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ARTICLE

Bibliometric analysis of genetic counseling publications in Asia: Insights and implications

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ABSTRACT

Purpose: Investigation of genetic counseling-related published papers offers a historical assessment of the cumulative scientific knowledge produced by members of the profession and can be the basis for future practice, training, and research. This paper aims to present a bibliometric analysis of genetic counseling publications in Asia.

Methods: We conducted a bibliometric analysis of genetic counseling-related manuscripts published in Asia from 1947 to 2023. We excluded articles published in 2024 given an incomplete year of data source. The articles were retrieved through the Scopus database using the search terms "genetic counsel*" OR "genomic counsel*" in the article titles. The bibliographic information was downloaded and analyzed descriptively through Microsoft Excel. Network visualization was done through VOSViewer.

Results: A total of 449 genetic counseling-related publications authored by at least one researcher from Asian countries were identified. The most common publication type was original articles (332, 74%) and a total of 299 manuscripts were published from 2012 to 2023, representing 66.5% (299/449) of total publications. Among Asian countries, India had the highest number of publications accounting for 19.4% of the total (n = 87) and publications from Israel had the most citations (n = 1882). Out of the 29 Asian countries represented in the document corpus, 15 have links with other Asian countries. The most common keywords used are *genetic counseling, prenatal diagnosis, genetic counselling, genetic testing,* and *genetics.*

Conclusion: There is an overall increase in the number of genetic counseling publications authored by at least one researcher affiliated with an Asian institution. This increase has corresponded to various developments in genetic counseling in the continent and is possibly driven by collaboration between and among Asian researchers and other researchers outside of Asia. The analysis of keywords also shows the evolution of topics of genetic counseling publications which also corresponded to the development of genetic counseling as a profession in the region.

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Introduction

The field of genetic counseling has experienced significant growth as a profession on a global scale over the past three decades.[1] As of 2018, there are approximately 7,000 genetic counselors worldwide, with an estimated 2,600 of them trained or working outside of the United States and Canada.[1,2] The expansion of the profession is accompanied by a steady increase in the number of publications related to genetic counseling from its inception in 1947 to the present.[3] This reflects the evolution of both the knowledge base and the practice of genetic counselors which entailed a shift of focus from the prevention of disability in 1947 to 1980 to adopting a more psychosocial-focused approach in the 1980s to 1990s.[4] More recently, genetic counselors have expanded their roles beyond the traditional medical genetics clinics into various clinical settings,[4,5] in the laboratory,[6] in public health,[7] and research.[8] These changes in the profession are influenced by a complex interplay of social, cultural, historical, regional, and technological factors.[4] Consequently, despite its origins in the United States in 1947 when Sheldon Reed coined the term 'genetic counseling,'[9] the current knowledge base of the profession has been enriched by the collective efforts and diverse perspectives of genetic counselors worldwide.

To better inform members of the profession on the advances and developments in the field, it is crucial to investigate published papers on genetic counseling as they represent the cumulative scientific knowledge that underpins both practice and research. One approach to do this is through a bibliometric analysis. Unlike meta-analyses or systematic literature reviews, which are typically applied to smaller datasets or specific research scopes, bibliometric analysis involves the examination of a large dataset with a broader focus.[10] Such an analysis can offer insights into the evolution of a scientific field, identify areas where further research is needed, and set direction for future inquiry.[10]

Notably, a recent bibliometric analysis conducted by Zakaria et al[3] covered genetic counseling publications from 1952 to 2021, highlighting the growth of the field parallel with the development of genomic medicine. Their analysis also identified key research themes within the period of their search.[3] Additionally, a content analysis of research articles published in the Journal of Genetic Counseling from 2011 to 2017 by Wallgren et al[11] provided insights into the characteristics of published studies including data analysis approaches, study samples, and topics investigated. While these studies have shown global trends and gaps in genetic counseling research[3] and those published in a specific journal,[11] none have specifically analyzed publications originating from Asia or those authored by a researcher affiliated with an Asian institution.

Genetic counseling is an emerging profession in Asia with nearly 500 clinically practicing genetic counselors estimated in 2018.[1] There are also 29 genetic counseling training programs established as of 2018 across 10 countries in Asia as well as nine in-country (i.e., in Japan, Taiwan, the Philippines, Malaysia, Indonesia, India, Israel, Saudi Arabia, and Singapore) and two regional genetic counseling professional societies (i.e., Professional Society of Genetic Counselors in Asia and the Arab Society of Genetic Counselors).[1,2,12] An analysis of the trend in genetic counseling publications in Asia can offer valuable insights into the context and significant events leading to the emergence of genetic counseling as a field in this region.[4] Furthermore, it can provide preliminary insights into knowledge gaps, serving as a foundation for setting a research agenda in the field of genetic counseling in Asia.[13]

The purpose of this paper is to present a bibliometric analysis of genetic counseling publications authored by researchers from Asia. The analysis is guided by the following questions:

1. What is the distribution of publications in this field from inception in 1947 to 2023?
2. Which countries in Asia and journals have been most prolific in publishing genetic counseling research?
3. To what extent do Asian countries collaborate in producing publications on genetic counseling?
4. Which publications have received the highest number of citations, and who are the leading authors in the field?
5. What are the most frequently studied topics or concepts as evidenced by the author keywords used in publications?

Materials and Methods

We conducted a bibliometric analysis following the guidelines outlined by Donthu et al.[10] Though framed in the context of business research, the guidelines authored by Donthu et al[10] have also been used in recent health-related bibliometric analyses.[14,15] We chose bibliometric analysis over other review methods such as scoping or systematic review because our aim was to provide a broad overview of scientific production in genetic counseling in Asia by examining various research constituents including authors, countries, institutions, and topics.[10,16] Similar to other previous bibliometric analyses,[3,14,17,18] the data we used for this study were gathered from a search of the Scopus database. The choice of the electronic database is a key decision point in undertaking a bibliometric analysis since each database has varied journal coverage.[10] In addition, using data from a search of multiple databases, while possible, necessitates consolidating metadata which is challenging because each database has its own bibliometric formatting.[10] Doing so may introduce human error which may affect the validity of the results.[10] It is in this context that Donthu et al[10] have recommended the use of only one appropriate database to mitigate the need for consolidation. Furthermore, the software we used for analysis (i.e., VOSViewer) can only currently support analysis of data from a single

database.[19] In this study, we chose Scopus because it is regarded as one of the most comprehensive bibliographic databases globally.[20]

Similar to a previous bibliometric analysis of genetic counseling publications worldwide,[3] our search terms included "genetic counsel*" OR "genomic counsel*" in the article title. Our search was limited to publications in English since this is the only language that is currently supported by VOSViewer.[21] We also limited the search to publications that are affiliated with countries and territories in Asia including India, China, Israel, Japan, Turkey, Taiwan, Hong Kong SAR, South Korea, the Philippines, Iran, Saudi Arabia, Malaysia, Indonesia, Singapore, Thailand, Qatar, Pakistan, Russian Federation, United Arab Emirates, Bangladesh, Lebanon, Jordan, Georgia, Vietnam, Sri Lanka, Kuwait, Iraq, and Bahrain. Except for erratum, we retrieved and analyzed all types of articles published from 1947 to 2023. We excluded documents published in 2024 since data for that year was not yet complete. The full search string which includes search limiters for language, publication date, and country of affiliation is found in Supplemental Table 1.

With guidance from health science librarians, the first author performed the initial search on 2 September 2023 and an updated search on 6 May 2024 to include documents published in 2023. The initial and updated search yielded a total of 408 and 449 documents, respectively. We exported the bibliographic data, citation information, and author keywords of these publications in a comma-separated values (CSV) file. To ensure data accuracy, we opened this file in Microsoft Excel, verifying that it corresponded to the number of retrieved publications and confirming that there were no missing, erroneous, or duplicate data.[10] Both the first and second authors screened and cleaned the document corpus.

In this paper, we report the results of the analysis of the updated search. We employed two analytical approaches: performance analysis and science mapping. Performance analysis entailed a descriptive examination of all retrieved publications.[10] In this study, we analyzed variables such as the number of publications per year, the types and sources of publications, their authors, and countries and territories of author affiliations through the 'analyze report' function of Scopus. Figures were generated through Microsoft Excel. For science mapping, we used VOSViewer (version 1.6.19)[22] to create visual representations of relationships among countries and author keywords. Science mapping was essential in analyzing the structural connections among the retrieved publications.[10] Specifically, we used co-authorship analysis of countries to examine the interaction and collaboration among scholars in the field of genetic counseling in Asia.[10] We used the *links* metric in VOSViewer to determine the number of co-authorship links between countries. We also used co-occurrence analysis of author keywords to examine the topics discussed in the publications. We used both network and overlay visualizations in VOSViewer to identify the main themes of publications and the usage of author keywords across the years, respectively. This analysis allowed us to determine notable "keywords" that may represent past and current topics examined by researchers and to derive insights into future trajectories of inquiry.[10]

Results

Publication output

Our Scopus database search yielded 449 publications. The most common publication type was original articles (332, 74%) followed by review articles (43, 10%). Letters to the editor, conference papers, book chapters, editorials, notes, and short surveys account for 17% of the remaining publication types. The earliest article we retrieved dated back to 1972. From 1972 to 1997, the annual number of publications remained in the single digits ranging from 0 to 7 per year. However, starting in 1998, there was a steady increase in the number of publications with peaks observed in 1998, 2006, 2010, and 2011. Subsequently, there was a significant increase in publications from 2012 to 2023, during which a total of 299 documents were published (Figure 1).

Publication by countries

Sixty-three (63) countries contributed to genetic counseling publications in Asia. Among Asian countries, India had the highest number of publications accounting for 19.4% of the total (n = 87). It was followed by China (70, 15.6%) and Israel (66, 14.7%) (Table 1). Publications from Israel have the most citations (n = 1882), followed by publications from India (n = 530) and Saudi Arabia (n = 514). In terms of average citations, publications from Saudi Arabia have the highest (42.83) followed by publications from Singapore (35.63) and Malaysia (32.70).

Co-authorship analysis of countries shows that among the 29 Asian countries represented in the document corpus, only 15 have links with other Asian countries (Figure 2). India and the Philippines each have links with 12 Asian countries, while Singapore, Taiwan, and Malaysia each have links with 11 Asian countries (Table 1). In terms of total links including with countries outside of Asia, Israel has the most (n = 41), followed by the Philippines (n = 32), India (n = 29), Taiwan (n = 29), Malaysia (n = 28), and Singapore (n = 28) (Table 1 and Figure 3). The country outside of Asia that has the most links is the United Kingdom (n = 49), followed by Canada (n = 45) and the United States (n = 42) (Supplemental Table 2). Notably, publications from Turkey, Iran, Pakistan, and Qatar did not have any links with other Asian countries but had links with countries outside of the continent (Table 1).

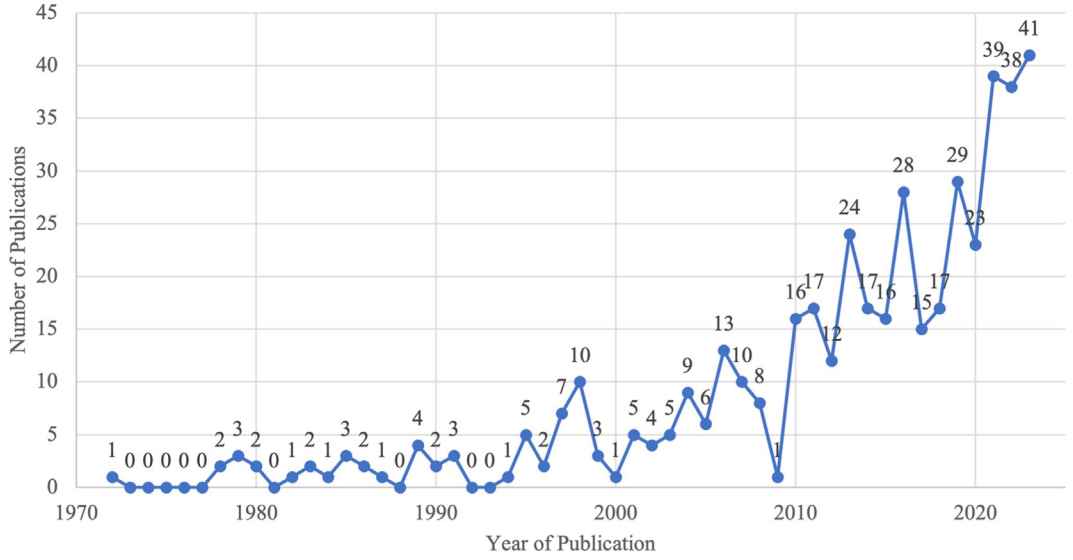

Figure 1 **Number of genetic counseling publications in Asia per year from 1972 to 2023**.

Publications by journal

Two hundred forty (240) journals have published at least one article on genetic counseling co-authored by at least one researcher affiliated with an Asian institution. The journals that have published the most articles in this area are the Journal of Genetic Counseling ($n = 49$), Taiwanese Journal of Obstetrics and Gynecology ($n = 17$), Genetic Counseling ($n = 10$), Genetics and Molecular Research ($n = 9$), Journal of Community Genetics ($n = 9$), the Indian Journal of Pediatrics ($n = 8$), and Molecular Cytogenetics ($n = 8$). Among the journals that have published the most articles (Table 2), the European Journal of Human Genetics has the highest Impact Factor (5.2) while the Journal of Genetic Counseling has the most citations ($n = 519$).

Publications by institutions

The Asian institutions with the most publications related to genetic counseling (Table 3) are Tel Aviv University, Israel ($n = 20$), the University of Hong Kong ($n = 16$), Sanjay Gandhi Postgraduate Institute of Medical Sciences, India ($n = 16$), and the University of the Philippines Manila ($n = 14$). Among these institutions, publications from Chaim Sheba Medical Center Israel have the most citations with 927 followed by Tel Aviv University, Israel

Table 1 Citation and co-authorship analysis of publications on genetic counseling from Asian countries

Rank	Country	Record	Citations	Average Citations	Links with Asian Countries Only	Total Links with Other Countries
1	India	87	530	6.09	12	29
2	China	70	477	6.81	8	13
3	Israel	66	1882	28.52	8	41
4	Japan	55	363	6.6	2	13
5	Turkey	29	163	5.62	0	6
6	Taiwan	28	360	12.86	11	29
7	Hong Kong SAR	20	122	6.1	8	15
8.5	Philippines	18	489	27.17	12	32
8.5	South Korea	18	337	18.72	8	24
10	Iran	15	67	4.47	0	1
11	Saudi Arabia	12	514	42.83	7	24
12	Malaysia	10	327	32.7	11	28
13	Indonesia	9	91	10.11	8	14
14	Singapore	8	285	35.63	11	28
15	Thailand	7	72	10.29	8	13
16	Russian Federation	6	127	21.17	2	19
17.5	Pakistan	5	32	6.4	0	1
17.5	Qatar	5	45	9	0	5

Note: This table only includes countries with a minimum of 5 documents. The column "Links with Other Asian Countries" and "Total Links with Other Countries" indicate the number of co-authorship links of researchers from the specific country to other countries. The "Total Links with Other Countries" indicates links with both Asian and non-Asian countries.

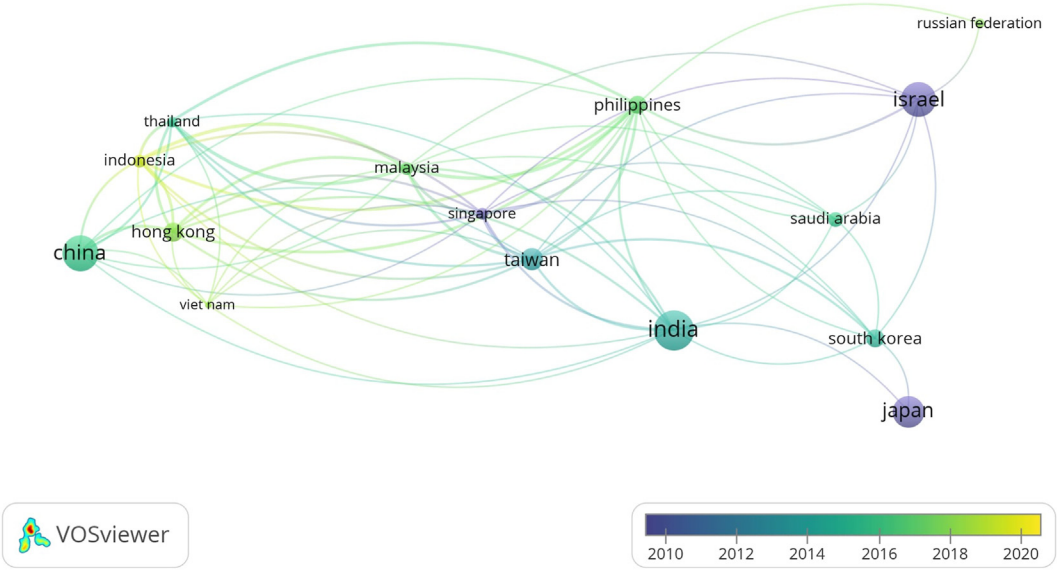

Figure 2 Overlay visualization of collaboration among Asian countries. The color gradient shows the average year of publication of documents emanating from a specific country. The size of the circle is directly proportional to the number of documents contributed by the country. The thickness of the line corresponds to the strength of the connection between countries.

(n = 902) and Hadassah University Medical Center, Israel (n = 803).

Publications by author

The most productive authors who have publications related to genetic counseling in Asia are from Taiwan, the Philippines, Hong Kong, Israel, China, India, and Malaysia (Table 4). The most productive authors are Chen CP (Mackay Memorial Hospital, Taiwan) and Laurino MY (University of the Philippines Manila) with 12 publications each followed by Shiloh S (Tel Aviv University, Israel), Wang W (Mackay Memorial Hospital, Taiwan), Chung BHY (The University of Hong Kong) and Zayts O (The

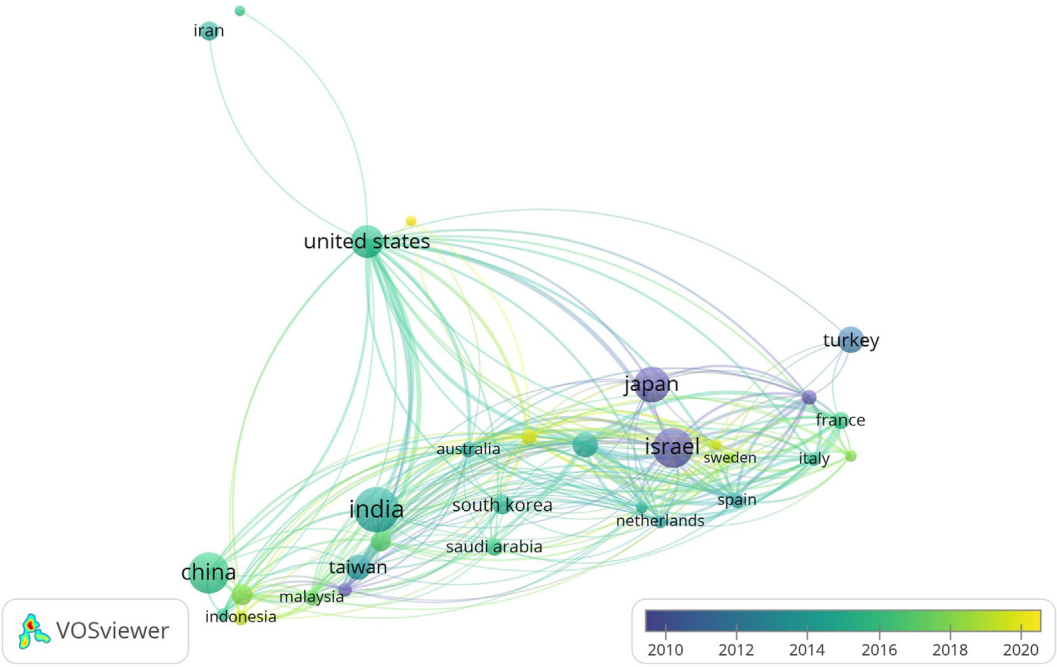

Figure 3 Overlay visualization of international collaboration of countries represented in the document corpus. The color gradient shows the average year of publication of documents emanating from a specific country. The size of the circle is directly proportional to the number of documents contributed by the country. The thickness of the line corresponds to the strength of the connection between countries.

Table 2 Journals that have published articles on genetic counseling in Asia ranked by number of publications

Rank	Journal	No. of Publications	Impact Factor (2022)	Cite Score (2022)	Citations	Average Citations
1	Journal of Genetic Counseling	49	1.9	3.7	519	10.59
2	Taiwanese Journal of Obstetrics and Gynecology	17	2.1	3.1	84	4.94
3	Genetic Counseling[a]	10	0.14[b]	0.4[c]	38	3.8
4.5	Genetics and Molecular Research	9	0.4	1.2	76	8.44
4.5	Journal of Community Genetics	9	1.9	3.5	71	7.89
6.5	Indian Journal of Pediatrics	8	4.3	6.7	64	8
6.5	Molecular Cytogenetics	8	1.3	3	16	2
8.5	Clinical Genetics	7	3.5	7.4	174	24.86
8.5	Frontiers in Genetics	7	3.7	5.2	32	4.57
10.5	European Journal of Human Genetics	6	5.2	9.1	275	45.83
10.5	International Journal of Human Genetics[a]	6	0.1	0.6[d]	4	0.67

[a]Discontinued in Scopus in 2016.
[b]Impact Factor data as of 2017.
[c]Cite Score data as of 2017.
[d]Cite Score data as of 2015.

University of Hong Kong) each having 10 publications. Among the productive authors, the most cited is Shiloh S (Tel Aviv University, Israel) with 687 citations, followed by Laurino MY (University of the Philippines Manila, 240 citations), Thong MK (University of Malaya Medical Center, Malaysia, 127 citations) and Zayts O (The University of Hong Kong, 123 citations). Since the number of author citations is a function of the length of time an author is active in research, we have also computed for each author's m-index. The m-index is a time-adjusted author-level metric calculated by dividing the author's h-index by the number of years since their first publication.[23] This, however, considers all of the author's publications and not just those about genetic or genomic counseling.[23] When the m-index is considered, Chung BHY has the highest (1.91) followed by Chen CP (1.06) and Wang W (0.92).

Top cited publications

Table 5 shows the 10 most cited publications in genetic counseling that have at least one author from an Asian institution. Three of the most cited publications are by the American Journal of Medical Genetics. Considering that the number of citations is a function of the length of time since publication, we also analyzed the relative citation ratio (RCR) of these 10 articles. Derived from data from PubMed, the RCR is an article-level metric that uses data from the article's co-citation network to field- and time-normalize the number of citations it receives.[24] As seen in Table 5, rank discrepancies are apparent when the RCR is used compared to the total citations.

The topics discussed in the top-cited articles are varied. Four articles were about concepts integral to genetic counseling such as risk perception, satisfaction, and perceived personal control, and how to measure them.[25-28] Three articles explored disease-specific genetic counseling considerations[29-31] and two articles were about the professional development of genetic counseling globally,[1,2] and one article was about an evaluation of a disease-specific genetic counseling service.[32]

Keyword visualization

The co-occurrence analysis of author keywords used in publications related to genetic counseling in Asia is shown in Figures 4 and 5. The co-occurrence analysis shows the

Table 3 Leading institutions in Asia that published articles on genetic counseling ranked by number of publications

Rank	Institution	Country/Territory	No. of Publications	Citations	Average Citations
1	Tel Aviv University	Israel	20	902	45.1
2.5	The University of Hong Kong	Hong Kong	16	149	9.31
2.5	Sanjay Gandhi Postgraduate Institute of Medical Sciences	India	16	67	4.19
4	University of the Philippines Manila	Philippines	14	261	18.64
5.5	Hadassah University Medical Center	Israel	13	803	61.77
5.5	China Medical University	Taiwan	13	277	21.31
5.5	Mackay Memorial Hospital Taiwan	Taiwan	13	94	7.23
8	Asia University	Taiwan	12	85	7.08
9	Chaim Sheba Medical Center Israel	Israel	11	927	84.27
10	National Yang-Ming University Taiwan	Taiwan	10	74	7.4

Table 4 Leading authors who published on genetic counseling in Asia ranked by number of publications

Rank	Author	Country of Affiliation	Records	Citations	Ave Citations	h-Index	m-Index
1.5	Chen, C.P.	Taiwan	12	85	7.08	36	1.06
1.5	Laurino, M.Y.	Philippines	12	240	20	15	0.79
2.5	Shiloh, S.	Israel	10	687	68.7	26	0.72
2.5	Wang, W.	Taiwan	10	55	5.5	24	0.92
2.5	Chung, B.H.Y	Hong Kong SAR	10	56	5.6	42	1.91
2.5	Zayts, O.	Hong Kong SAR	10	123	12.3	13	0.87
7.5	Liu, R.Z.	China	8	75	9.38	18	0.86
7.5	Phadke, S.R.	India	8	47	5.88	28	0.85
9.5	Chern, S.R.	Taiwan	7	50	7.14	23	0.85
9.5	Thong, M.K.	Malaysia	7	127	18.14	26	0.9

Note: h-index is based on 2024 data reflected in the author profile in Scopus. The m-index is computed by dividing the h-index by the number of years since the author's first publication.

relatedness of author keywords based on the number of documents they occur together. There are a total of 979 author keywords used but we limited our analysis to the 74 keywords which have occurred at least three times in the document corpus. The most common keywords used in publications on genetic counseling in Asia are *genetic*

Table 5 Top cited articles published by authors affiliated with an Asian institution ranked by total citations (TC) and Relative Citation Ratio (RCR)

Rank by TC	TC	Rank by RCR	RCR	Authors	Article Title	Year of Publication	Journal	Document Type
1	365	2	7.79	Charron et al.	Genetic counselling and testing in cardiomyopathies: A position statement of the European Society of Cardiology Working Group on Myocardial and Pericardial Diseases	2010	European Heart Journal	Note
2	192	1	11.44	Abacan et al.	The Global State of the Genetic Counseling Profession	2019	European Journal of Human Genetics	Review
3	138	7	3.39	Berkenstadt et al.	Perceived personal control (PPC): A new concept in measuring outcome of genetic counseling	1999	American Journal of Medical Genetics	Article
4	136	4	4.64	Mcallister M et al.	The Genetic Counseling Outcome Scale: A new patient-reported outcome measure for clinical genetics services	2011	Clinical Genetics	Article
5	133	8	2.09	Buiting K et al.	Sporadic imprinting defects in Prader-Willi syndrome and Angelman syndrome: Implications for imprint-switch models, genetic counseling, and prenatal diagnosis	1998	American Journal of Human Genetics	Article
6	129	6	3.96	Memish & Saeedi	Six-year outcome of the national premarital screening and genetic counseling program for sickle cell disease and -thalassemia in Saudi Arabia	2011	Annals of Saudi Medicine	Article
7	105	3	5.71	Ormond et al.	Genetic counseling globally: Where are we now?	2018	American Journal of Medical Genetics, Part C: Seminars in Medical Genetics	Note
8	105	5	4.03	Shiloh S et al.	Satisfaction with genetic counseling: Dimensions and measurement	1990	American Journal of Medical Genetics	Article
9	90	9	1.48	Köhler et al.	Androgen insensitivity syndrome: Somatic mosaicism of the androgen receptor in seven families and consequences for sex assignment and genetic counseling	2005	Journal of Clinical Endocrinology and Metabolism	Article
10	84	na	nd	Shiloh S.	Perception Of Risk In Genetic Counseling	1989	Psychology & Health	Article

Note: The article by Shiloh S is not found in PubMed and thus an RCR is not computed. As mentioned, RCR takes into account only those indexed in PubMed. *na*, not applicable; *nd*, no data available.

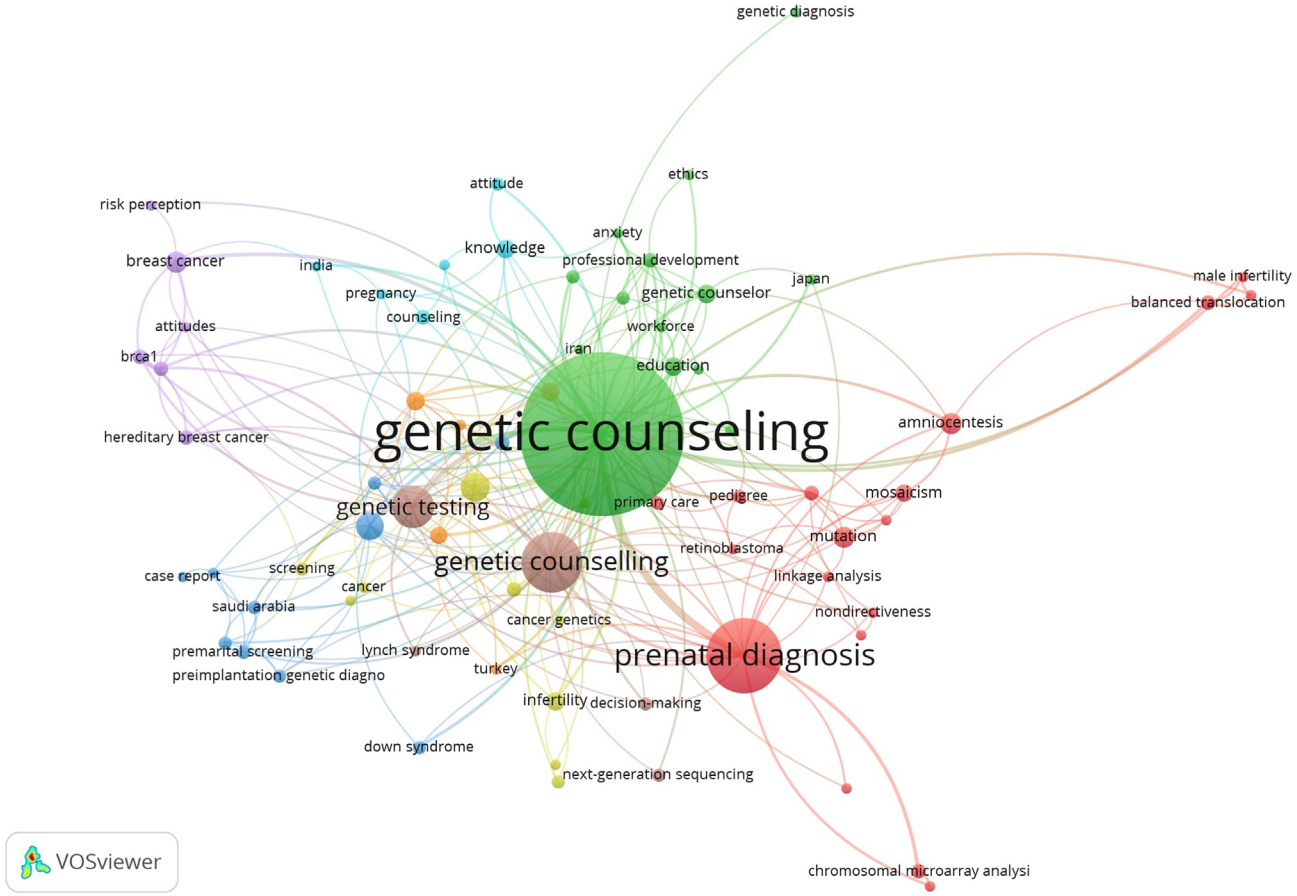

Figure 4 Network visualization of author keywords used in genetic counseling publications in Asia. The colors indicate different thematic focus based on author keywords co-occurrence analysis. The size of the circle is directly proportional to the number of publications containing the keyword. The thickness of the lines indicates the strength of the relationship between keywords.

counseling ($n = 162$), *prenatal diagnosis* ($n = 52$), *genetic counselling* ($n = 38$), *genetic testing* ($n = 23$), and *genetics* ($n = 13$) (see Supplemental Table 3). Figure 4 shows a mapping of the eight identified publication 'hotspots' based on co-occurrence analysis of author keywords. Cluster one (red) contains 18 keywords which mainly focused on prenatal testing and diagnosis. Cluster two (green) has 14 keywords pertaining to genetic counseling training, education, and professional development. Cluster three (blue) has 10 keywords that focus on consanguinity and premarital screening. Cluster four (gold) has 10 keywords that are focused on genetics, hereditary cancer, and newborn screening. Cluster five (purple) has six keywords that are focused on hereditary breast cancer. Cluster six (light blue) also has six keywords focused on attitudes and knowledge in genetics. Cluster seven (orange) has five keywords that are focused on COVID-19 and genetics services. Cluster eight (brown) also has five keywords focused on decision-making, genetic testing, and counseling.

We also analyzed the keywords used through the years and we saw changes in the topics in genetic counseling publications in Asia from 2005 and earlier to 2023 (Figure 5 and Supplemental Table 3). The keyword *risk perception* was used most commonly by publications in 2005 or earlier. From 2006 to 2010, the most common keywords used were *mutation, primary care, India, Israel, linkage analysis, non-directiveness,* and *retinoblastoma*. From 2011 to 2015, publications have commonly used the following keywords: *genetic counseling* or *genetic counselling*, *prenatal diagnosis, consanguinity, amniocentesis, breast cancer, hereditary breast cancer, mental retardation, Pakistan, ethics, genetic, genetic diagnosis, Iran, MECP2, polymorphism,* and *Turkey*. For the years 2016 to 2020, the most common keywords are *genetic testing, genetics, education, genetic counselor/s, infertility, knowledge, Philippines, genetics services, mosaicism, balanced translocation, BRCA1/2, counseling, genetic disorders, professional development, Asia, attitude, decision-making, Down syndrome, pedigree, preimplantation genetic diagnosis, premarital screening, training,* and *translocation.* More recently from 2021 to 2023, the most common keywords are *chromosomal microarray analysis, next generation sequencing, screening, Lynch syndrome, newborn screening, noninvasive prenatal testing (*now referred to as *prenatal screening by cell-free DNA),* and *workforce.*

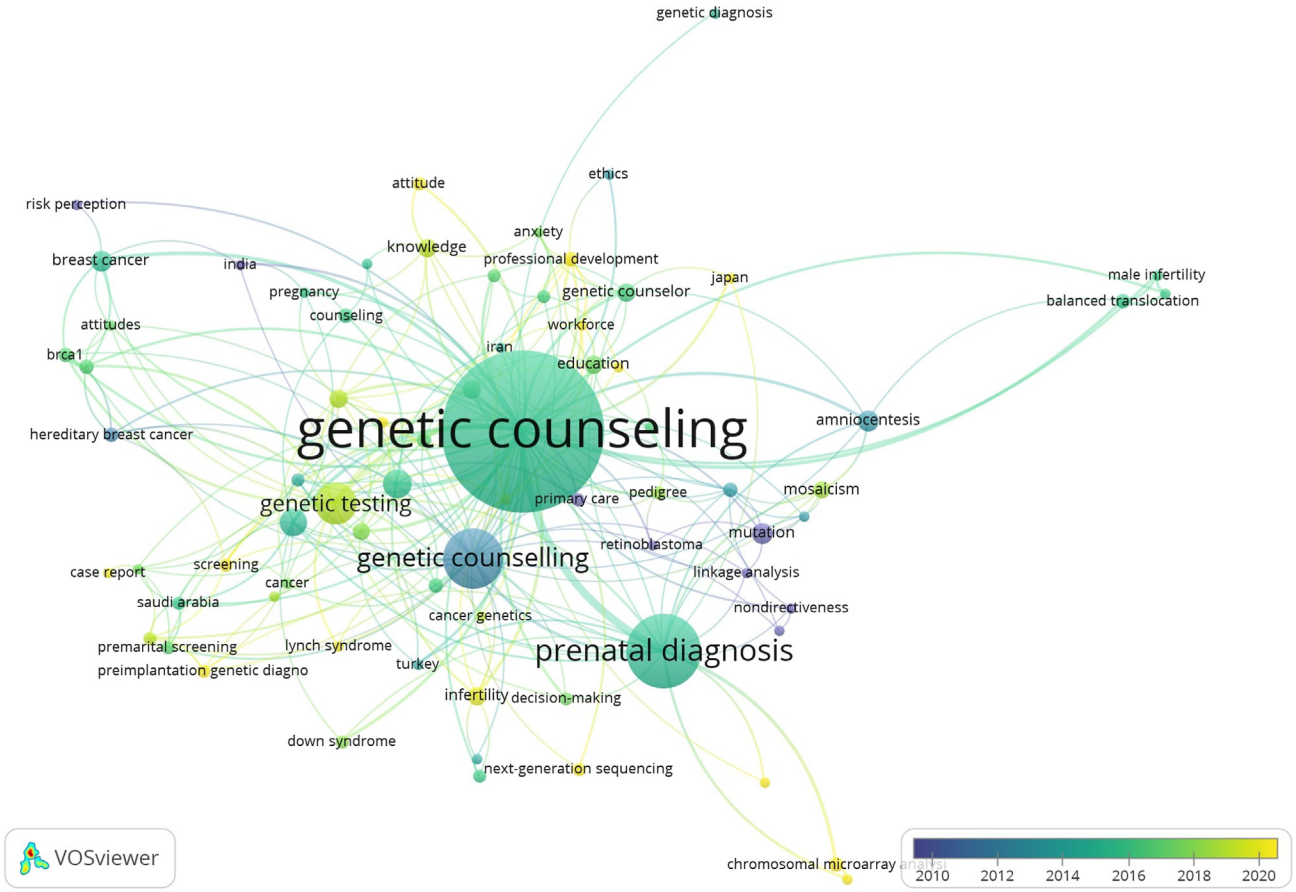

Figure 5 Overlay visualization of author keywords used in genetic counseling publications in Asia. The color gradient indicates the average year of publication of articles that used specific author keywords. The size of the circle is directly proportional to the number of publications containing the keyword. The thickness of the lines indicates the strength of the relationship between keywords.

Discussion

In this study, we have identified the trend in genetic counseling publications in Asia through a bibliometric analysis. We have shown the contributions of Asian countries and researchers from these countries in generating knowledge on genetic counseling including their collaboration within the continent and across the globe. Our results show that there is a cumulative increase in the number of genetic counseling publications in Asia from 1972 to 2023. This trend is consistent with the observed increase in the number of genetic counseling publications globally.[3]

Specifically, we observed that from a single-digit total annual publication from 1972 to 1997, the number of publications has consistently increased from 1998 to 2023 with the most significant increase from 2012. This observation can be attributed to various developments in genetic counseling in the region including the establishment of several master's programs in genetic counseling, and in-country and regional professional societies. In 1997, the first master's program in genetic counseling in Israel was established at Hebrew University – Hadassah Medical School.[33] Across the region, several training programs, mostly at the master's level, have also been established in the following countries (in parenthesis was the year the first training program started)[1,12]: Japan (2000s), Taiwan (2003), Saudi Arabia (2005), South Korea (2006), Indonesia (2006), India (2007), Hong Kong (2010), the Philippines (2011), and Malaysia (2015). These training programs have focused on preparing non-physicians to provide genetic counseling services within the context of each country's health system and population needs.[1,2]

Even prior to training programs being established, professional societies in genetic counseling were established as early as 1982 in Japan with the Japanese Society of Clinical Genetics (which was the precursor of the Japanese Society of Genetic Counseling)[34] and in 1989 in Israel with the Israel Association of Genetic Counselors.[33] Specifically, the Israel Association of Genetic Counselors was established by genetic counselors, mostly non-physicians, who were trained abroad and moved to the country.[33] Subsequently, several country professional societies have also been established including the Taiwan Association of Genetic Counselors (2008), Board of Genetic Counseling India (2015), Genetic Counseling Society Malaysia (2018), Indonesian Society of Genetic Counselors (2020),

Philippines Society of Genetic Counselors (2021), and more recently, the Singapore Genetic Counselor Society (2023).[1,12,35] Two regional societies have also been established: the Professional Society of Genetic Counselors in Asia (PSGCA, 2015)[12] and the Arab Society of Genetic Counselors (ASGC, 2019).[36] These societies were instrumental in creating an avenue for genetic counselor collaboration and networking as well as the exchange of resources (e.g., information and tools) which may eventually impact patient care.[12,13,36]

Overall, the increase in the number of publications through the years is an indication of an active knowledge production in genetic counseling in this region. This is important especially when we consider the need for context-specific evidence to support the praxis of genetic counseling including but not limited to exchanging best practice guidelines, defining the critical elements of genetic counseling tools and resources including being culturally appropriate, and determining feasible infrastructure to support implementation of genetic counseling training programs.[13] The disseminated knowledge may help inform further insights in addressing cross-cutting issues in Asia such as unequal access to and provision of genetic counseling services in the context of highly diverse sociocultural and health systems across the continent.[13]

Our results also show that the Asian countries with the most publications related to genetic counseling are those with a significant presence of genetic counselors and where genetic counseling training programs have been in existence for several years. Except for Thailand, Pakistan, and the Russian Federation, all the countries presented in Table 1 have been reported to have practicing genetic counselors in various capacities.[1,12,37,38] Similarly, genetic counseling training programs exist in all countries in Table 1 except for Turkey, Iran, Pakistan, Russian Federation, Singapore, and Thailand where there are no master's level training programs reported in the literature.[1,12,39] Thailand, however, has a short course training in genetic counseling for local healthcare providers that focuses on thalassemia and Down syndrome.[12] The paper by Abacan et al[1] is a good reference on the global state of genetic counseling as they attempted to describe as fully as possible the extent of the global expansion of the profession, similar to what we have found in this paper in terms of publications and production of scientific knowledge. Ormond et al[40] has just recently updated the data from the Abacan et al[1] article to capture the changes in the genetic counseling global landscape from 2017 to 2022.

It is also unsurprising that the leading Asian institutions that have published articles related to genetic counseling have significant roles in the expansion of the field in Asia. To illustrate, the master's program in genetic counseling in the Philippines is established at the University of the Philippines Manila[35] while the Hadassah University Medical Center hosted Israel's first genetic counseling master's program.[33] The other leading Asian institutions reported in this study have either been the main referral centers or the primary training institutions for clinical genetics in their respective countries.[41] For example, the Sanjay Gandhi Postgraduate Institute of Medical Sciences Lucknow in India is the only institution in the country to train clinical geneticists for many years. This finding is expected since genetic counseling is also a recognized role in the medical specialty of clinical genetics.[41] However, this also begs the question of who writes about genetic counseling in Asia. While we have presented in Table 4 the most productive authors in the field in Asia, we did not have a reliable method to ascertain the background of these authors, whether they are genetic counselors, clinical geneticists, or from other disciplines. This matter is important especially if we consider that authors tend to write based on their worldviews and the philosophical underpinnings that they subscribe to. Moreover, this also extends the discussion on the delineation of "genetic counseling as a *practice*" and "genetic counseling as a *profession*", the latter with its own shared values, practice models, and tenets.[42]

We also noted considerable linkages between and among Asian countries and countries outside of Asia in producing publications related to genetic counseling. These collaborations are possibly enabled by the establishment of regional professional societies such as the PSGCA and the ASGC which, as mentioned, were primarily established to promote genetic counselor collaboration.[12,36] Training and research collaborations were two of the priority areas for genetic counselor professional development that were identified when the PSGCA was established in 2015.[13] Existing ties with other genetic counseling professional societies outside of Asia have also possibly driven the increase in publications seen in this study. For example, many Asian countries with genetic counseling training programs are part of the Transnational Alliance for Genetic Counseling, an international group primarily consisting of genetic counselor educators from more than 20 countries.[43] Moreover, many Asian genetic counselors, who are mostly pioneers in their own countries, have received their training elsewhere most notably in the United States, United Kingdom, and Australia.[12] It is possible that they brought their ties with their country of education as they established genetic counseling services in their country of origin. The outcomes of these collaborations do not only include joint publications but also partnerships in the training and supervision of genetic counseling students. Notable examples of this include the "sister programs" between the University of the Philippines Manila and the Stanford Genetic Counseling program[44] as well as the cross-training in genetic counseling between Diponegoro University in Indonesia and Radboud University Nijmegen Medical Center in the Netherlands.[45] These existing (and even future) collaborations are important and should be encouraged to facilitate scientific exchange and to promote a culture of research among individuals involved. This will not only benefit individual professional development, but it may also help overcome contextual and methodological challenges in conducting research, facilitate the advancement of genetic counseling as a rigorous academic area of study,[46] and

support improving patient care, especially with clinician-researcher collaboration.[47] It would be informative, however, to determine the growth of collaborative activities over time but we have limited access to such data and could be a focus of future analysis.

The keyword visualization provided insights into the topics of publications and how these topics evolved over time. In general, the topics of publications were mainly on prenatal diagnosis and screening, genetic counseling education and professional development, consanguinity and premarital screening, hereditary cancer, and genetics services and testing. These results may have been driven by both the research interests of the genetic counseling community in Asia and the healthcare needs within the region. For example, consanguinity remains a concern in many areas in Asia particularly in the Middle East[48] while hereditary cancer especially Hereditary Breast and Ovarian Cancer has been receiving much attention considering the availability of genetic screening and access to early diagnosis and treatment.[49] The trends we saw in the publication topics were consistent with the content areas of articles published from 2011 to 2017 in the Journal of Genetic Counseling in which many addressed the topic of genetic counseling services and have represented the cancer and prenatal specialties.[11] A similar trend was reported by Zakaria et al[3] when they did a bibliometric analysis of genetic counseling publications globally. However, none of the keywords analyzed have pointed to neurogenetics and psychiatry as a topical area of published genetic counseling papers from Asia. This is contrary to what was reported by both Wallgren et al[11] and Zakaria et al[3] that psychiatry is a prevalent topical area in genetic counseling papers globally and those published in the Journal of Genetic Counseling within the period of 2011 to 2017. This result may be due to the limited awareness and understanding about psychiatric genetic counseling and the inevitable differences across countries in Asia in terms of available resources which may have been diverted to other priority areas such as prenatal diagnosis, genetic counseling training, and hereditary cancer.[39]

Interestingly, the keywords on training and professional development of genetic counselors have started to emerge in publications from 2016. This time period corresponded to the maturation of several established master's training programs in genetic counseling including those in Japan, India, South Korea, Taiwan, Indonesia, and Hong Kong, and the establishment of several others especially in the Philippines and Malaysia.[1,12] This was also the time when the PSGCA was established, and several papers were published to document the emergence of the genetic counseling profession in the region.[12,13] The emergence of publications discussing genetic counseling training and professional development does not only increase awareness of the expansion of the profession, but it may also benefit countries without training programs in eventually establishing their own. It may also enable building connections among genetic counseling leaders and educators to exchange best practices in advocating and advancing the profession and in training genetic counselors.[46]

Our analysis of author keywords also provides some insights into possible topics of future research that can broaden the production of knowledge in genetic counseling in Asia. Aside from the keywords *attitude, anxiety, decision-making, knowledge, risk perception,* and *ethics*, there are no other keywords used that may point to the examination of the psychosocial aspects of providing genetic counseling. Even these keywords have been used sparingly in publications as indicated by the number of times they occurred in the document corpus. Also, apart from the keywords *genetic counseling, genetic counselling, genetic counselor,* and *genetics services*, there are no other keywords used that may indicate the examination of the implementation of genetic counseling services including service delivery models. These indicate the need for further studies that explore the psychosocial aspects as well as the processes and outcomes of genetic counseling.[46] It may be possible that publications that examined these topics used generic author keywords such as *genetic counseling*, which may partly explain why this keyword is most frequently occurring. This particular point supports the need for further content analysis to derive a more nuanced understanding of the topics of publications.

Our analysis also provides some insights into the trajectory of genetic counseling research albeit in the short-term. From the overlay visualization of keywords, we saw newer publications (from 2021-2023) about *next generation sequencing*, *noninvasive prenatal testing* (now referred to as *prenatal screening by cell-free DNA*), *newborn screening*, *Lynch syndrome*, and *workforce.* In the short term, we would expect more publications on these topics given the increasing interest in the roles of genetic counselors in the era of genome-based medicine,[5] the widespread clinical use of noninvasive prenatal testing (*prenatal screening by cell-free DNA*),[50] and the global progress in newborn screening especially among developing countries.[51] We would also expect more publications on professional development, education, and training considering the continued interest in international genetic counseling and the worldwide expansion of the profession.[1,2,52]

This bibliometric analysis has several limitations. First, our analysis was limited to articles indexed in the Scopus database. While Scopus is considered to be one of the most comprehensive electronic databases for scholarly articles,[20] there may be articles that are relevant that are not currently indexed in the database. This is especially true for national and local journals. Second, searching primarily using the article title for inclusion is another limitation. While a search through the article title only has provided a more targeted document corpus, doing so may have inadvertently excluded other relevant articles. Third, because VOSViewer does not currently support non-English articles, we limited our search to those published in English. We acknowledge that this decision may have excluded relevant articles written in other languages. Despite these limitations, this study still provided valuable insights into the trends in genetic counseling publications from Asia and those authored by at least one researcher affiliated with an Asian institution.

In conclusion, this bibliometric analysis showed an overall increase in the number of genetic counseling publications authored by at least one researcher affiliated with an Asian institution. This increase has corresponded to various developments in genetic counseling in the region, most notably the institution of master's level training programs primarily for non-physicians and the establishment of various country and regional societies. Collaboration between and among Asian researchers and other researchers outside of Asia has also contributed to this trend. This bibliometric analysis has also shown the evolution of topics of genetic counseling publications which has also corresponded to the development of genetic counseling as a profession in the region. Future research should consider conducting a content analysis of genetic counseling publications from Asia as this can provide deeper insights into topical areas explored in these articles. Such an undertaking can inform research agenda-setting for the field of genetic counseling in Asia.

Data Availability

Data is available upon request from the corresponding author.

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Ethics Declaration

This project did not collect individual-level human data, and therefore was exempt from human subjects review.

Conflict of Interest

The authors are officers of their respective in-country genetic counseling societies. Peter James Abad and Ma-Am Joy Tumulak are the current President and President-elect, respectively of the Philippine Society of Genetic Counselors (PSGC). Mercy Laurino is a past president of the Professional Society of Genetic Counselors in Asia (PSGCA). Sook-Yee Yoon is the current President of the Genetic Counselor Society Malaysia (GCSM). Qurratulain Hasan is the current President of the Board of Genetic Counseling India (BGCI). The authors did not receive any compensation in these capacities.

Additional Information

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