

## BRIEF COMMUNICATION



## Global trends and themes in genetic counseling research

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This article seeks to highlight the most recent trends and themes in genetic counseling that are of broad interest. A total of 3505 documents were published between 1952 and 2021, with a trend toward increase in paper/year. The most common documents are original articles (2515, 71.8%), followed by review articles (341, 9.7%). Journal of Genetic Counseling publishes the highest number of genetic counseling articles (587, 16.7%), followed by Clinical Genetics (103, 2.9%) and the South American Journal of Medical Genetics (95, 2.7%). Co-occurrence analysis revealed five research themes: genetic testing, cancer, genetic counselor, prenatal diagnosis, and psychiatry. The genetic counselor theme contained most of the recent keywords, including “covid-19,” “underrepresented population,” “service delivery models,” “workforce,” “disparities,” “service delivery,” “professional development,” “cultural competence,” “access,” “diversity,” “telemedicine,” and “health literacy.” Genetic counseling researchers may use these keywords to find topics pertinent to their future research and practice.

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## INTRODUCTION

The term “genetic counseling” was first coined by an American scientist, Sheldon C. Reed, in 1947 [1]. According to the Ad Hoc Committee of the American Society of Human Genetics, genetic counseling addresses human issues related to the incidence or risk of a genetic disorder in a family [2]. Seymour Kessler later defined genetic counseling as psychological contact or psychotherapy [3]. The 2006 National Society of Genetic Counselors task force described genetic counseling as helping people understand and adapt to the physical, psychological, and familial implications of genetic contributions to disease [4].

Future trends in clinical genetic and genetic services are fast evolving and have been the subject of recent publications [5–8]. However, the global situation of genetic counseling research, as well as trends and subjects of broad interest in this area throughout the last 70 years, have not been adequately covered since the focus was mainly on specific journals [5] or countries [6–8]. We, therefore, mapped genetic counseling research globally using the Scopus database and the VOSviewer tool to provide academics and practitioners in this field, information on the scientific landscape as well as important and emerging themes.

## MATERIALS AND METHODS

## Search strategy

The literature was searched and retrieved from the Scopus database on January 19, 2022. The Scopus database is regarded as one of the most significant and exhaustive collections of scientific information in the world [9]. The following search string was used in the article title: “genetic counsel\*ing”. All document types written in the English language and released between 1952 and 2021 were included, except for erratum. The retrieved documents were subjected to bibliometric analysis using VOSviewer.

## RESULTS

## Publication outputs

A total of 3505 documents were retrieved. The most common document type was original articles (2515, 71.8%), followed by review articles (341, 9.7%). Letters, book chapters and conference papers, each contributed about 4.7–4.8% of the documents. Notes, editorials, short surveys and books contributed about 4.4% in total. From 1952 to 1967, the document number was one digit. From 1968 to 2010, the number of papers increased to two digits; from 2011 to 2021, it was three digits (Fig. 1).

## Journals

The top genetic counseling journals published 1163 documents, accounting for 33.2 per cent of all publications (Table 1). The Journal of Genetic Counseling had 587 publications with a Cite-Score of 3.8 and SJR 2020 of 0.867. Only three journals, the American Journal of Human Genetics, Genetics in Medicine, and Journal of Medical Genetics, have an SJR greater than two. The Scopus database has discontinued its coverage of the two of the top journals, while the American Journal of Medical Genetics has been renamed the American Journal of Medical Genetics, Parts A, B, and C.

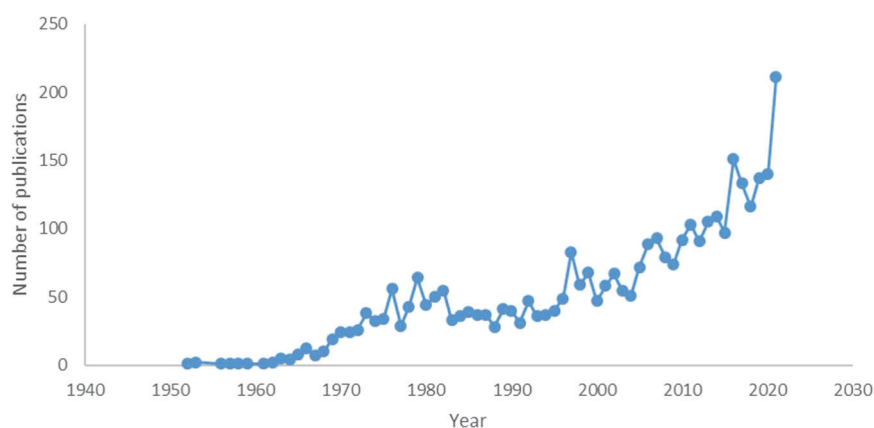
## Author Keywords

Network visualization of keyword co-occurrence map was constructed with a threshold of nine keyword co-occurrences in VOSviewer. The top keywords ranked by frequency were ‘genetic counseling’ ( $n = 1156$ ), ‘genetic testing’ ( $n = 205$ ), ‘prenatal diagnosis’ ( $n = 104$ ), ‘breast cancer’ ( $n = 97$ ), ‘genetic counselor’ ( $n = 68$ ), ‘genetics’ ( $n = 59$ ), ‘brca1’ ( $n = 49$ ), ‘education’ ( $n = 48$ ), ‘brca2’ ( $n = 45$ ), and ‘hereditary cancer’ ( $n = 44$ ).

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**Fig. 1** Trends in the number of publications from 1952 to 2021 in the field of genetic counseling.

**Table 1.** Top publishing journals on genetic counseling research.

Source Title	TP	TC	Publisher	Cite Score	SJR 2020	SNIP 2020
Journal of Genetic Counseling	587	8732	Springer Nature	3.8	0.867	0.9
Clinical Genetics	103	2346	Wiley-Blackwell	7.3	1.543	1.52
South American Journal of Medical Genetics	95	2702	Wiley-Blackwell	-1.0	1.131	1.643
Birth Defects Original Article Series <sup>a</sup>	78	442	March of Dimes Birth Defects Foundation	-1.0	0.166	-1.0
Journal of Medical Genetics	72	2627	BMJ Publishing Group	9.7	2.439	1.883
American Journal of Human Genetics	49	2359	Elsevier	16.7	6.661	2.976
Genetics In Medicine	49	1493	Springer Nature	13.7	3.509	2.73
Genetic Counseling <sup>a</sup>	47	197	Editions Medecine et Hygiene	0.4	0.155	0.347
Patient Education and Counseling	43	1103	Elsevier	4.6	1.098	1.583
European Journal of Human Genetics	40	1336	Springer Nature	6.1	1.587	1.383

TP total number of publications, TC total citations, Cite-Score average citations received per document published in serial, SJR SClmago Journal Rank, SNIP Source Normalized Impact per Paper.

<sup>a</sup>discontinued in Scopus.

To identify themes in the literature, we studied the terms in each cluster. Five clusters appear from the map in Fig. 2A with the following themes: genetic testing (red cluster), cancer (green cluster), genetic counselor (blue cluster), prenatal diagnosis (yellow cluster) and psychiatry (purple cluster). The overlay visualization of keyword co-occurrence map indicates the publishing dates when specific topics were most popular. Many newer keywords (yellow nodes) were found mainly in the blue cluster (genetic counselor theme) compared to other clusters (Fig. 2B).

## DISCUSSION

The increasing number of publications per year may indicate the development trend in the field of genetic counseling. The observed trend may be attributable to the rapid evolution of clinical genetic services [5–8]. Genetic counselors collaborate with geneticists and other medical specialists to provide genetic counseling services. Genetic counseling research, which strives to improve the quality and availability of genetic counseling services, benefits from these clinical data. Original articles contributed to a large proportion of the publication outputs and increased significantly over the last decade. This finding indicates that the field is expanding in line with the development of genomic medicine [10]. Funding for implementation and outcome research is urgently needed to expand the scientific evidence for genetic counseling practice [10].

The present analysis revealed that 1163 documents or 33.2% of studies were published in the top 10 journals. Of these, more than

half of the relevant documents were published in the *Journal of Genetic Counseling*. This indicates that this is the most active journal publishing a large number of documents and has a substantial impact on the field of genetic counseling. Among the top journals, the results revealed only three journals have SClmago Journal Rank (SJR) greater than two; the *American Journal of Human Genetics*, *Genetics in Medicine*, and *Journal of Medical Genetics*. As a result, it can be deduced that approximately 170 documents, or 4.9% of all genetic counseling publications, were published in high-impact journals.

The keyword co-occurrence map identified five major clusters in the genetic counseling field (Fig. 2A). These are genetic testing (red cluster), cancer (green cluster), genetic counselor (blue cluster), prenatal diagnosis (yellow cluster) and psychiatry (purple cluster) themes. Some of these topics (cancer and prenatal) have earlier been identified by content analysis study [5]. Genetic counseling should be regarded as an integral part of the genetic testing process [11]. Thus, genetic testing appeared as the main theme with the largest cluster. There are several categories of genetic testing that require genetic counseling. In diagnostic genetic testing, the test is performed on symptomatic individuals. While pre-test genetic counseling may not always be necessary, post-test genetic counseling should be offered especially when the result is positive. Conversely, pre-and post-test genetic counseling should be offered in other types of genetic testing such as predictive, susceptibility (risk), carrier, prenatal, preimplantation genetic diagnosis (PGD), and genetic screening. For pharmacogenetic testing, the need for genetic



counseling service, more studies are needed to look into the requirements and obstacles of patients, family members, and specialists particularly, in different settings/countries to ensure equitable access to all patients [12].

Genetic counselor, the third theme, is a health practitioner who has been trained to evaluate genetic testing results, both scientifically and medically, while also considering psychological, ethical, and family concerns [13]. The genetic counselor theme has the highest number of recent keywords based on the number of yellow nodes (Fig. 2B). These keywords with the average publication year 2018 onwards include 'covid-19', 'underrepresented population', 'service delivery models', 'workforce', 'disparities', 'service delivery', 'professional development', 'cultural competence', 'access', 'diversity', 'telemedicine' and 'health literacy'. These keywords are related to the genetic counselors' challenges and needs related to genetic service delivery models.

The fourth theme in the genetic counseling field is prenatal diagnosis. The need for prenatal genetic counselling is increasing worldwide due to advances in prenatal screening and diagnostic tests [14]. Given the complexity of genetic testing information, the role of genetic counseling at pre-and post-test is to increase knowledge, decrease anxiety, avoid decisional conflicts, and aid in the interpretation of test results, thereby helping an individual make an informed decision [14, 15].

The last theme in the genetic counseling field is psychiatry. Psychiatric genetic counseling has benefits for people with psychiatric disorders and their families despite the limited genetic testing available [16–18]. The role of genetic counseling is to help people understand that mental illness is not their fault, but there are things that they can do to protect their mental health [19]. It is an emerging speciality within clinical genetics [20].

The main limitation of our study is that, despite Scopus' vast coverage [9], we might have overlooked some important studies in other databases. Second, new research is being released almost daily, and the data in this analysis was up to January 19, 2022. Third, we might have missed several relevant documents if the authors had not included the term "genetic counseling" in the article titles. Lastly, the research themes in this study were derived from keyword co-occurrence thematic analysis without a deeper analysis.

## CONCLUSIONS

The field of genetic counseling is growing as genomic medicine develops. Genetic counseling research has identified the following themes: genetic testing, cancer, genetic counselor, prenatal diagnosis, and psychiatry. Future directions in genetic counseling research may be related to the requirements and challenges of genetic counselors, according to the emerging topics.

## DATA AVAILABILITY

The datasets generated during and/or analysed during the current study are available from the corresponding author on reasonable request.

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## AUTHOR CONTRIBUTIONS

WNAZ designed the study; RZ and AW contributed to data extraction and prepared the first draft of the manuscript; and all authors reviewed and provided feedback on the manuscript.

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## COMPETING INTERESTS

The authors declare no competing interests.

## ADDITIONAL INFORMATION

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