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ORIGINAL ARTICLE

The evolving role of genetic counselors in South Africa: A decade of growth

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Abstract

Genetic counseling has evolved from focusing solely on congenital anomaly prevention to incorporating psychological counseling. Traditionally, its goal has been to enhance patient care by providing clinical information and facilitating psychological adjustment. However, with the rise of genomics and complex testing technologies, genetic counselors now engage in education, research, clinical practice, and laboratory roles. A previous study from 2013 described the role of genetic counselors in South Africa. This current study provides an updated perspective on these findings and how these roles have diversified. Registered independent genetic counselors were invited via email to complete an electronic survey adapted from the previous study conducted in 2013. The survey collected data on information pertaining to (1) demographics, (2) genetic counseling experience and employment, and (3) work/clinical practice. Data were analyzed using descriptive statistics. Of 44 contacted genetic counselors, 29 responded. Most were female (97%), under 40 years old (60%), and had practiced for less than 10 years (59%). Although many genetic counselors took on multiple responsibilitiessuch as clinical, administrative, research, and teaching—the proportion primarily working in clinical roles declined from 75% (12/16) in 2013 to 45% (10/22) in 2023. Notably, genetic counselors involved in teaching and training had significantly fewer years in practice ($p = 0.0396$; CI 95%), suggesting a loss of expertise in academic centers. A new trend observed since 2013 is laboratory-based responsibilities, with 68% of participants reporting duties in this area. The role of genetic counselors in South Africa has expanded substantially over the past decade, with a shift away from predominantly clinical roles. These findings highlight the need for adaptation of training programs and strategic workforce planning to retain expertise in academic centers and ensure the continued growth of the profession.

KEYWORDS
genetic counseling roles, genetic counsellor practice, LMIC, workforce evolution

the study. A follow-up email was distributed midway through the recruitment period to encourage participation. Individuals who had not completed their training and registration at the time recruitment ended were excluded. The email included a link to the electronic survey and the participant information sheet. Informed consent was inferred for those who completed the survey. Ethics clearance was obtained from the Human Research Ethics Committee (Medical) of the University of the Witwatersrand (Ethics Clearance Certificate no. M231039).

2.2 | Instrumentation

The electronic survey used in this study was based on the survey employed by Kromberg et al. (2013) in the original study on the roles of genetic counselors in South Africa. The questionnaire was updated to ensure the changing roles of genetic counselors in South Africa could be adequately interrogated in fulfillment of the aims of this study. The main survey comprised three sections: (1) Demographic Information, (2) Genetic Counseling Experience and Employment, and (3) Practice Information. Section three included questions on the various roles and responsibilities fulfilled by the participant, including clinical, educational, research, laboratory, marketing, and administrative. This section also included three open-ended questions on the future of genetic counseling in South Africa and participants' thoughts on how the role of genetic counselors might change in South Africa. The survey was administered using Google Forms to make the participation as accessible as possible. A copy can be found in the Supplementary Material.

2.3 | Data analysis

All data were compiled and analyzed using the Microsoft Excel statistical package. The Shapiro–Wilk test was used to determine if the data were normally distributed and were reported as means and standard deviations, while data that were not normally distributed were reported as medians and interquartile ranges. To assess statistical differences between independent groups, unpaired two-tailed t-tests were performed. This test was chosen due to its suitability for comparing the means of two unrelated groups. Comparisons were conducted between various independent groups, including participants engaged in specific disciplines (e.g., teaching vs. non-teaching), those practicing within South Africa versus abroad, and individuals with differing qualification levels. These groupings were analyzed in relation to continuous variables such as age and years of experience to assess statistically significant differences. A p-value of less than 0.05 was considered indicative of statistical significance. Descriptive statistics were used to describe the data.

Responses to the open-ended questions were analyzed using qualitative content analysis as described by Krippendorff (2018). The analysis focused on identifying and categorizing the content from participant responses. Two researchers (MA and KF) independently read through the responses multiple times to familiarize themselves with the data and then developed an initial set of data-driven categories. These categories were iteratively refined through comparison and discussion. The frequency of responses within each category was noted to identify patterns across participants. Representative quotes were selected to illustrate each category. This inductive approach allowed the analysis to remain grounded in participants' perspectives, without being constrained by pre-existing theoretical frameworks. The process remained grounded in the principles of content analysis, focusing on the systematic identification and classification of patterns in textual data while allowing insights to emerge from participants' perspectives (Krippendorff, 2018).

3 | RESULTS

3.1 | Sample characteristics

A total of 29 genetic counselors completed the online survey, of which 75.9% (22/29) were working as genetic counselors in South Africa. Those practicing in South Africa reported working in three of the nine provinces, with the majority located in Gauteng Province. Forty-one percent (9/22) of participants in South Africa had <5 years of experience as a genetic counselor. The demographic characteristics of the participants are presented in Table 1.

3.2 | Distribution of time spent on various roles

Participants reported how their time was distributed across various professional activities. The largest share of time (31.8%) was spent on direct patient care, reflecting the continued importance of clinical counseling. Administrative duties—such as report writing, patient scheduling, billing, and clinic preparation—made up the second-largest portion (21.5%), suggesting a significant non-clinical workload that supports service delivery. Laboratory-based responsibilities (12.0%) included result interpretation, gatekeeping, and test selection support. Teaching and training activities (11.1%) encompassed undergraduate and postgraduate instruction, intern supervision, and curriculum development. Research activities (10.5%) varied from student supervision to personal research projects and participation in funded studies. Smaller percentages of time were spent on outreach, advocacy, and multidisciplinary meeting coordination, although the latter was often described in qualitative responses rather than specifically quantified in the time allocation section.

Participants were categorized based on the highest percentage of time spent on a specific activity, with their primary job focus identified as clinical, research, laboratory, teaching, or a combination of these if time allocation was evenly split across multiple categories. As illustrated in Figure 1, 68% (15 out of 22) of participants identified their clinical role as a primary focus. Among them, five participants reported an equal focus on additional roles, including research (3/5), teaching (1/5), and a combination of teaching and laboratory (1/5).

student supervision to degree completion as their main focus. The remaining respondents reported conference participation (26.7%; 4/15) and product development (6.7%; 1/15) as their primary research outputs.

3.6 | Laboratory roles

Among the 22 genetic counselors currently practicing in South Africa, 68.2% (15/22) reported involvement in laboratory-based duties. While the majority allocated approximately 10%–15% of their time to laboratory roles, one participant indicated that 90% of their time was dedicated to such tasks. The most common laboratory responsibility was result interpretation and reporting, reported by 93.3% (14/15) of participants, followed by ensuring appropriate test selection for incoming genetic testing requests, reported by 73.3% (11/15).

3.7 | Qualitative content analysis

Using an inductive content analysis approach, the open-ended responses were examined to allow key topics to emerge from the data. These topics were not predetermined but developed through a process of open coding and iterative refinement by the researchers. This analysis revealed four main areas that reflect participants' perspectives on the current state and anticipated evolution of the genetic counseling profession in South Africa. These topics highlight both the opportunities for professional expansion and the systemic barriers that may influence future growth.

3.7.1 | Perceptions of the Profession's future in South Africa

When asked about their perception of the future of the genetic counseling profession and the role of genetic counselors in SA, most participants felt that the genetic counseling service would expand and become more broadly accessible throughout the country, citing factors such as increasing awareness of genetic conditions among the public and healthcare providers, the growing integration of genomics into mainstream medical practice, and the anticipated demand for personalized medicine approaches:

> Hopefully we will be able to service all the provinces in SA within the next 10 years.
>
> (GC11)

> Genetic counselling will continue to grow as new genomic studies are being introduced and will require this workforce for assistance in the delivery of these results.
>
> (GC19)

Most participants believed that genetic counseling would eventually become a standard part of patient care. They expressed hope that both state and private sectors would continue to expand the availability of these services:

> It should become a standard part of patient care.
>
> (GC08)

> I hope that there is a future where genetic counsellors are members of staff in any and all hospital settings and that our value is realised.
>
> (GC17)

However, many participants felt that this expansion was contingent on formal legislation surrounding the use of genetic counseling and testing services, as well as recognition from medical insurance providers and national health departments:

> If the need is not highlighted in legislation, the community doesn't put out committee statements and regulations on the appropriate use of genetic testing/referral to genetic specialists; genetic counsellors are not utilised in the day-to-day of medicine.
>
> (GC15)

> There's a need for formal legislation and a concerted effort to include GCs.
>
> (GC23)

3.7.2 | Broadening the scope of genetic counselors

Other participants felt that the future of the genetic counseling profession in South Africa would see a broadening of the scope of genetic counselors, particularly into non-traditional roles in laboratories and industry. There was a common belief that genetic counselors' transferable skills could be utilized in a wide range of sectors beyond the clinical setting:

> [Genetic counselors] will move into non-traditional roles more often as our transferable skills are in high demand.
>
> (GC27)

> I think non-clinical genetic counsellors will become more prevalent, with more GCs working in labs, industry etc.
>
> (GC20)

Participants also saw potential for genetic counselors to become integral members of multidisciplinary teams in clinical settings, particularly in specialties where genetic counseling is currently underutilized:

experience, an age and experience profile that has been observed in other countries such as Australia and New Zealand (Kanga-Parabia et al., 2024). In the South African context, this is likely a consequence of the temporary cessation of training in the early 2010s, with the first genetic counseling cohort to qualify again being in 2017 at UCT and 2019 at WITS. Additionally, the availability of qualified, registered genetic counselors in training units limits the intake of new students (Wessels et al., 2024). Nonetheless, the predominance of younger professionals can be viewed as a positive indicator of the profession's renewed growth, signaling increased visibility and interest in genetic counseling among the next generation of healthcare workers. A younger workforce may also offer opportunities for long-term sustainability and innovation, particularly as genetic services continue to expand in South Africa. This aligns with the National Department of Health's strategic emphasis on developing a skilled, future-oriented health workforce capable of addressing the country's evolving healthcare needs (National Department of Health, 2023). Although data on race and ethnicity were collected, they were not reported to maintain participant anonymity, given the small size and close professional networks within the South African genetic counseling community. However, it is notable that not all respondents identified as white, indicating a positive shift toward increased diversity within the profession. Although no formal studies have examined the reasons for this shift, both training institutions and the GC-SA committee have made increased efforts to actively recruit students from diverse backgrounds and regions.

Unlike many other countries, more genetic counselors reported working in the private rather than state healthcare sector. The reported ratio of genetic counselors practicing in state versus private healthcare is approximately 2:1. However, this does not accurately represent population access, which is closer to 5:1 in favor of state healthcare (Cowling, 2023) showing a significant imbalance in the supply and demand of genetic counseling services. As there are currently only seven state healthcare posts in existence (two with the Department of Health in the Western Cape and five with the NHLS in Gauteng), this reflects the lack of government-supported jobs available to genetic counselors in South Africa. This is supported by Gomes et al. (2024), who reported that in South Africa, the number of genetic counselors in private healthcare is sufficient to serve 22% of the population. In contrast, only 2% of the population relying on state healthcare has access to genetic counselors employed by the government.

Genetic counselors in South Africa embody the phrase 'jack of all trades'. Given the shortage of genetic counselors, there is little to no opportunity to specialize in one area of interest, such as prenatal or pediatric practice, or in a specific role, such as researcher or lecturer. The majority of participants reported fulfilling a mixed role consisting of various responsibilities, and while clinical practice is still reflected as the main focus, the percentage of genetic counselors who reported clinical practice as their core focus decreased over the last decade from 75% reported by Kromberg et al. (2013) to 45% reported in this study. This reflects a subtle but definite shift away from the traditional patient-facing role.

This shift reflects both the influence of private stakeholders, such as private laboratories aligning with international standards, and the proactive efforts of qualified genetic counselors seeking to establish their roles within the evolving healthcare landscape. Importantly, however, this evolution is not solely a reflection of adaptation to a changing healthcare system: it also represents a pragmatic survival strategy in a resource-constrained setting where clinical genetic counseling positions, particularly within the public sector, are scarce (Gomes et al., 2024; Wessels et al., 2024). As such, many genetic counselors are compelled to diversify their skill sets and pursue roles outside of traditional clinical care in order to secure employment and sustain their careers in the profession. This pattern aligns with broader trends of professional migration and survival within healthcare systems that offer limited institutional support (Khan, 2021).

The majority of participants in this study worked predominantly in a cancer setting. This is similar to findings in the USA and Canada, where adult cancer cases were cited as the most common area of practice for genetic counselors (at 41% and 42% respectively: Canadian Association of Genetic Counselors, 2022: National Society of Genetic Counselors, 2024). The South African study of 2013, in contrast, showed comparatively little cancer genetic counseling at the time (Kromberg et al., 2013). Genetic counseling and available testing for cancer genetics, specifically breast cancer, have increased significantly over the last few years in South Africa, as predicted by Kromberg et al. (2013) who noted the upward trend of these cases starting around 2012. This could be due to the South Africa Department of Health (DoH) clinical guidelines for breast cancer control and management released in 2019, outlining various goals and strategic objectives aimed at reducing the burden of breast cancer by 2030. The decreased cost of next-generation sequencing and the increased accessibility of panel testing have also resulted in higher throughput and increased cost-effectiveness of testing, leading to the criteria for testing being broadened (Oosthuizen et al., 2023). Other contributing factors could also be increased awareness of breast cancer, as well as the increased screening availability for at-risk individuals. Cancer treatments such as PARP-inhibitors, which are administered based on an individual's genetic results, have also increased the need for oncologists to request genetic testing to assist with appropriate management.

Apart from oncology, prenatal practice is often reported as a focus area for a large proportion of genetic counselors in other countries (Canadian Association of Genetic Counselors, 2022: Kanga-Parabia et al., 2024; National Society of Genetic Counselors, 2024). Although prenatal genetic counseling was not a primary focus for many participants in this study, a significant number expressed a desire to become more actively involved in this area. The underrepresentation of genetic counselors in the prenatal setting appears to reflect limited opportunities and insufficient support from

genetic counselors, with educational programs adapting to include laboratory-based and genomic competencies as core components of practice. In line with this, the HPCSA is in the process of updating both the intern training curriculum and scope of practice to formally recognize these roles (HPCSA GC Representative, personal communication, 2024).

4.5 | Imagining the future of genetic counseling in South Africa

The qualitative findings of this study highlight several key perceptions and expectations regarding the future of the genetic counseling profession in South Africa. Participants expressed a largely hopeful outlook, envisioning the expansion and increased accessibility of genetic counseling services across the country. There was a strong sense that genetic counseling could become integrated into mainstream medicine and available in all nine provinces, consistent with global trends showing significant growth in the profession (National Society of Genetic Counselors, 2024).

However, participants also identified systemic barriers that may inhibit this anticipated growth. These included the absence of formal legislation on genetic counseling and testing, limited support from medical insurance providers, and insufficient recognition by national health authorities. Similar challenges have been described internationally, where genetic counselors often face issues related to professional regulation, recognition, and incorporation into healthcare systems (Ormond et al., 2024; Wessels et al., 2024).

A common view expressed by participants was that genetic counselors in South Africa would increasingly occupy non-traditional roles. Participants identified opportunities for involvement in laboratories, the healthcare industry, policy development, administration, and consulting for institutions such as insurance providers and national health departments. These perspectives align with recent international reports describing the expansion of genetic counselors' roles beyond clinical settings (Ormond et al., 2024).

Participants also highlighted the potential value of professional specialization. It was suggested that focusing on particular clinical or non-clinical domains could enhance credibility, foster deeper expertise, and support integration into underutilized areas of the healthcare system. Genetic counselors in South Africa have historically held versatile roles, including in education, research, and advocacy (Kromberg et al., 2013), and many appear eager to see these contributions grow in scope and impact.

That said, some participants were more cautious in their outlook, expressing concerns that the profession's growth would remain constrained by systemic challenges. They pointed to a lack of awareness among healthcare professionals and policymakers, limited stakeholder engagement, and few innovative strategies for expanding service delivery. These concerns echo previous findings highlighting persistent underutilization of genetic counseling services in South Africa (Thom & Haw, 2021).

It is important to acknowledge that participants' optimism may appear somewhat at odds with the reality of systemic limitations facing the genetic counseling profession in South Africa. Since the publication of Kromberg et al. (2013), subsequent studies have demonstrated that the expansion of services has been slow, with persistent challenges in workforce capacity, funding, and policy integration (Gomes et al., 2024; Wessels et al., 2024). This stagnation occurs within the broader context of South Africa's healthcare system, which is shaped by a high burden of disease, substantial public-private disparities, economic constraints, and shifting political priorities (Harvard Global Health Institute, 2021; Kahn et al., 2018; Vaughan et al., 2020). The COVID-19 pandemic further strained resources and diverted attention away from health services like genetic counseling (Boulle et al., 2021). Despite these realities, the optimism expressed by participants in this study may reflect a growing professional identity, increased training opportunities, international exposure, and hope for the future. This disconnect between perceived potential and structural limitations underscores the need for realistic strategic planning grounded in the South African context.

Although this study offers rich insights into participants' expectations, it is important to note that the open-ended survey design did not explore the reasoning behind participants' views in depth. As such, while key topics such as professional expansion, specialization, and non-traditional roles were commonly identified, the underlying motivations behind these perspectives remain less well understood. Future qualitative research could use interviews or focus group discussions to explore these views more deeply and capture the contextual nuances that could not be accessed through written survey responses alone.

Altogether, these findings highlight the dual reality facing genetic counseling in South Africa: one of clear ambition and perceived opportunity, and another shaped by structural limitations. Addressing this gap will require systemic support, coordinated advocacy, and deliberate planning to ensure that the profession can fully contribute to South Africa's evolving healthcare landscape.

5 | CONCLUSION

Over the past decade, the role of genetic counselors in South Africa has expanded beyond traditional clinical practice to encompass significant contributions in education, research, laboratory-based roles, policy development, and advocacy. The findings of this study underscore the dynamic evolution of the profession, with an increasing number of genetic counselors actively participating in multidisciplinary teams, engaging in non-clinical roles, and contributing to research-driven initiatives.

Despite these advancements, several challenges persist, particularly in relation to workforce distribution, professional recognition within the healthcare system, and equitable access to genetic counseling services across all provinces. To facilitate the continued development of the profession, strategic efforts should be directed

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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