cleanliness has a relation to ear discharge?	Yes = 11 (78.6)	Yes = 21 (91.7)
(9) Do you think ear disease/otitis media is transmitted from person-to-person?	Yes = 3 (21.4) Don't know = 1 (7.1)	Yes = 7 (29.2) Maybe = 1 (4.2)
(10) Do you believe that immunization can help in preventing ear discharge?	Yes = 10 (71.4) Maybe = 1 (7.1)	Yes = 24 (100)

^a Items a–k are answers provided by interviewees

^b One respondent answered use of chicken bones to clean the ears

demonstrate change in knowledge from pre- to post-counseling and are indicative of the willingness of participants to accept new information when a plausible explanation on the cause of the condition was provided. The community lectures and genetic counseling provided greater understanding on the role of environmental factors in increasing their risk to develop OM. In this study, we found that environmental factors as a cause of OM became secondary to a bigger appreciation of familial predisposition to OM for those whose tests revealed that they were carriers of the *A2ML1* variant post-genetic counseling. This increased awareness of familial inheritance as a factor influencing the development of OM could be attributed to the education and information given during the genetic counseling session.

Genetic counseling allowed participants to receive another perspective about their condition. However, it remained an individual's choice to use this information to change his health status and health-seeking behavior. Even with new information received during counseling, some beliefs contrary to medical knowledge shared during counseling persisted. For instance, even after the community lectures and genetic counseling, swimming, bathing, and breast- and bottle-feeding were still recognized as potential causes of OM. Swimming and bathing per se do not contribute to OM or OM recurrence unless the tympanic membrane is perforated. Additionally, breastfeeding (as opposed to bottlefeeding) protects against OM (Chonmaitree et al. 2016), and it was stressed to all counselees during the lecture and genetic counseling sessions that

Table 3 Bivariate analyses with p values < 0.1

Post-counseling responses	Independent variable per household	p value
Q3e: Belief that poor personal hygiene <i>does not lead</i> to otitis media	Increased proportion of <i>A2ML1</i> variant carriers	0.03*
Q3e: Belief that poor personal hygiene <i>does not lead</i> to otitis media	Increased number of previously tested for the <i>A2ML1</i> variant	0.04*
Q3e: Belief that poor personal hygiene <i>leads</i> to otitis media	Increased number of newly submitted DNA samples	0.0002*
Q3f: Otitis media is inherited in families	Increased number of newly submitted DNA samples	0.09
Q4a: Self-clean ears with discharge	Increased number of diagnoses of chronic otitis media	0.05
Q4b: See a doctor for ear discharge	Interviewed pre-counseling	0.047*
Q4b: See a doctor for ear discharge	Increased number of previously tested for the <i>A2ML1</i> variant	0.09
Q4b: See a doctor for ear discharge	Increased number of <i>A2ML1</i> variant carriers	0.06
Q5: Belief that otitis media is heritable	Increased number of <i>A2ML1</i> variant carriers	0.02*
Q5: Belief that otitis media is heritable	Increased proportion of <i>A2ML1</i> variant carriers	0.02*
Q5: Belief that otitis media is heritable	High school education or higher	0.048*
Q5: Belief that otitis media is heritable	Increased proportion of otitis media	0.06
Q6: Belief that only children can have ear discharge	Younger respondent age	0.03*
Q7: Belief that personal hygiene is related to otitis media	Younger respondent age	0.01*

* p value < 0.05

the high breastfeeding rates within the population (82%) should be continued (Santos-Cortez et al. 2016b). Though these were discussed during the community lecture and individual counseling, participants continue to associate OM with observable behavior present among those who developed the disease. Changing these persistent misconceptions requires multiple health education and genetic counseling sessions.

Results of the study also show that the belief that it is normal for children to have ear discharge and hearing loss persisted post-counseling, albeit with some decrease in proportion of respondents with these false beliefs. It is possible that *A2ML1* variant carriers who were informed about the results of their genetic test presumed that OM naturally occurs among those with the variant; however, this will need further validation. Conversely, the observation of a very high prevalence of OM (which manifests as ear discharge with or without hearing loss) in children within the community due to genetic predisposition may be interpreted as the community's "norm," and this belief based on their own observations may not be easily corrected. Though majority of the respondents believe that ear discharge affects both children and adults, approximately one-fifth of them, mostly younger respondents, believe that the condition only presents in children. This perception may be due to OM being more common in children than adults (Santos-Cortez et al. 2015, 2016b). Furthermore, younger respondents who have no personal history of OM are less likely to be aware of adults in the community who have active OM persisting in adulthood.

Improvement in the knowledge about preventive measures for OM, specifically on the role of immunization, is apparent post-counseling. This improvement in knowledge on preventive measures rather than curative management of OM is likely because the community lecture and genetic counseling

focused on health promotion and disease prevention. For treatment strategies, self-cleaning of the ears with cotton swabs, which can further cause infection and trauma if not done correctly, remains the most common response when it comes to addressing ear discharge. This is reflective of the community's lack of access to health care including ear specialists and surgical facilities.

Survey results show a significant reduction on the number of participants who identified consulting with a physician as a plan of management for individuals with OM. This decrease in the proportion of participants willing to seek consultation for OM may be due to the perception that this condition is "normal" for children and may also result from a personal sense that nothing can be done for OM given that they have the genetic variant. The latter is encapsulated in the typical Filipino expression of "Bahala na," which is interpreted as submission of one's fate to a higher being and is a common defense mechanism used when facing adversity (Abad et al. 2014). Even though multiple prevention strategies were included in the lecture and during the counseling sessions, the participants may still have difficulty understanding what it means to carry an *A2ML1* variant and how this information on their genetic susceptibility to OM can help them utilize strategies to prevent otitis media.

Challenges in genetic counseling

The difficulty of genetic counseling in this population stems from multiple factors, including language barrier, low educational level, cultural practices, the complexity of the disease due in part to gene-environment interaction, and poor health access.

Language barrier and literacy level

The Philippines is a linguistically diverse nation which presents challenges in communicating health information. While majority of Filipinos understand and speak the national language (i.e., Filipino), miscommunication can happen for individuals where the local dialect is more widely spoken than Filipino and for those with only primary or secondary level of education. In this study, majority of the clients were unable to finish elementary education and are not fluent in Filipino. It would have been ideal if, instead of employing a translator, the genetic counselors providing the counseling are able to speak the local dialect. However, this is not yet feasible given the current lack of genetic counselors in the country.

In the Philippines, the Master of Science in Genetic Counseling (MSGC) is a program established in 2011 and is offered solely by the University of the Philippines (Laurino and Padilla 2013). Since its establishment, the MSGC program has produced eleven genetic counselors, but only ten genetic counselors currently provide genetic counseling services. Eight genetic counselors are based in the country's capital, Manila, within the northern island of Luzon while the other two work within centrally located islands that require at least a day of travel to the indigenous community and speak a language different from the indigenous population's dialect.

Though the MSGC program enrolled several trainees from across the Philippines, many of whom speak local dialects, the sheer number of ethnolinguistic groups in the country makes provision of genetic counseling services in all possible dialects impracticable (Simons and Fennig 2017). Therefore, language barrier is to be expected in the practice of genetic counseling in the country. It has been shown that language barrier is likely in any genetic or genomic research (Nyika 2009), especially when the research participants in a community do not share the same dialect as the researchers.

Communicating medical and genetic concepts and terminologies to the participants who do not share the same language was a challenge recognized by the genetic counselors for this particular community. Therefore, a research assistant fluent in the local dialect, who has also been part of the study since the time of community engagement, was employed to assist in the translation of genetics education, especially for the participants with poor understanding of Filipino. The use of lay interpreters is a recommended measure to bridge the gap and address the language barrier between the genetic counselor and the client (Zayts et al. 2013).

Various strategies were also exhausted to break down the complex genetic information into terms that the community can understand. The community lecture and individual counseling provided made use of visual tools, such as flipcharts and drawings. Using flipcharts, drawings, and figures as visual aids is known to complement verbal communication (Ellington and Race 1993; Maduna 2002), especially in

presenting seemingly abstract concepts of genetics into meaningful interpretations (Greenwood Genetic Center 2013). A culturally appropriate genetics education flipchart, which was previously utilized in another population genetics study involving other indigenous populations of the Philippines, was also used during the community lecture to facilitate learning. Analogies using familiar objects and scenarios in the community were also used to explain genetic concepts. According to Mozdzierz et al. (2014), the use of analogies increases cognitive flexibility to help understand concepts from a different perspective. In a similar thought, using analogies in genetic counseling can help tailor a personalized understanding of genetic concepts. For example, there is no direct translation of the word “gene” to the local dialect, so to explain the abstract concept of a “gene,” analogies were employed. One example used is the “cookbook analogy,” which explains the gene akin to a recipe inside a cookbook, containing instructions on how to make a specific dish (Carver et al. 2008; Condit and Condit 2001; Condit et al. 2002). A cookbook is explained as similar to how genes contain information that makes the human body work and is responsible for physical traits like one's skin color and hair texture, among others. While it can be argued that using analogies such as the “cookbook or recipe metaphor” can be deterministic in nature (Condit et al. 2002), in a population where there is low genetic literacy, receiving genetic information using an analogy familiar to them may at least provide some level of understanding (Carver et al. 2008; Condit and Condit 2001; Condit et al. 2002). For this community, the genetic counselors considered the participants' demographics, such as their occupation and educational level, in determining which analogies and examples to use to further explain genetic concepts. However, a one-time genetic counseling intervention is not expected to fully address the genetics education and counseling needs of the participants. Several sessions of health education and genetic counseling are necessary for effective learning.

Cultural background and beliefs on hearing loss and otitis media

The indigenous community, where the study took place, is a relatively closed social system. Members are born into the community and social relations with external communities are often limited. Given the relative social isolation of the community, they have maintained certain cultural beliefs and practices, which have impact on health education and management. This may also result to difficulties in accepting “foreign” concepts, such as the idea of genetic susceptibility to OM. In this study, familial susceptibility to OM has been identified post-counseling particularly if asked as a yes/no question, but majority of the respondents (75%) did not recognize it as a factor influencing development of ear discharge when asked as an open-ended question (Table 2).

Persistent beliefs such as ear discharge and hearing loss being normal among children may also be an effect of living in a community where children often develop ear discharge and/or having close relationships with community members that their health conditions were usually shared with each other. There also exist some cultural notions on the cause and management of ear discharge, such as the concept of “lihi” (or maternal impressions) to explain certain tendencies or behavior known to result to the condition (Abad et al. 2014), or the use of chicken bones to clean the ear. These notions were acknowledged during the genetic counseling session and alternative explanations were provided non-directively.

Being a tightly knit community composed of intermarried households, genetic counseling and pre- and post-genetic testing become a challenge because results of the genetic test do not only affect the individual or their household but may impact related households as well (even those who refused genetic testing).

Counseling about an otitis media susceptibility gene with autosomal dominant inheritance and incomplete penetrance

Genetic counseling in this community was a challenge given the language barriers and literacy level of respondents but was further compounded by the complexity of genetic information to be relayed. The *A2ML1* variant discovered in this indigenous community is considered a susceptibility factor rather than a causative one inherited in an autosomal dominant pattern with incomplete or reduced penetrance of ~80% (Santos-Cortez et al. 2015). Relaying this complicated concept in lay language is a challenge for the genetic counselor. Understanding this information, even when given in simple terms, is difficult for participants found to be *A2ML1* variant carriers, especially with the uncertainties it implies.

The goals of the genetic counselors influence the method and practice of genetic counseling provided (Lewis 2002). In this study, the main goal of genetic counseling is to inform. Since this is the first genetic counseling session attended by the participants and given the limited time to provide counseling, the aim of the intervention is to provide knowledge about OM, the results and implications of genetic testing, and the genetic risks of the family. The teaching model, which highlights the client's grasp of the genetic information as its main goal, was used in the genetic counseling practice. This model assumes that an informed client can make use of their knowledge to make informed decisions (Lewis 2002).

Different strategies were also used to simplify the complicated genetic information, analogies (such as “draw lots” to demonstrate the odds of passing down the genetic variant to an offspring), visual tools (like flipcharts and infographics to explain the 80% penetrance), and simple words were used to

explain the concept. The use of both verbal and visual information has been known to improve understanding and retention of disclosed information (Nyika 2009). Using infographics or pictographs was also found to be beneficial in understanding probabilities, especially among individuals with low numeracy levels (Fagerlin et al. 2005; Hawley et al. 2008). While other forms of numerical risk figures were discussed (e.g., 50% or 1 in 2) to explain autosomal dominant risk of inheritance, the concept of “drawing of lots” may further complement understanding of risks as it offers another format which may be preferred by the community (Hallowell et al. 1997; Lautenbach et al. 2013).

In conclusion, in the indigenous Filipino community described, genetic counseling led to improvement in knowledge, particularly in helping respondents understand their increased susceptibility to OM due to the *A2ML1* duplication variant. However, optimizing genetic counseling service delivery in this indigenous community requires trained genetic counselors who can speak the local dialect or training of interpreters on basic genetics prior to community engagement. Insights drawn from the experiences of genetic counselors in this community suggest that additional genetics education and counseling may be needed to correct misconceptions and improve genetic literacy in this community. In clinical practice, cultural competence in the delivery of genetic counseling services is needed to enable better health education and support to individuals and families. Employing genetic counselors fluent in the local language or interpreters educated on basic genetics would likely optimize the delivery of genetic counseling services. Further development of culturally and linguistically appropriate materials to support the counseling provided will also help in improving the service. Furthermore, it is recommended that genetic counselors be involved at the start of any genetics research to identify culture-based strategies that can assist in providing better health education and promotion.

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Compliance with ethical standards

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

Ethical approval The study was conducted according to the principles of the Declaration of Helsinki. Prior to initiation, it was approved by the University of the Philippines Manila Research Ethics Board, the National Commission on Indigenous Peoples (NCIP), and the Colorado Multiple Institutional Review Board.

Informed consent Community consent was obtained as part of the NCIP approval process prior to the start of the study. In addition, informed consent was obtained from individual participants included in the study.

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