




# Current Issues, Challenges, and Future Perspectives of Genetic Counseling in Korea

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Genetic testing has become increasingly integrated into all areas of healthcare, and complex genetic testing usage continues to grow; thus, the demand for genetic counseling (GC) is likely to increase. However, it is unclear whether the current clinical GC capacity is sufficient for meeting the existing demand. This review describes the current issues, challenges, and future perspectives of GC in Korea based on a professional survey conducted among laboratory physicians. In view of the growing GC demand in the clinical setting, participants expressed a concern about the lack of support from the national healthcare insurance policy and legal requirements, such as certification, for GC practice. The implementation of genetic testing in the overall healthcare system in Korea is in an early phase. Proper implementation can be achieved through education and training of specialists, collaboration among healthcare personnel, proper regulatory oversight, genomic policies, and public awareness. Understanding the current GC capacity, issues, and challenges is a prerequisite for effective strategic planning by healthcare systems considering the expected growth in the demand for clinical genetic services over the next few decades.

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**Key Words:** Genetic counseling, Genetic services, Specialists, Healthcare system, Policy

## INTRODUCTION

Genetic testing usage in routine clinical practice has greatly increased over the past decade. As genetic testing expands with the growth of new genetic technologies, there is an emerging need for genetic counseling (GC). Although the demand for GC is increasing, and 94.2% of laboratory physicians concede the advantages of GC performed by professional personnel, several challenges and concerns regarding GC exist, especially regarding the need for support from national healthcare insurance policy and for laying out legal requirements for GC to ensure standardization and quality [1].

We assessed the status of GC and its present and future challenges in Korean hospitals based on professional surveys. An electronic survey was designed and conducted among 54 certified laboratory physicians associated with Korean Society of Laboratory Medicine between July and September 2020. The involvement of human participants was reviewed and approved by the Institutional Review Board of Soonchunhyang University Seoul Hospital, Seoul, Korea (IRB approval number 2020-07-002). The participants provided written informed consent to participate in the study. The contents of the survey responses were sufficient to complete a descriptive assessment and gain an in-depth understanding of the GC status in Korea. The ques-

tionnaire comprised three sections that probed for opinions on GC models in terms of status, challenges, and perspectives. The survey participants described the present and future GC workforce characteristics, current service delivery models, issues and challenges in clinical practice, and attitudes of healthcare providers and patients toward GC practitioners.

## GENETIC COUNSELING STATUS AND TRENDS

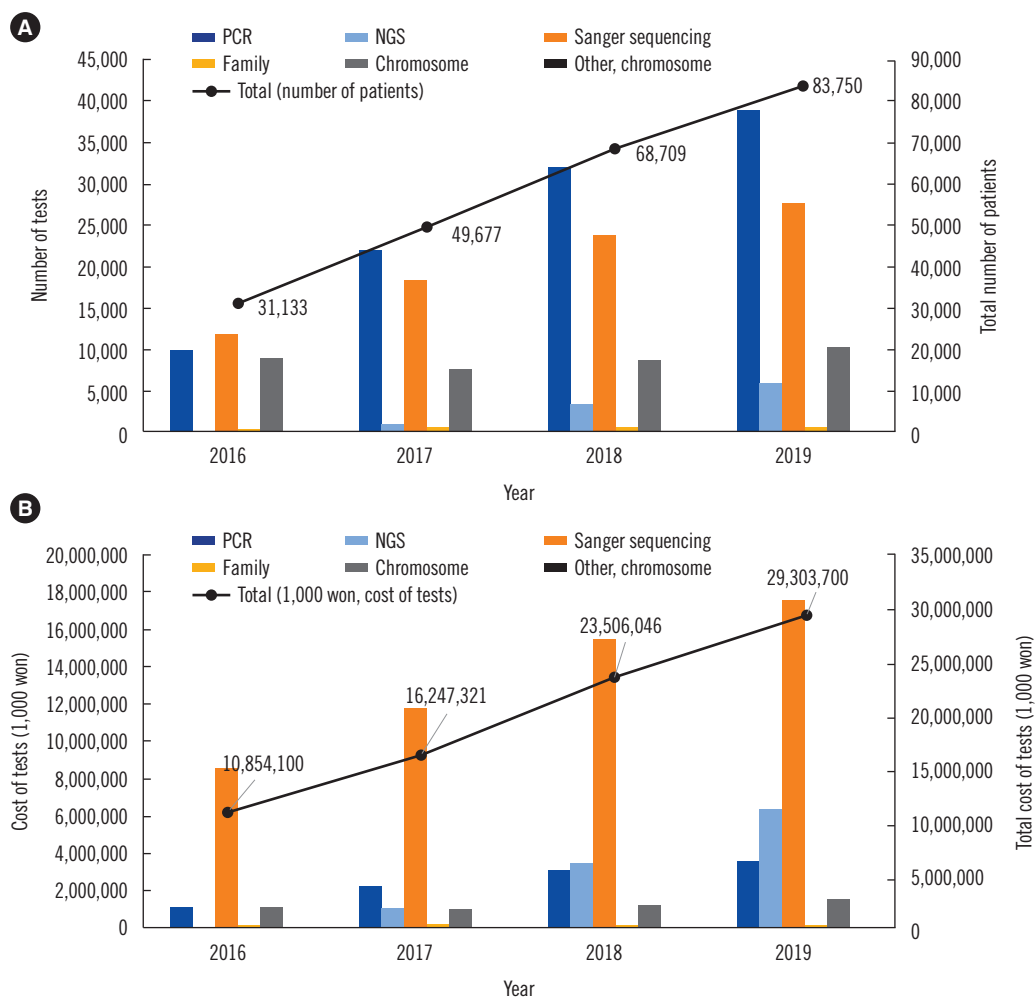
### Expansion of genetic testing in clinical practice

Healthcare is increasingly moving toward personalized medicine, which involves the use of genetic testing for decision-making related to disease prevention, diagnosis, and treatment. With the integration of the progress in genetics and overall healthcare

systems, the demand for genetic testing in clinical practice has increased rapidly in Korea. This trend has been driven by financial support from the Korean National Health Insurance System (KNHIS) for targeted next-generation sequencing panels for detecting inherited disorders and malignancies since 2017. The scope of KNHIS reimbursement for genetic testing has continued to expand over the past few years. Thus, the number of patients undergoing genetic testing and the cost of genetic testing continue to grow (Fig. 1A, B).

### Increased complexity of genetic testing

As genetic testing is increasingly used throughout the healthcare system, healthcare providers will require an understanding of the complexities of genetic testing for decision-making. As



**Fig. 1.** Growing trend in genetic testing. (A) Number of patients who underwent germline genetic testing in Korea between 2016 and 2019. (B) KNHIS reimbursement for genetic testing for inherited disorders between 2016 and 2019. The karyotyping and microarray were categorized as chromosome tests, whereas special chromosome analysis for fragile X and chromosome breakage were categorized as other chromosome tests. The data were retrieved using the Electronical Data Interchange code from <http://opendata.hira.or.kr/>. Abbreviations: KNHIS, Korean National Health Insurance System; NGS, next-generation sequencing; CMA, chromosomal microarray analysis.

**Table 1.** Data from a 2020 professional survey of the GC status in Korea conducted by the Genetic Counseling Committee of the Korean Society for Genetic Diagnostics

Variables	Participants, N (%)
GC status (N = 54)	
Providing GC	44 (81.5)
Not providing GC	10 (18.5)
Service type (N = 44)	
Outpatient clinic	19 (43.2)
Consultation	18 (40.9)
Support online or by phone	33 (75.0)
Multidisciplinary team	9 (20.5)
GC cases per month (N = 44)	
≤ 10	32 (72.7)
11–20	5 (11.4)
≥ 21	7 (15.9)
GC sessions per week (N = 44)	
Irregular	26 (59.1)
1	4 (9.1)
2	3 (6.8)
≥ 3	11 (25.0)
Time spent on pre-GC activities (N = 44)	
New patients	
< 30 min	9 (20.5)
30–59 min	15 (34.1)
60–119 min	10 (22.7)
> 120 min	5 (11.4)
Not applicable	5 (11.4)
Follow-up patients	
< 30 min	12 (27.3)
30–59 min	18 (40.9)
Not applicable	14 (31.8)
Time spent on GC (N = 44)	
New patients	
< 15 min	9 (20.5)
15–30 min	18 (40.9)
31–60 min	9 (20.5)
Not applicable	8 (18.2)
Follow-up patients	
< 15 min	13 (29.5)
15–30 min	15 (34.1)
Not applicable	16 (36.4)

(Continued to the next)

**Table 1.** Continued

Variables	Participants, N (%)
Scope of GC (N = 44)	
Mendelian disorders	28 (63.6)
Cancer genetics	28 (63.6)
Family test	26 (59.1)
Genetic predisposition (genetic susceptibility)	19 (43.2)
Prenatal genetics	17 (38.6)
Other genetic disorders	12 (27.3)
GC practitioner qualification (N = 54)	
Genetics and disease-related profession	35 (64.8)
Interpretation of genetic testing results	35 (64.8)
Psychological and emotional support provision	28 (51.9)
Disease management	22 (40.7)
Challenges and concerns regarding GC (N = 54)	
Lack of financial support	30 (55.6)
Collaboration with other specialists for disease management	9 (16.7)
Effort load for acquiring latest medical scientific knowledge	3 (5.6)
Lack of consensus about standard GC	3 (5.6)
Lack of specialized training program	1 (1.9)

Abbreviation: GC, genetic counseling.

complex genetic testing usage progresses toward genomic or epigenomic levels, the burden on healthcare providers further increases. Accordingly, the need for clinical geneticists has increased. The clinical genetics workforce will likely encounter a greatly elevated demand for its services because it is difficult not only to select the most appropriate and cost-effective test, given that genetic tests become more complex, but also to interpret test results, such as those of variation of uncertain significance (VUS) and clinical significance reclassification, or secondary results [2, 3]. Many clinical geneticists find that VUS is quite difficult to explain, and many patients may not be able to grasp the concept. There are concerns regarding the potential negative repercussions of non-clinical geneticists misinterpreting the significance of VUS [4].

### Increased GC demand

Patients and non-clinical geneticists depend on a specialized professional to provide appropriate GC and education before and after testing [5]. According to a professional survey in 2020 conducted by the Genetic Counseling Committee of the Korean Society for Genetic Diagnostics, the number of institutions providing GC increased from 25% (13/52) in 2018 to 81% (44/54)

in 2020, and the numbers of GC cases and GC sessions have also increased compared with those in 2018 [1]. The scope of GC and the targeted disease categories have also expanded (Table 1).

Apart from privacy concerns, when describing the genetic test results to a patient, a GC practitioner must deal with the complex and often unanticipated psychological and informational impacts of genetic testing. In addition, GC involves activities of varying complexity, ranging from single consultations for diagnosis and counseling to clinical and laboratory investigations of other family members. Of the respondents to the professional survey in 2020, 51.9% answered that dealing with these complex aspects professionally is an important responsibility of a GC practitioner.

### Lack of preparedness of GC for genetic medicine

Non-clinical geneticists have insufficient knowledge of genetics and are generally unprepared to counsel their patients regarding genetic testing results [6]. The combination of a lack of experience and the increasing complexity of genetic testing can lead to negative outcomes in patients and their families [7]. According to the Organization for Economic Co-operation and Development Reviews of Public Health South Korea, while the genomic medicine field is growing rapidly, there is no clinical geneticist specialization in Korea [8]. Although the wide adoption of genetic medicine has been implemented by the Korean government, the establishment of programs that inevitably derive from GC is necessary to ensure that the service is safe, qualified, and cost-effective. Many countries have taken steps to focus on the unmet need for GC and increase genetic testing to promote the profession of GC practitioners [9-11]. A national plan to accommodate GC in the Korean healthcare system needs to be developed promptly and thoroughly by representatives of genetic practitioners, the health system, and the health insurance system.

## CURRENT ISSUES

### Financing of GC

The absence of facilitating factors for the easy implementation of GC models, insufficient availability of qualified personnel, and dearth of specialized centers are barriers to appropriate GC provision. Further, GC practitioners or centers struggle due to a lack of financial support, which is mainly influenced by the KNHIS policy.

In professional surveys in 2018 conducted by the Genetic Counseling Committee of the Korean Society for Genetic Diag-

nostics, most respondents reported that they spent 30–59 minutes on pre-visit activities, such as reviewing medical records or literature prior to a patient's initial visit, and <30–59 minutes on follow-up visits, whereas the mean duration of in-person visits is generally 6.2 minutes in outpatient clinics in Korea [12]. In addition to the huge workload, compared to other specialists, GC practitioners require specialized training to address the medical, psychological, and familial implications of patients with genetic diseases [13]. A team approach may be required to deliver a sufficiently useful and effective service, as the management of genetic conditions can vary and is difficult to generalize [14]. Given these peculiar aspects of GC, the training and employment of professionals would require enormous investments of time, expertise, and money, which are impossible without adequate financial support.

In our survey, 55.6% of the respondents answered that they had difficulty in opening or maintaining a clinic due to a lack of financial support. This indicates that the availability of genetic specialists and specialized centers has not kept pace with the increase in genetic discoveries and demand for genetic testing and counseling services due to financial issues. However, this survey indicated that 81.5% of respondents are providing GC regardless of the current challenges to meet the clinical demands. Nonetheless, currently, it is impossible to provide high-quality services consistently.

### National regulation governing GC practice

The barriers and facilitating factors indicated by the respondents are related to not only financial support but also national healthcare system legislation. The main issue that GC professionals face concerning GC provision is the lack of a national legislation or policy for the integration of GC in the overall healthcare system.

As national certification is not available, the genetic counselor profession is currently unregulated in Korea. As currently there is no legal definition or regulation for GC, non-geneticists claim to be geneticists, and many healthcare professionals practice GC without proper training.

Standards defining training, practice, and registration requirements for genetic counselors have been developed in Canada by the Canadian Association of Genetic Counsellors and in the United States (US) by the Accreditation Council for Genetic Counseling [9]. Core competencies for genetic counselors have also been defined in Europe and Australia [9, 15, 16]. Certifications of national associations are important to guarantee the proper provision of GC by qualified healthcare providers. National regu-

**Table 2.** Regulation states of GC in the US, Canada, UK, EU, and Australia/ New Zealand

Country	National or state regulation	Type of professional regulation	Accredited training programs	Requirements	Professional GC organizations for licensure and regulation
US	Yes (22 states with regulation)	Certification	Yes	Accredited training program; case log record	American Board of Genetic Counseling
Canada	None	Certification	Yes	Accredited training program; case log record; references	Canadian Association of Genetic Counseling
UK	Yes	Registration	Yes	Accredited training program; post-graduate work experience; case log record; references	Genetic Counsellor Registration Board
EU	Yes	Registration	Yes	Accredited training program; post-graduate work; case log record; references	European Board of Medical Genetics
Australia/ New Zealand	In progress	Certification	Yes	Accredited training program; post-graduate work experience; case log record; academic publication	Human Genetics Society of Australasia

Abbreviations: GC, genetic counseling; US, United States; UK, United Kingdom; EU, European Union.

lations currently only exist in the United Kingdom, Norway, Israel, Saudi Arabia, and South Africa, with projects to implement national regulations underway in Australia and New Zealand. State-level regulation exists in 22 states in the US (Table 2) [10].

There is a need for a unified approach for the appropriate regulation of quality and competence in GC provision. The adoption of practice standards can harmonize differences in GC education and practice among institutions. Although attempts are made in Korea, they are not driven by the government or organizations that have been fully delegated the authority of the government. As the standards are only asserted by academic societies or expert groups, expert groups have not reached a consensus.

## STANDARDIZATION OF GENETIC COUNSELING PRACTICE

All regulations and policies should be designed based on standardized GC guidelines. In a professional survey in 2020, 5.6% of the respondents pointed out this issue. Standardization is essential because it improves the quality of practice by reducing potential errors and variability in clinical outcomes. Given the differences in the medical system and social background between Korea and other countries, it is necessary to adopt national standards for GC, which are internationally compatible. To standardize GC optimized for Korea, expert groups must reach a consensus considering the characteristics of the Korean medical system and medical resources. This may lead to increased acceptance and prompt implementation of the new standards nationwide.

## FUTURE PERSPECTIVES

### Shortages and insufficient GC capacity to meet the growing demand

It is unclear how the GC workforce will be able to meet the growing demand. The recent trend in personalized medicine has many implications. In the near future, the GC workforce will face demands from patients and their families, non-clinical geneticists, and the public. Lack of professional resources in medical genetics is the most frequently cited critical issue [10].

The implementation of GC in predictive genetic testing requires proper interpretation of test results to understand their implications for the patient. This requirement needs to be swiftly addressed given the critical issues related to the provision of direct-to-consumer services, such as lack of access to qualified counseling and proper interpretation of test results. The integration of GC into population screening programs has not yet been widely implemented. Such integration would enhance referrals to GC and facilitate the identification and follow-up of patients at a high risk of inherited disorders [17].

### Strategic planning and implementation of a GC model by expert groups

To allow the best possible practice, a unified Korean-based registration for individuals qualified in medicine who work in human genetics diagnostics needs to be established. Similar efforts have been made by the European Society, which established core competences and a qualification registration schema [18]. As a part of these efforts, the Korean Society for Genetic Diagnostics established the Genetic Counseling Committee in

2015. The committee is composed of GC experts, holds an annual GC workshop to foster knowledge and good practices in GC, and monitors changes in GC nationwide by surveying the status of GC in medical fields. With the ongoing education for qualified GC training throughout academic societies in Korea, concerted efforts are highly recommended [1, 19, 20].

### Establishment of guidelines applicable to the Korean policy of national health insurance and legal requirements

The main factor in current GC delivery models in Korea is clinical demand, and the main barriers are inadequate financial support and lack of legislation. Since Korea has a consistent national health insurance policy and has instituted legal requirements for domestic medical personnel, it is necessary to establish regulations related to GC. The regulatory definition of GC needs to contain a detailed definition along with the aims of GC, scope, content, quality assessment, and education. Based on this definition, financial support policies need to be designed considering the workload and expertise. Various proposals have been suggested and are being developed by academic associations.

Despite the fast and promising development of GC practice, there are concerns about how to ensure high standards [18, 21]. Clear guidelines for best practice will ensure that GC provision develops in a way that is beneficial to its customers, health professionals, and the public [22]. Before implementation in clinical practice, GC should be evaluated and assessed based on available data on their efficacy and cost-effectiveness [11]. Clinical pathways should be monitored to reduce inappropriate GC provision and ensure high-quality standards.

GC monitoring should be based on standardized GC guidelines, and ideal guidelines should include (1) regulations, essential education, and work experience requirements for GC professionals, (2) standardization for qualified GC based on the consensus of expert groups, (3) financial support and legal protection for the provision of GC, and (4) social and ethical issues related to genetic testing and GC.

### CONCLUSION

We presented survey responses of professional clinical practice personnel to describe the different aspects of and issues with GC provision in Korea. The survey data suggest that the integration of genetic testing in the national healthcare system of Korea is still in an early phase. Despite the growing demand, there are currently few legislative frameworks in Korea that specifically

target GC and related services. Certification of medical and non-medical GC staff trained in genetics through national and international associations should be mandatory. The implementation of GC should be governed by appropriate legislative frameworks that can ensure quality by setting standards, evaluating performance, and monitoring service outcomes. Appropriate financial support for the current insurance system is of critical importance. A detailed approach involving professional organizations working with government agencies is necessary to ensure that GC and related services maintain a high quality. Thus, an ideal GC model requires a good level of genetic knowledge, adequate funding, public policies, and public understanding of genetics and genomics applications.

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### AUTHOR CONTRIBUTIONS

Kim N summarized the data and wrote the paper. Kong S-Y and Yoo J designed the study and advised on the study methodology. Kim D-H and Seo SH critically revised the study. Kim J performed the study and supervised the entire study. All authors read and approved the final manuscript.

### CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

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