



# Genetic Counseling Status and Perspectives Based on a 2018 Professional Survey in Korea

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**Background:** Genetic counseling (GC) provides many benefits, including the identification of patients appropriate for testing, patient education, and medical management. We evaluated the current status of and challenges faced by GC practitioners in Korean hospitals.

**Methods:** An electronic survey was designed and conducted in 52 certified laboratory physicians belonging to the Korean Society of Laboratory Medicine, from August to September 2018. The questionnaires addressed three main categories of information: (1) current status of GC in hospitals; (2) essential qualifications of GC practitioners; and (3) challenges and perspectives for GC. Fisher's exact test was applied to analyze categorical data.

**Results:** Among a total of 52 participants who initially responded, 12 (23.1%) were performing GC either by direct or indirect care. GC clinics were opened regularly for one (33.3%) or more than three sessions (25.0%) per week; most respondents spent more time for pre-visit activities than in-person visits, both for a initial visit patient and for a follow-up visit patient. All laboratory physicians provided genetic information to their patients. Most recommended family genetic testing when indicated (91.7%), discussed disease management (75.0%), and/or ordered additional genetic testing (58.3%), and some referred patients to other specialists (8.3%).

**Conclusions:** Both patients and laboratory physicians concede the advantage of GC performed by clinical geneticists; however, the practice of GC involves several challenges and raises some concerns. The cost and support required to implement GC need to be addressed in order to provide qualified GC in Korea.

**Key Words:** Genetic counseling, Genetic testing, Laboratory physician, Survey

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## INTRODUCTION

Health care is increasingly moving towards personalized or precision medicine, which involves the use of genetic or genomic information to guide decision-making with regard to disease prevention, diagnosis, and treatment. To date, personalized medicine has mainly been integrated into oncology and the manage-

ment of disease through pharmacogenomics [1]. In addition, the diagnosis of rare genetic disorders has been achieved more readily through the advancement of molecular techniques, with several successful cases of treatment using personalized or targeted therapies [2]. In line with the promise of personalized medicine, the demand for genetic testing has increased rapidly in Korea since 2017, when the costs of targeted next-generation

sequencing (NGS) panels for inherited disorders and malignancies were first covered nationally [3].

As genetic testings are expensive, time-consuming, and may be performed only once in a patient's lifetime, their use should be carefully considered and optimized to provide the greatest benefit for patients. Genetic testings have been found to be cost-effective when combined with genetic counseling (GC), the economic and societal impact of which have been well-studied [4, 5]. Complete GC services provided by a clinical geneticist are more cost-effective than brief counseling provided by a general physician; appropriate patient communication depends on the physician's understanding of genetic testing procedures, results, and the implications thereof [6, 7].

Patients are largely in favor of GC, especially for rare genetic disorders, although a substantial proportion of patients or families in Korea have not undergone GC and have no access to relevant information [8]. Primary-care physicians and non-genetic specialists are often uncomfortable communicating the results of even a single gene testing to their patients or have some negative bias towards the value of genetic testing for their patients [7]. Additional barriers to the use of genetic testing results in clinical care include limited understanding of genomic information, confusion regarding terminology, and the volume of information arising from genetic testing [9].

With an increase in demand for GC in clinical settings, we evaluated the current status of and challenges faced by GC practice in Korean hospitals. To our knowledge, this study is the first to examine current GC practice in Korea.

## MATERIALS AND METHODS

As the majority of genetic services in Korea are provided and managed by departments of laboratory medicine, a national volunteering electronic survey was conducted in 108 certified laboratory physicians belonging to the Korean Society of Genetic Diagnostics including working members of non-teaching or teaching hospitals [10]. During August 2018, the survey was planned and the questionnaires were critically reviewed by experts within the Korean Society for Genetic Diagnostics genetic counseling committee. The data were collected using a prospective cross-sectional survey and merged in September 2018. The survey addressed three main categories of information: (1) current status of GC in hospitals; (2) essential qualifications of GC practitioners; and (3) challenges and perspectives of GC. Categories 1 and 2 were addressed through closed-ended questionnaires, and information for Category 3 was gathered through an open-

ended question in the survey. This study was exempted for approval by the Institutional Review Board of National Health Insurance Service, Ilsan Hospital, since it was anonymous and participation was voluntary.

Reponses were calculated as a percent of total respondents of each questionnaire. Statistical analyses were performed using R software, version 3.3.3, 64 bit (The R Foundation of Statistical Computing, Vienna, Austria). Fisher's exact test was used to determine whether the characteristics of hospitals performing GC differed or not. A two-tailed  $P < 0.05$  was considered statistically significant.

## RESULTS

The characteristics of the 52 respondents (survey response rate of 48.1%) are summarized in Table 1. Twelve respondents (23.1%) were performing GC either by direct or indirect care, such as participating as members of the multidisciplinary team or clinical support, and 40 respondents (76.9%) were not yet performing GC. The GC service differed among different-sized hospitals but did not differ significantly among service type ( $P > 0.05$ ). GC service did not significantly differ among geographical regions and did not vary by number of working laboratory physicians; however, the service was provided in all teaching hospitals.

Of the 12 respondents providing GC services, 10 had outpatient clinics for GC, and eight were providing GC by consultation. Most respondents reported 10 or fewer GC cases per month (66.7% of 12 respondents), while 16.7% of them reported 51–100 patients per month and clinics were open regularly. Most respondents spent 30–59 minutes on pre-visit activities prior to a patient's initial visit, whereas the time spent with patients during follow-up visits varied from <30 to 59 minutes. The time spent for in-person visit activities on new patients (such as collection of medical and family history, running risk models for the patient or family members, educating about genetics and conditions, explaining genetic testing results, and providing psychosocial support) was 15–30 minutes for nine respondents (75.0% of 12 respondents), and 15–30 minutes were spent on patient follow-up (58.3%; Table 2).

All respondents reported providing genetic information to their patients, recommending family genetic testing when indicated (91.7%), discussing disease management (75.0%), ordering additional genetic testing (58.3%), and referring patients to other specialists (8.3%). Further, they indicated that professional training in genetics and disease is essential, and the capacity to provide emotional support (59.6%) and information regarding the

**Table 1.** Summary of the characteristics of the 52 survey respondents

Characteristics	N of respondents (%)			<i>P</i>
	All (N=52)	Providing GC (N=12)	Not providing GC (N=40)	
<b>Hospital size</b>				
≤ 500 beds	8 (15.4)	1 (8.3)	7 (17.5)	0.078
501–1,000 beds	36 (69.2)	7 (58.3)	29 (72.5)	
1,001–2,000 beds	6 (11.5)	4 (33.3)	2 (5.0)	
> 2,000 beds	2 (3.9)	0 (0.00)	2 (5.0)	
<b>Type of hospital by care service</b>				
Tertiary hospital	28 (53.9)	7 (58.3)	21 (52.5)	0.754
Non-tertiary hospital	24 (46.2)	5 (41.7)	19 (47.5)	
<b>Geographic region within Korea</b>				
Seoul	20 (38.5)	5 (41.7)	15 (37.5)	0.861
Gyeonggi-do	12 (23.1)	3 (25.0)	9 (22.5)	
Busan	6 (11.5)	1 (8.3)	5 (12.5)	
Daegu	4 (7.7)	1 (8.3)	3 (7.5)	
Daejeon	3 (5.8)	2 (16.7)	1 (2.5)	
Gangwon-do	2 (3.9)	0 (0.0)	2 (5.0)	
Incheon	2 (3.9)	0 (0.0)	2 (5.0)	
Jeonrabook-do	2 (3.9)	0 (0.0)	2 (5.0)	
Gyeongsangnam-do	1 (1.9)	0 (0.0)	1 (2.5)	
<b>Academic status</b>				
Teaching	49 (94.2)	12 (100.0)	37 (92.5)	1.000
Non-teaching	3 (5.8)	0 (0.0)	3 (7.5)	
<b>Number of laboratory physicians* employed</b>				
1	5 (9.6)	0 (0.00)	5 (12.5)	0.235
2	7 (13.5)	1 (8.3)	6 (15.0)	
3	6 (11.5)	1 (8.3)	5 (12.5)	
4	18 (34.6)	3 (25.0)	15 (37.5)	
5	9 (17.3)	3 (25.0)	6 (15.0)	
6	0 (0.0)	0 (0.0)	0 (0.0)	
≥7	7 (13.5)	4 (33.3)	3 (7.5)	

\*Certified members of Korean Society of Laboratory Medicine.  
Abbreviation: GC, genetic counselling.

interpretation of genetic testing results (92.3%) and disease management (73.1%) is important in order to offer GC to patients. Majority of respondents considered general professionalism, knowledge of genetics and diseases, efficient provision of diagnoses, and the ability to communicate with other specialists and conduct scientific research can be the advantages of laboratory physicians related to GC (Table 3).

Although 94.2% of laboratory physicians conceded the advantages of GC performed by a clinical geneticist, we identified

several challenges and concerns regarding GC practice that can be categorized into the cost and support required to implement GC (Table 3).

## DISCUSSION

GC provides many benefits to patients, including the identification of patients appropriate for testing, patient education, and appropriate medical management [11]. From the data collected

**Table 2.** Status of the 12 respondents providing GC

Variables	N of participants (%)
<b>Service type*</b>	
Outpatient clinic	10 (83.3)
Consultation	8 (66.7)
Support online or by phone	3 (25.0)
Multidisciplinary team	2 (16.7)
<b>N of GC (or support) cases per month</b>	
≤ 10	8 (66.7)
11–50	2 (16.7)
51–100	2 (16.7)
<b>N of GC sessions per week</b>	
Irregular	4 (33.3)
1	4 (33.3)
2	1 (8.3)
≥ 3	3 (25.0)
<b>Time spent on pre-visit activities</b>	
<b>New patients</b>	
< 30 minutes	2 (16.7)
30–59 minutes	5 (41.7)
60–119 minutes	4 (33.3)
> 120 minutes	1 (8.3)
<b>Follow-up patients</b>	
< 30 minutes	5 (41.7)
30–59 minutes	5 (41.7)
Not applicable	2 (16.7)
<b>Time spent on in-person visits</b>	
<b>New patients</b>	
< 15 minutes	1 (8.3)
15–30 minutes	9 (75.0)
31–60 minutes	2 (16.7)
<b>Follow-up patients</b>	
< 15 minutes	3 (25.0)
15–30 minutes	7 (58.3)
Not applicable	2 (16.7)

\*Multiple-choice questionnaires.

Abbreviation: GC, genetic counselling.

in 2016, it has been estimated that there is only one genetic counselor per 89,324–16,390,000 residents (median 1:2,000,000) and one clinical geneticist per 262,867–10,930,000 residents (median 1:2,480,000) in the Asia-Pacific region, [12]; it is estimated that there are 4,242 certified genetic counselors in the United States [13]. In Korea, approximately 80 and 30 genetic

counselors were trained through a GC program offered by the Korean Society of Genetic Diagnostics and the Korean Board of Medical Genetics, respectively; however, only a limited number of clinicians can offer GC and only for certain disorders [8]. As GC services are not currently well recognized to be a valuable part of clinical genetics services in Korea, residency programs for training clinical geneticists are yet to be organized, and significant workforce shortages are expected in the near future.

Despite the general lack of genetic counselors, patients in other countries report being satisfied with the emotional ramifications of going through GC and the help they receive in making important decisions when the length of the counseling session is approximately 90 minutes [14], whereas in Korea the mean duration of in-person visits is generally 6.2 minutes in outpatient clinics [15]. In addition to the time spent during inpatient visits, an average of 7 and 3.5 hours are required for new patient and follow-up patient-related activities, respectively [16], which comprise a greater workload than any other specialty [17].

In addition to the workload, GC practitioners require certain training and certifications in order to address the medical, psychological, and familial implications of patients with genetic diseases [18]. In view of clinical practice, medical roles, psychosocial support, and case management practices are mostly needed [19]. However, such roles may not be sufficiently provided by one person, as management of genetic conditions can vary and is difficult to generalize [20, 21]. Integrating genomic information in medical curricula may help in providing genetic knowledge to the physicians enabling better communication. A flexible rather than a uniform GC model may be more adequate realistically; however, this requires effective communication between specialty physicians, as well as transforming genomic information into clinical laboratory results. Laboratory physicians are proficient at medical communication and play an essential role in translating genetic findings into clinical reports [22].

Given the uniform national healthcare insurance policy and legal requirements for medical personnel in Korea, establishing a GC service within the Korean National Health Insurance System (KNHIS) presents several difficulties. The main reason for the shortage of GC providers is lack of financial support, which is mainly influenced by the KNHIS. From a cost perspective, pre-testing GC can guide suitable genetic testing, which can minimize unnecessary testings, and post-testing GC can offer precise disease management for specific diseases [23]. The benefits of providing qualified GC either in a patient-specific manner or as a public health measure are invaluable. To secure qualified GC service in Korea, the KNHIS policy regarding reimburse-

**Table 3.** Concerns and difficulties (or issues) in performing GC as a laboratory physician

Variables	N of participants (%)
Current practice <sup>*,†</sup>	
Provide genetic information	12 (100.0)
Recommend family genetic testing	11 (91.7)
Management	9 (75.0)
Additional genetic testing	7 (58.3)
Referral to other specialties	1 (8.3)
Disease information	1 (8.3)
GC practitioner qualification <sup>*</sup>	
Genetics and disease-related profession	52 (100.0)
Interpretation of genetic testing results	48 (92.3)
Disease management	38 (73.1)
Ability to provide emotional support	31 (59.6)
Advantage of laboratory physician over genetic counselor <sup>*</sup>	
General professionalism	49 (94.2)
Genetics and disease-related profession	45 (86.5)
Communication with other specialists	36 (69.2)
Efficient diagnosis	32 (61.5)
Conduct scientific research	1 (1.9)
Challenges and concerns regarding GC practice <sup>‡</sup>	
Cost or support	
Time and effort load for outpatient clinic	8 (21.6)
Financial support for outpatient clinic setting	5 (13.5)
Cost-effectiveness of outpatient clinic management due to a small number of patients	3 (8.1)
No current reimbursement rate for GC	3 (8.1)
Effort load for acquiring latest medical scientific knowledge	1 (2.7)
Education or experience	
Lack of GC experience or concerns regarding the lack of such an experience	7 (18.9)
Lack of communication with patients	4 (10.8)
Management and conflicts	
Collaboration with other specialists for disease management	6 (16.2)
Conflict of interest with other specialists regarding GC clinic	1 (2.7)
Lack of consensus regarding the need for GC	1 (2.7)

\*Multiple-choice questionnaires; †Twelve respondents currently performing direct GC; ‡Thirty-seven respondents.  
Abbreviation: GC, genetic counselling.

ment for GC should be amended.

As noted in our survey, 23.1% of laboratory physicians are currently providing GC services regardless of the current challenges, and some are preparing to establish new GC clinics. This finding reflects increasing demand for genetic testing, especially using the NGS panel. Where GC cannot be directly provided by genetic counselors, general physicians may perform GC, espe-

cially in oncology clinics. As adoption of the knowledge and skills required to perform specific genetic tasks takes years and the annual growth rate for GC providers in Korea is far lower than in other countries [12, 13], providing GC services by laboratory physicians or clinicians familiar with genetics seems to be a reasonable alternative solution at this time.

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## AUTHOR CONTRIBUTIONS

Jieun Kim participated in summarizing data and writing of the paper. Sun-Young Kong participated in research design and advised on research methodology. Sung-Hee Han participated in research design and advised on research methodology. Jong-Won Kim participated in critically revising the study and provided support. Chang Ho Jeon participated in critically revising the study and provided support. Jongha Yoo participated in the performance of the research and supervised the full study.

## CONFLICTS OF INTEREST

None declared.

## RESEARCH FUNDING

None declared.

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