



## SPECIAL ARTICLE

# The role of the Latin American Professional Society of Genetic Counseling (SPLAGen): Advancing genetic counseling in Latin America



D. Diaz Caro<sup>1,\*</sup>, L. Simone<sup>2</sup>

<sup>1</sup>Columbia University Department of Neurology, Columbia University Irving Medical Center, New York; <sup>2</sup>Center for Genetic and Genomic Medicine, Hackensack University Medical Center, Hackensack, New Jersey

### ARTICLE INFO

#### Article history:

Received 9 February 2024

Received in revised form

9 July 2024

Accepted 11 July 2024

Available online 16 July 2024

#### Keywords:

Awareness

Education

Genetic counseling

Latin America

Professional society

### ABSTRACT

The increasing importance of genomics and its expanding clinical application underscore the significance of genetic counseling in disseminating this vital information. In many countries, including regions of Latin America and the Caribbean, there is a shortage of adequately trained genetics providers thus limiting access to these essential services. Moreover, providers practicing genetic counseling are scarce and experience a lack of professional support. To address these challenges, the Latin American Professional Society of Genetic Counseling (SPLAGen, for its Spanish and Portuguese abbreviation) was established with the objective to increase awareness of and access to genetic counseling services in the region. Through this commentary, we aim to provide insights into the formation and organizational structure of SPLAGen, outlining our impactful work, as well as the current and future obstacles we foresee in our endeavor. SPLAGen's efforts aim to serve as a critical step toward bridging the gap in genetic counseling services and promoting better health care outcomes for patients living in Latin America and the Caribbean.

© 2024 The Authors. Published by Elsevier Inc. on behalf of American College of Medical Genetics and Genomics. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

## Introduction

Latin America and the Caribbean, home to over 600 million people, boasts a rich diversity of cultures and languages.<sup>1,2</sup>

The region has made significant contributions to the field of medical genetics, enriching global knowledge and understanding.<sup>3,4</sup> Despite these contributions, the region confronts notable challenges in providing equitable genetic services.<sup>5-8</sup>

A critical issue is the shortage of qualified genetics professionals, severely restricting access to crucial genetics

services.<sup>5-7</sup> For example, in Mexico, the ratio of genetic professionals is 1 to 100,000 citizens and in Brazil, it is 1 for 770,000 citizens.<sup>2,3</sup> Furthermore, the administration of services such as genetic counseling is performed by medical geneticists or other health care providers who lack specialized genetics training, the latter leading to concerns regarding the quality of care patients receive.<sup>5-9</sup> Organizations such as Red Latinoamericana de Genética Humana (RELAGH) created in the 1990s have worked to develop collaborative projects to improve education, research,

The Article Publishing Charge (APC) for this article was paid by the SPLAGen board of directors.

\*Correspondence and requests for materials should be addressed to Daniela Diaz Caro, SPLAGen, 30 E 33rd Ave, Po Box 51166, Eugene, OR 97405.

Email address: [dd3094@cumc.columbia.edu](mailto:dd3094@cumc.columbia.edu)

doi: <https://doi.org/10.1016/j.gimo.2024.101870>

2949-7744/© 2024 The Authors. Published by Elsevier Inc. on behalf of American College of Medical Genetics and Genomics. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

assistance, and representation of medical genetics in the region.<sup>10</sup> However, no organization in Latin America has yet focused solely on the topic of genetic counseling.

Genetic counseling is an important aspect of delivering genetic care to patients. It is a process designed to help individuals understand and come to terms with the medical, psychological, and familial implications of genetic contributions to disease.<sup>11,12</sup> To achieve this, genetic counseling uses specific strategies, such as the reciprocal-engagement model, to equip professionals with the necessary tools to deliver care that is centered around the patient's needs.<sup>13</sup> The objectives of genetic counseling include facilitating patient decision making, providing education regarding genetic conditions or risks, offering psychosocial support, and anticipatory guidance for the patient and their family members.<sup>11-13</sup>

Regions such as Europe and Asia have established educational programs to train nonphysician genetic counselors; yet, this remains largely absent in most Latin America and the Caribbean.<sup>14</sup> Although genetic counseling is not recognized as an independent profession in most of Latin America, Brazil, and Cuba offer a master's degree program in genetic counseling,<sup>14,15</sup> whereas Chile has a certificate program in genetic counseling primarily for nongenetics physicians and nurses.<sup>16</sup> As a result, most genetic counseling services in Latin America are carried out by medical geneticists or health care professionals with various degrees of training.

The establishment of the Latin American Professional Society of Genetic Counseling (SPLAGen) marks a significant step toward addressing these challenges. By advocating for the recognition of genetic counselors and promoting greater educational opportunities for health care professionals, this society aims to drive toward the equitable global application of genetic services. This initiative represents a collective effort to overcome the barriers to access and to enhance the provision of genetic services across the region, aligning with the goal of fair and equitable health care for all Latin Americans.

## The birth of the Latin American Professional Society of Genetic Counseling

The idea of a professional society emerged from conversations among genetic counselors practicing in the United States with ties to Latin America and their counterparts in the region. In 2020, during a meeting through the Minority Genetics Professional Network, the concept to crowdsource a list of working professionals providing genetic counseling services in Latin America was born. This initiative highlighted the lack of knowledge that US-based genetic counselors had of clinical professionals providing genetic counseling and genetics services in Latin America. A conversation between Laurie Simone, MS, CGC, a genetic counselor in the United States, and Sonia Margarit, MS, CGC, a clinical genetic counselor working in Chile,

emphasized the lack of support for genetic counselors in Latin America. Further conversations with genetic counselors and geneticists from the region provided direction and the necessary impetus to form a group committed to supporting the field of genetic counseling in Latin America.

Our first step involved assessing interest among those providing genetic counseling services in Latin America regarding the formation of a dedicated organization. To achieve this, we developed an Interest and Needs Assessment (INA) survey in Spanish and Portuguese. This survey aimed to gather contact information, determine the perceived necessity of a professional society, identify the forms of support needed, collect ideas for the society's objectives, and gauge interest in active participation. We received 53 responses from individuals providing genetic counseling services in Latin America, including 27 medical geneticists, 11 physicians from other specialties, 9 genetic counselors, 5 researchers, and 1 genetic counseling student (SPLAGen internal communications, 2022). This survey revealed a strong desire for additional professional support, educational opportunities, and improved access to genetic counseling services. In response, Laurie Simone, MS, CGC, Daniela Diaz Caro, MS, CGC, and Amanda de Leon, MS, CGC, devised strategies to address these needs expressed in the INA survey. We formulated a plan for developing a professional society based on the INA survey findings and a shared desire to establish a robust organizational foundation, ensuring the longevity and effectiveness of the group's objectives for future genetic counselors practicing in Latin America.

## Laying the groundwork for future success

The development of a professional society required a systematic approach: laying the foundation through the establishment of a name, detailed goals, bylaws, and a financial support plan. A crucial step in building a platform for future success was establishing our name: La Sociedad Profesional Latinoamericana de Asesoramiento Genético in Spanish and Sociedade Profissional Latino-americana de Aconselhamento Genético in Portuguese, or SPLAGen for its Spanish and Portuguese abbreviation. Our initial goals were formulated from survey responses and aimed to promote awareness of and access to genetics services and genetic counseling in Latin America.

Leveraging knowledge from other organizations, we determined the best approach to achieve these goals was through the development of educational opportunities, professional and patient specific advocacy, research, public policy, and networking. Our bylaws and organizational structure are decided upon to ensure effective decision making aligned with SPLAGen's goals, prioritizing the interests of Latin American genetic counseling providers. For example, we established that our board of directors must include regional representatives from South America, Central America, the Caribbean, and Mexico. The leadership

structure includes executive roles (eg, president, secretary, and treasurer), regional representatives, and committee chairs. Our committees, which play pivotal roles in driving our project initiatives, include the Educational Committee, Research and Ethics Committee, Membership and Marketing Committee, and Public Policy and Access Committee. Projects for each committee are selected based on feedback from member surveys and guidance from the board of directors. Furthermore, we actively encourage members to initiate their own projects, fostering a collaborative environment in which support from fellow members is readily available. The official launch of our organization coincided with our first board of directors meeting and website ([splagen.org](http://splagen.org)) launch in August 2021.

Within SPLAGen, membership is categorized in 4 distinct groups: full members who are genetic counselors, full members from other professions practicing genetic counseling, allied professionals, and students interested in our mission. All members enjoy benefits such as access to educational resources, committee meetings, networking opportunities, and professional development. However, leadership roles, including positions on the board and committee chairs, are exclusively available to full members. This structure ensures a diverse yet focused leadership, guiding the organization toward fulfilling its goals and mission.

To secure financial support for our initiatives, we obtained a 501(c)(3) nonprofit status in the United States, which was granted by the Internal Revenue Service in November of 2023. Obtaining a 501(c)(3) status is important because it exempts us from federal income tax, allows donors to receive tax deductions for donations, and provides us access to federal grants. Overall, our nonprofit status improves our ability to access funding opportunities that directly support our mission while minimizing organizational tax liabilities. Our funding model, still in development, aims to incorporate member contributions, private donations, and grants. These financial resources will be instrumental in bolstering the initiatives of our various committees under the guidance of our board of directors.

### **Accomplished and ongoing projects**

In the early stages of SPLAGen, we have concentrated on several key projects to establish a robust foundation for the field of genetic counseling in Latin America. Our initial endeavor, led by the Public Policy and Access Committee in 2021, involved creating a directory of Latin American geneticists and genetic counselors, including those in the United States who offer services in Spanish and Portuguese. This grant-funded initiative involved hiring software developers to help build an interactive directory map, enabling users to search for providers by language, location, and specialty. Concurrently, our Marketing and Communications Committee established an international internship program targeted at students from Latin America or of Latin

American descent interested in the field of genetic counseling. Under the guidance of genetic counselors, our interns are taught how to communicate genetic concepts to the general public and are provided with mentorship support when they apply to genetic counseling programs. In 2023, SPLAGen's Educational Committee launched virtual educational webinars for continuing professional development. These webinars aimed to enhance awareness of genetic counseling, covering topics from various specialties, condition-specific content, and current research. All educational webinar recordings are publicly accessible through our YouTube channel.<sup>17</sup> The Research Committee has played a pivotal role in guiding student research projects, providing academic support, and fostering international research collaborations. Finally, our board of directors has been instrumental in shaping organizational goals, mission statements, and securing our nonprofit status.

### **Current and foreseeable obstacles for the implementation of the genetic counseling profession in Latin America**

The obstacles we have encountered thus far provide insight into potential future barriers to the establishment and recognition of the genetic counseling profession in Latin America. A study completed in collaboration with SPLAGen identified several key issues in implementing genetic counseling in the region.<sup>18</sup> One major challenge is the limited awareness that health care providers have regarding the distinct role of genetic counselors, whether as independent professionals or as part of a specialized skill set in healthcare.<sup>7,18</sup> This lack of knowledge about the unique roles of genetic counselors, compared with other healthcare professionals, such as geneticists, hinders the recognition and integration of their valuable skill set within the healthcare system.

Additionally, a significant gap exists in the literature regarding the practice of genetic counseling in Latin America. More studies are needed that address the psychosocial and cultural nuances involved in delivering genetic counseling to diverse patient populations across various medical specialties in the region. Comprehensive and region-specific research is essential to effectively understand and adapt genetic counseling to the unique contexts of Latin America.

The shortage of trained and experienced genetic counselors in Latin America reinforces these barriers by impeding progress in raising awareness, advancing research, and delivering patient care. To date, we are aware of 5 providers in the region who have completed a master's level training program in genetic counseling, with 3 of them also possessing a medical degree in genetics (SPLAGen, internal communication, 2022). To address this shortage, the development of educational infrastructure is essential. Graduate programs are needed to train genetic counselors within the region for local practice. Currently, because these

programs are in their nascent stage,<sup>14</sup> many professionals are left to seek educational opportunities abroad. Studying abroad comes with a significant financial burden and English language requirements that can make obtaining a master's degree in genetic counseling inaccessible for many aspiring genetic counselors in Latin America.<sup>19,20</sup>

To support the growth of genetic counseling in Latin America, SPLAGen is committed to establishing strong support mechanisms, including research and educational opportunities. We recognize and value the dedication of those who volunteer their time to contribute to this field.

This support serves not only as a backbone for our sustained success but also a catalyst for the future evolution of the profession in this vibrant region. By emphasizing a unified vision of Latin America, rather than viewing it as individual countries, we hope to harness our collective strength. Future collaborations with already established organizations will be needed to ensure continued progress, while engaging with stakeholders in the region. We also plan to maintain ongoing awareness and engagement through our regular committee meetings and outreach operations. This approach not only empowers us to serve our patients more effectively but also aligns our efforts toward a shared, impactful goal. Together, we can transform challenges into opportunities and ensure that the field of genetic counseling thrives, benefitting both professionals and patients across Latin America.

## Conclusion

The journey of SPLAGen, from an idea born in conversations among genetic counselors to an impactful organization shaping the future of genetic counseling in Latin America, is a testament to collective vision and dedication. We have successfully laid a strong foundation, addressing the urgent need for increased awareness, education, and professional support within the field in the languages of the people. Our commitment to overcoming barriers, such as the scarcity of trained professionals, recognition of genetic counselors, lack of research, and access to genetic counseling services in Latin America, continues to drive our pursuits. As we forge ahead, our focus remains on enhancing education, promoting public policy, fostering research, and growing networking opportunities to empower current and future genetic counselors in the region. Embracing a unified vision for the region, SPLAGen stands at the forefront of a burgeoning opportunity for the globalization of genetic counseling and is poised to ensure that genetic counseling effectively serves the diverse populations of Latin America.

## Data Availability

The data that support the findings are available from the corresponding author, D.D.C., upon request.

## Acknowledgments

The authors like to thank Amanda de Leon, MS, LCGC for her thoughtful review and comments of this commentary.

## Funding

The research described in this manuscript was not supported by a grant or other funding source.

## Author Contributions

Conceptualization: L.S., D.D.C.; Writing original draft: D.D.C.; Writing-review and editing: L.S, D.D.C.

## Ethics Declaration

No informed consent was required for this manuscript.

## Conflict of Interest

Daniela Diaz Caro, MS, CGC is a co-founder and chair of the Research Committee for the Latin American Professional Society of Genetic Counseling. Laurie Simone, MS, CGC is a co-founder and president of the Latin American Professional Society of Genetic Counseling.

## References

1. Population, total – Latin America & Caribbean. World Bank Open Data. Accessed January 28, 2024. <https://data.worldbank.org/indicator/SP.POP.TOTL?locations=ZJ>
2. Sanabria H. Anthropology, Latin America, and the Caribbean. In: Sanabria H, ed. *The Anthropology of Latin America and the Caribbean*. 1. Routledge; 2019:30. <http://doi.org/10.4324/9781315560427>
3. Sohail M, Palma-Martínez MJ, Chong AY, et al. Mexican Biobank advances population and medical genomics of diverse ancestries. *Nature*. 2023;622(7984):775-783. <http://doi.org/10.1038/s41586-023-06560-0>
4. Barahona A. Medical genetics and the first studies of the genetics of populations in Mexico. *Genetics*. 2016;204(1):11-19. <http://doi.org/10.1534/genetics.116.191767>
5. Ashton-Prolla P, Goldim JR, Vairo FP, da Silveira Matte U, Sequeiros J. Genomic analysis in the clinic: benefits and challenges for health care professionals and patients in Brazil [published correction appears in *J Community Genet*. 2015;6(3):285]. *J Community Genet*. 2015;6(3):275-283. <http://doi.org/10.1007/s12687-015-0238-0>
6. Bucio D, Ormond KE, Hernandez D, Bustamante CD, Lopez Pineda A. A genetic counseling needs assessment of Mexico. *Mol Genet Genomic Med*. 2019;7(5):e668. <http://doi.org/10.1002/mgg3.668>
7. Margarit SB, Alvarado M, Alvarez K, Lay-Son G. Medical genetics and genetic counseling in Chile. *J Genet Couns*. 2013;22(6):869-874. <http://doi.org/10.1007/s10897-013-9607-1>

8. Rodas-Pérez C, Clarke A, Powell J, Thorogood M. Challenges for providing genetic counselling in Colombian genetic clinics: the viewpoint of the physicians providing genetic consultations. *J Community Genet.* 2015;6(3):301-311. <http://doi.org/10.1007/s12687-015-0237-1>
9. Klitzman R, Chung W, Marder K, et al. Attitudes and practices among internists concerning genetic testing. *J Genet Couns.* 2013;22(1):90-100. <http://doi.org/10.1007/s10897-012-9504-z>
10. Rojas-Martínez A, Giraldo-Ríos A, Jiménez-Arce G, de Vargas AF, Giugliani R. RELAGH – the challenge of having a scientific network in Latin America: an account from the presidents. *Genet Mol Biol.* 2014;37(1):305-309. <http://doi.org/10.1590/s1415-47572014000200017> (suppl).
11. National Society of Genetic Counselors' Definition Task Force, Resta R, Biesecker BB, et al. A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. *J Genet Couns.* 2006;15(2):77-83. <http://doi.org/10.1007/s10897-005-9014-3>
12. Ormond KE, Hayward L, Wessels TM, Patch C, Weil J. International genetic counseling: what do genetic counselors actually do? *J Genet Couns.* 2024;33(2):382-391. <http://doi.org/10.1002/jgc4.1735>
13. Veach PM, Bartels DM, Leroy BS. Coming full circle: a reciprocal-engagement model of genetic counseling practice. *J Genet Couns.* 2007;16(6):713-728. <http://doi.org/10.1007/s10897-007-9113-4>
14. Abacan M, Alsubaie L, Barlow-Stewart K, et al. The global state of the genetic counseling profession. *Eur J Hum Genet.* 2019;27(2):183-197. <http://doi.org/10.1038/s41431-018-0252-x>
15. Cruz AL. An overview of genetic counseling in Cuba. *J Genet Couns.* 2013;22(6):849-853. <http://doi.org/10.1007/s10897-013-9635-x>
16. de Asesoramiento Genético D., de Medicina Facultad Clínica Alemana Universidad del Desarrollo. <https://medicina.udd.cl/ver-diplomado/diplomado-de-asesoramiento-genetico/>. [Accessed 9 June 2024].
17. SPLAGen. YouTube. Accessed January 28, 2024. <https://www.youtube.com/@SPLAGen>
18. Venezia R. *Exploring the perspectives of genetics providers in Latin America on genetic counseling and the barriers to establishing the field in the region.* Hinxton, United Kingdom: Oral presentation at: World Congress on Genetic Counselling; October 2023.
19. Akgumus G, Shah D, Higgs L, Valverde K. Professional issues of international genetic counseling students educated in the United States. *J Genet Couns.* 2016;25(4):664-676. <http://doi.org/10.1007/s10897-016-9947-8>
20. Sabbadini M, Naldi M, Packman W, Youngblom J, Weil J. International genetic counseling students' perspective on their training experience in the United States. *J Genet Couns.* 2013;22(6):817-829. <http://doi.org/10.1007/s10897-013-9648-5>