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RESEARCH

Genetics services in Latin America: a descriptive study of availability and utilization of genetics in healthcare

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Abstract

Genetic services are expanding globally, but access remains limited in low-resource regions such as Latin America. Understanding current service availability, barriers, and facilitators is critical to guide capacity building and improve patient care. We conducted a cross-sectional survey of healthcare professionals providing genetic services in Latin America. The survey, available in Spanish and English, assessed genetic services, referral patterns, testing availability, barriers, facilitators, and perceived needs. Descriptive statistics summarized quantitative data, and thematic analysis was applied to open-ended responses. Eighty-five respondents from 18 countries reported broad clinical activity across pediatric, cancer, and adult-onset genetic conditions. Commonly ordered tests included karyotype, gene panels, and exome sequencing, with many sending samples abroad. Key barriers included high costs, workforce shortages, and limited clinician familiarity with testing. Facilitators included clinician interest and laboratory partnerships. Respondents emphasized the need for expanded test access, workforce development, and increased training opportunities. Genetic services in Latin America show substantial growth but face persistent barriers. Strengthening local infrastructure, training, and collaborative efforts is essential to improve access, reduce diagnostic delays, and enhance patient outcomes.

Keywords Genetic services · Latin America · Genetic testing · Capacity building

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Introduction

Genetic services are expanding globally as sequencing costs decrease, and genetic knowledge is integrated into clinical practice. Clinical genetics expertise aids in diagnosing complex genetic conditions and guiding cost-effective medical management (Dimmock et al. 2021). Implementing genomic medicine has improved health outcomes and reduced costs in prenatal (Migliavacca et al. 2024; Wang et al. 2022), pediatric (Szigety et al. 2022), and cancer clinical settings (Pritchard et al. 2022; Rao et al. 2024; Szigety et al. 2022). Additionally, genomic technologies are useful for population-based screening (Allen et al. 2024) such as newborn screening (NBS) (Ceyhan-Birsoy et al. 2019). However, additional genetics expertise and infrastructure are needed to make genomic medicine feasible and effective in low-resource settings.

Studies have shown that genetics expertise in clinical practice is key to providing an accurate and timely diagnosis for genetic conditions (Chen et al. 2023; Hendricks-Sturrup et al. 2020). Often, a genetic diagnosis can inform

medical management and treatment decisions. Genetic conditions like phenylketonuria (PKU) can be treated with diet adjustments. While others, such as cystic fibrosis and X-linked adrenoleukodystrophy, have treatments to cure, halt or slow progression and mediate the severity of disease. Without access to testing, genetic conditions such as these may remain undetected and untreated. Cost is a major barrier to implementing genetic testing in clinical care in both high and low resource settings (Hackl et al. 2023; Powell et al. 2024). However, the cost-saving potential of an accurate diagnosis without the delays to clinical management caused by barriers to genetic evaluation are often overlooked in healthcare policy (De Rosa et al. 2024).

In Latin America (LATAM), genetic services are provided through a variety of professionals and contexts. Previous descriptions of genetic services in Latin America vary by country are outdated and lack uniformity (Bonilla et al. 2022; De Castro and Restrepo 2015; Guio et al. 2018; Larrandaburu et al. 2015; Paz y Miño et al. 2015; Penchaszadeh 2013; Toland et al. 2018; Torre-Hernandez et al. 2018; Vishnopolska et al. 2018). In contrast, the landscape of genetic services in the United States (Jenkins et al. 2021), Europe (Unim et al. 2019), and Asia (Abacan et al. 2019; Kim et al. 2022; Thong et al. 2018) is well described in publicly accessible resources. Although genetic services in LATAM have been developing rapidly in recent years (De Rosa et al. 2024; Diaz Caro and Simone 2024), this development is not well described in scientific literature. This gap highlights the need for research describing current genetic services availability and utilization in LATAM.

Recent reports highlight a lack of training opportunities and awareness of genetic services in LATAM (Bucio et al. 2019; Fernández-Ramires, Morales-Pison, Rucatti, Echeverría, San Martín, Cammarata-Scalisi et al., 2024; Giugliani et al. 2022; Ormond et al. 2024). Calls to action stress the need for national NBS programs, a unified definition of rare disease, and a push for legislative action to advance the implementation of genomics in healthcare (De Rosa et al. 2024; Giugliani et al. 2022; Wainstock and Katz 2023). A necessary step towards action is to catalogue the landscape of genetic services in LATAM. The availability and accessibility of such information can facilitate advocacy and collaboration between established genetics centers and developing clinics to meet patients' needs and improve clinical care (MacFarland et al. 2022; Wainstock and Katz 2023). We seek to address the gap in recent literature on genetic services in LATAM. We aim to (1) catalogue available genetic services, their context and implementation (2) assess barriers, facilitators and needs, and (3) evaluate the adequacy of genetic services in terms of genetic testing, genetic counseling, and medical management from a healthcare provider perspective.

Materials and methods

This study was approved by the Baylor College of Medicine Institutional Review Board (Protocol # H-55131). A survey was developed through an iterative process with healthcare providers with experience providing genetic services in LATAM. Survey creation was guided by the Consolidated Framework for Implementation Research (CFIR) (Damschroder and Lowery 2013), as no standardized measure exists to assess availability and utilization of genetic services in LATAM. After three rounds of drafting and revision, the survey included questions about the patient population, genetic testing, demographic information, and five-point Likert scale questions addressing the adequacy of genetic testing, genetic counseling, medical management, demand for genetic services, and workforce size. Free text questions were included to allow respondents to provide context for their answers if desired. The survey was translated into Spanish and reviewed by native speakers to ensure linguistic accuracy. It was distributed via REDCap (Harris et al. 2009, 2019) through research groups, national genetics societies, and direct contacts in LATAM to reach healthcare professionals providing genetic services. English and Spanish versions of our survey are available as supplemental material **(SM1)**.

Snowball sampling was used to expand the survey reach. Inclusion criteria required participants to (1) read and write in Spanish or English and (2) have direct involvement in genetic services in a Latin American country in the last 12 months. Genetic services were defined as ordering genetic testing, evaluating patients with genetic conditions, or providing genetic counseling. To maintain respondent anonymity, no race, ethnicity, or sex data were collected. Data were gathered from May 2024 to November 2024 and analyzed using STATA 18 BE statistical software for descriptive analysis. Inductive thematic analysis was used to summarize and organize qualitative responses into themes for reporting. All free text responses were reviewed by two authors (R.J.G., E.A.) and the themes represented in all responses were agreed upon between the two.

Results

Enrollment and dataset

During the enrollment period we collected 134 responses. After reviewing eligibility and completeness of data, 85 responses (63%) were included in the final analysis. In ten cases, most (>50%), but not all questions were answered. These responses were included in the analysis but may lead to different denominators for different items (75 up to 85). Respondents were predominantly medical geneticists with MD-level training (36/75, 48%), the remainder had a master's degree (17/75, 23%),

doctorate (14/75, 19%), vocational training (3/75, 4%), bachelor's degree (1/75, 1%), or other training (4/75, 5%). Those who selected "Other" reported achieving a medical degree with an additional specialization or degree (Table 1).

Referral information

Respondents selected common reasons for referral to genetic evaluation in their clinical context. The most common referral indications were pediatric genetic conditions (69/85, 81%), cancer (51/85, 60%), adult-onset conditions (42/85, 49%), newborn screening (38/85, 45%), prenatal (32/85, 38%), infertility (32/85, 38%). Write-in responses to the "Other referral" option included after pharmacogenetic testing (3/85, 4%), preconception/infertility (2/85, 2%), and cardiogenetics (1/85, 1%). A wide variety of specialties referred patients to genetics, including pediatrics (66/85, 78%), neurology (59/85, 69%), oncology (51/85, 60%) and obstetrics and gynecology (43/85, 51%). Less common referral sources endorsed by respondents included self-referrals (26/85, 31%), maternal fetal medicine referrals (24/85, 28%) and psychiatry referrals (22/85, 26%).

Genetic testing

The most ordered genetic tests included karyotype (69/84, 82%), panel genetic testing (55/84, 66%), exome sequencing (52/84, 62%) and chromosomal microarray (CMA) (43/84, 51%). The least commonly ordered tests were genome sequencing (16/85, 19%), single gene testing (12/85, 14%) and research-based testing (11/85, 13%). Respondents from Cuba indicated that they do not perform sequencing tests larger than single gene testing. Additional data regarding genetic testing is reported in Table 2.

Table 1 Respondent demographic information. Note: 75 of 85 respondents included in the data set responded to the demographic portion of the survey

Training Level, n (%) *N* = 75		Country, n (%) *N* = 75	
MD	36 (48.0)	Argentina	4 (5.3)
Master's	17 (22.7)	Bolivia	1 (1.3)
Doctorate	15 (18.7)	Brazil	6 (8.0)
Bachelor's	1 (1.3)	Chile	11 (14.7)
Vocational	3 (4.0)	Colombia	8 (10.7)
Other	4 (5.3)	Costa Rica	2 (1.3)
Service Modality, n (%) *N* = 75		Cuba	10 (13.5)
In person	36 (48.0)	Dominican Republic	1 (1.3)
Telemedicine	2 (2.6)	Ecuador	2 (2.7)
Both	33 (44.0)	Guatemala	6 (8,0)
No patient care	4 (5.3)	Honduras	1 (1.3)
Language, n (%) *N* = 75		Mexico	11 (14.7)
Spanish	69 (89.3)	Nicaragua	1 (1.3)
English	13 (17.3)	Panama	3 (4.0)
Portuguese	6 (8.0)	Paraguay	1 (1.3)
French	1 (1.3)	Perú	3 (4.0)
Creole	1 (1.3)	Uruguay	2 (2.7)
Indigenous Language	0 (0.0)	Venezuela	2 (2.7)
		More than one country	0 (0.0)

Table 2 Genetic testing data. *Participants may have selected more than one response; thus, the percentages do not sum to 100%. +Other payment sources include philanthropy and research funding. #Other tests include multiplex ligation-dependent probe amplification (MLPA), triplet primed PCR, fluorescence in situ hybridization (FISH), APOE testing, spectral karyotype (SKY), and chromosome breakage studies

Testing Location* n (%) *N* = 84		Common Genetic Testing Orders* n (%) *N* = 84	
Commercial, out-of-country	49 (58)	Karyotype	69 (82.1)
Commercial, in-country	42 (50)	Panel genetic testing	56 (66.7)
In-house/Hospital lab	29 (34.5)	Exome sequencing	52 (61.9)
Government Lab	20 (23.8)	Chromosomal Microarray (CMA)	43 (51.2)
Payment Methods* n (%) *N* = 84		Other[#]	36 (42.9)
Public health care/government-based insurance	54 (70.9)	Single gene testing	24 (28.6)
Self-pay	50 (62.8)	Genome sequencing	15 (17.9)
Private commercial health insurance	24 (27.9)	Biochemical testing	12 (14.3)
Other[+]	7(8.1)	Research genetic testing	11 (13.1)
Unknown	1 (1.2)		

Respondents indicated that genetic testing was most often performed at out-of-country commercial laboratories (49/84, 58%), followed by in-country commercial laboratories (42/84, 50%) and in-house/hospital laboratories (30/84, 36%). All ten respondents from Cuba performed testing exclusively at a government laboratory. Genetic testing sent out-of-country was performed in the United States (35/47, 75%), Germany (24/47, 51%), Spain (11/47, 23%), South Korea (9/47, 19%), China (2/47, 4%), and Portugal (1/47, 2%). Some testing was also sent out-of-country within South America to Brazil (10/47, 21%), Colombia (4/47, 9%), and Argentina (2/47, 4%). Of respondents who performed genetic testing out-of-country, 32% (15/47) stated all genetic testing they ordered was performed at an out-of-country commercial lab. Respondents described sending genetic testing to another country for the following reasons: cost-effectiveness (29/49, 59%), absence of local testing options (18/49, 37%), lack of local laboratory (10/49, 18%), and lack of certification to perform testing (10/49, 18%).

Barriers and facilitators

We also evaluated the barriers and facilitators to genetic services (Fig. 1A and B**)**. The most selected facilitators were (1) doctors have a positive attitude towards genetic testing (50/84, 60%), (2) patients have a positive attitude towards genetic testing (38/84, 45%), (3) workplace culture encourages genetic testing (37/84, 44%), and (4) partnerships with laboratories (35/84, 42%). Cost was the barrier most selected by respondents (62/84, 74%), followed by lack of clinicians (38/84, 45%), and lack of knowledge about genetic testing (36/84, 43%). Other barriers included distance to a genetic specialty clinic (14/84, 17%), fear of genetic testing (7/84, 8%), cultural or religious beliefs (6/84, 7%), language barriers (5/84, 6%), and lack of awareness from patients (5/84, 6%). Distrust was not selected as a barrier to genetic services by any respondent.

An optional open-response question assessed the area of greatest need from the clinician perspective. A total of 60 respondents disclosed 67 needs that were categorized into five themes. Responses included (1) need for better access to genetic testing (28/67, 42%), (2) need for genetics providers (17/67, 25%), (3) genetic evaluation for a subspecialty (adult, cancer, prenatal, and pediatric) (12/67, 20%), (4) training and technology for local genetic services (6/67, 9%)and (5) government & public recognition (4/69, 6%). Exemplative qualitative responses are reported in Table 3.

Genetic services adequacy

We queried respondents about the adequacy of genetic counseling, genetic testing, medical management, and number of providers through Likert scale questions (Fig. 2.)

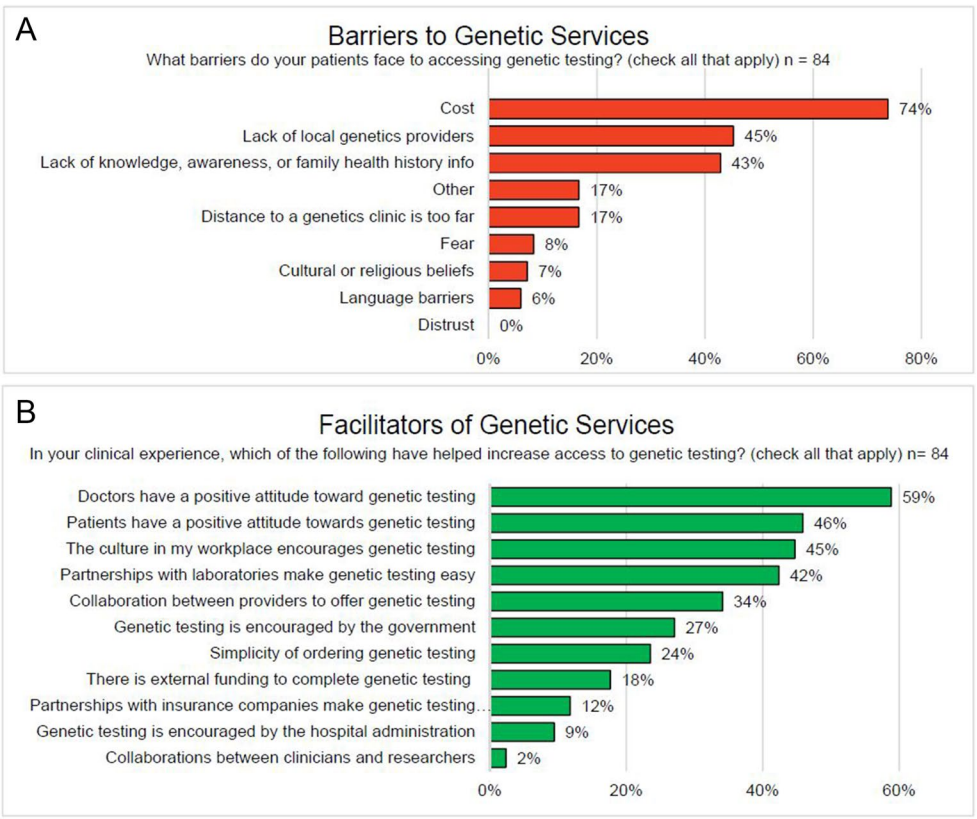

Fig. 1 Barriers and Facilitators of Genetic Services for healthcare providers in LATAM

Table 3 Highlighted clinician quotations from each theme of the areas of need open-response section

Theme	Quotation
Theme 1: Better Access to Genetic Testing	*"I work in the private sector. Not all medical insurances cover genetic studies and the current cost is about $600 dollars. Not everyone can do it."* (Participant 36, Argentina)
Theme 2: Need for genetics providers	"*More geneticists to cover more regions of the country. In addition, we do not have genetic counselors in my country, so the clinical geneticist is the one who also performs this function.*" (Participant 35, Chile)
Theme 3: Specialty-specific Genetic Evaluation	*"In many of the public hospitals there is no genetics service for adults, specialties are not prepared for genetic diseases, and less attention is given to these pathologies. [Referral] can take a long time."* (Participant 119, Mexico)
Theme 4: Training and technology for local genetic services	"*More geneticists are needed. Genetic counsellors are needed. Better quality of testing locally. Lower prices in local labs, [since the] intermediate lab is charging excessive costs. Collaboration for certified training in [the] USA, Canada or Europe is a must have. The government needs to realize the impact of genetic testing."* (Participant 111, Costa Rica)
Theme 5: Government & Public Recognition	"*The government must comply with the existing law […] to insure and pay for genetic testing and treatment of patients [with] rare diseases. [The] government does not do this."* (Participant 129, Panama)

Eighty-seven percent (73/84, 87%) of respondents agreed or strongly agreed that there is a need for additional providers of genetic services in their area. When asked whether there is insufficient demand for genetic services in their area, 73% (61/84) of respondents disagreed or strongly disagreed. Regarding the adequacy of genetic testing, genetic counseling, and medical management, around half of respondents strongly disagreed or disagreed that these needs are adequately met for their patients, (45%, 38/84), (45%, 38/84), and (55%, 46/84), respectively.

Discussion

Our study found that genetic healthcare providers in Latin America feel that cost is the main barrier to accessing genetic services and that more providers are needed to meet patient demand. This provides valuable insight into the availability and utilization of genetic services in LATAM. In a scoping review, (Raspa et al. 2021) identified limited genetics specialists, and poor reimbursement as barriers to genetic services in the United States. These barriers are similar to those facing genetics providers in LATAM. Raspa et al. proposes telehealth genetics services as a model to overcome certain access barriers to genetic services. This model is already in use in LATAM, as reported by 46.6% of our respondents who already provide services via telehealth. Other models

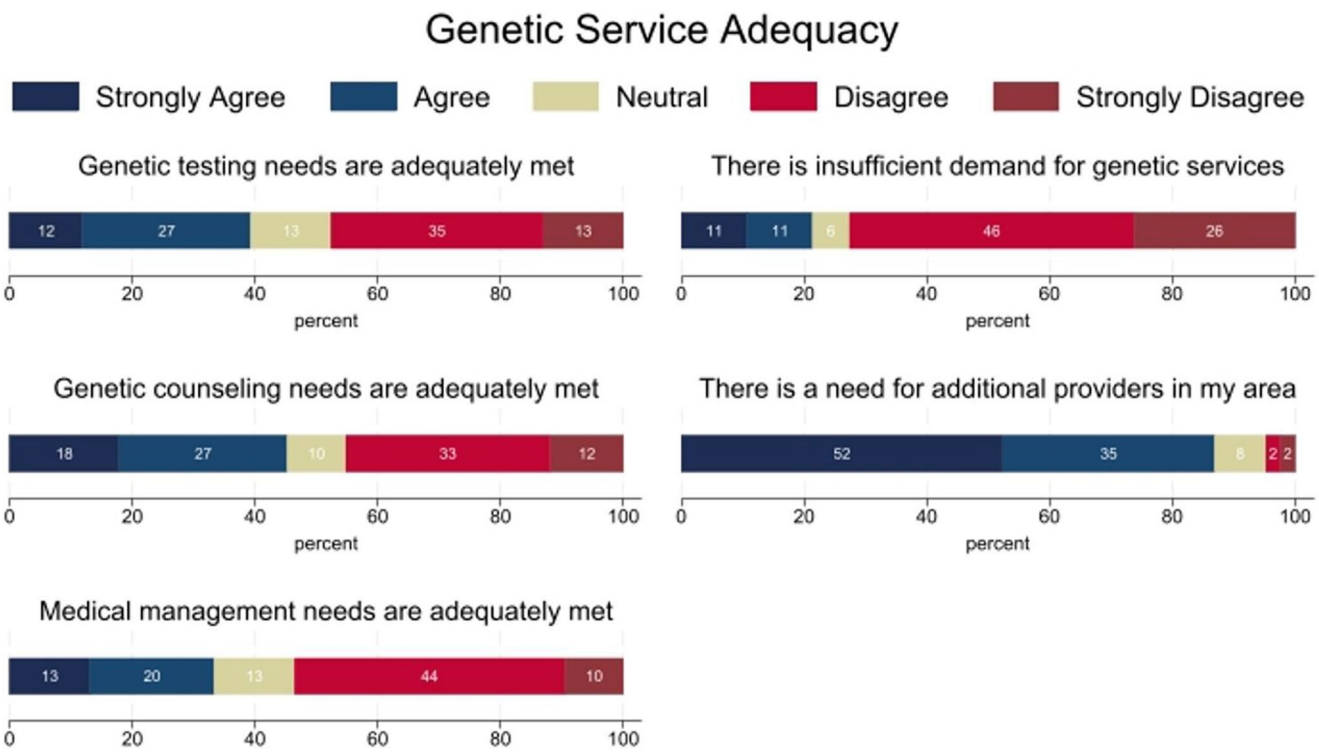

Fig. 2 Clinician-perceived adequacy of genetic testing, genetic counseling, and medical management on a five-point Likert scale

proposed by Raspa et al.(Raspa et al. 2021) include partnerships between genetic and non-genetic healthcare providers, and additional training opportunities. Our respondents likewise identified these as current facilitators that are increasing the access to genetic testing for their patients.

Capacity building is a key part of improving access to genetic services. In resource-limited settings, genetic services are still feasible with creative and customized solutions that fit the communities they intend to serve. One such example is a pilot implementation of genomic risk assessment for cancer risk and genetic services sustainment in Mexico by Blazer et al. (Blazer et al. 2021). They worked within the context of the Mexican public healthcare system and were able to offer online training to clinicians and low-cost targeted analysis of *BRCA1* and *BRCA2*. With the increased local capacity, the clinicians were able to identify 195 patients with *BRCA1* or *BRCA2* pathogenic variants. With this information they can consider risk reducing surgeries and targeted therapy options that may be beneficial for both the patient's health and the overall cost savings to the healthcare system. Capacity building can serve local patients for decades to come and build a foundation for future training and investment in genetic services. Currently, the main avenue for a healthcare provider to offer genetic services is training for physicians to specialize in genetics (Bucio et al. 2019; De Lima et al. 2024; Diaz Caro and Simone 2024; Margarit et al. 2013; Rodas-Pérez et al. 2015), which could explain the high number of physician respondents to our survey. In Latin America, master's level courses in genetic counseling are only available in Cuba (Roblejo Balbuena and Marcheco Teruel 2017) and Brazil (Ormond et al. 2018). In lieu of a master's degree or medical fellowship for physicians to specialize in genetics, certificates programs and graduate diplomas in genetic counseling or cancer genetics(Margarit et al. 2013) have attracted Latin American healthcare providers who see a need for genetics knowledge to better serve their patients.

Of interest, distrust was not selected as a barrier to genetic services by any respondent. Previous research about barriers to genetic services for Latin American patients based in the United States has reported medical distrust as a major barrier to genetic services (Hamilton et al. 2016; Hann et al. 2017; Ramirez et al. 2015). This could be an intrinsic feature of our study population, who are healthcare providers, rather than patients receiving genetic services as in other studies or could reflect a difference in perspectives between US-based Latin American patients and patients in Latin American countries. Another possibility is that this is the result of language and cultural concordance between Latin American healthcare providers and their patients, which is less common among Latin American individuals in US-based studies. Ethnic, linguistic, and cultural concordance have been shown to benefit the patient-clinician relationship (Daggett et al. 2023; Diaz et al. 2024).

Our study must be considered in the light of its limitations. While we had representation from eighteen countries, these results may not be generalizable because of the small number of responses from each country. However, it is worth noting that in some cases, even a small sample size in a country may represent many of the licensed clinical geneticists working there. For example, we received two responses from Costa Rican healthcare providers, one of which stated that there are only three clinical geneticists working there. This number matched the number of practicing clinical geneticists listed on the Costa Rican College of Physicians and Surgeons register of medical providers as of April 2025. This may be the case in other countries as well, meaning this data may be more representative than it appears with a small sample size. Similarly, the inclusion of high response countries (Chile, Mexico, Cuba) may cause averages across the total sample to not reflect the reality at a country level. Further, responses from Cuba were vastly different with respect to access to genetic services compared to other countries. Respondents from Cuba highlighted the adequacy of genetic providers and training, historical embargos affecting the available genetic tests, and exclusive use of in-country government laboratories. These differences in access to genetic services between Cuba and other Latin American countries are well documented (Ormond et al. 2024; Roblejo Balbuena and Marcheco Teruel 2017).

Additionally, we recognize that we are likely missing responses from Portuguese-speaking Brazil since our survey was only offered in English and Spanish. As the landscape of genetic services in large Brazilian cities is well documented (Bonilla et al. 2022; Horovitz et al. 2024), we did not expect to advance the aims of our study by actively recruiting Portuguese-speaking providers in Brazil. A special issue on community genetics in Brazil was recently published in the Journal of Community Genetics covering advancements in healthcare policy, access to genetic services, and research efforts which provides up-to-date information on the landscape of genetic services in Brazil. In contrast to the much of LATAM, this special issue reports that Brazil has an established and expanding NBS program (De Souza et al. 2025), robust policy and reference centers for rare disease (Félix et al. 2024; Giugliani et al. 2025). However, though comparatively advanced, these programs are not without challenges. Our study identifies similar barriers to those reported in Brazil by Rodrigues et al. 2024; including lack of expertise, laboratories, and patient advocacy groups (Rodrigues et al. 2024).

Self-selection bias may have affected our study, as participants with a vested interest in this topic area may be more likely to respond. The recruitment methods used may

overrepresent clinical geneticists with MD-level training, since non-physician genetics providers may be less likely to be affiliated with national genetics societies or research studies. However, other survey-based studies have shown a growing number of non-physician healthcare providers offering genetic services with a variety of training levels (Fernández-Ramires, Morales-Pison, Rucatti, Echeverría, San Martín, Cammarata et al., 2024). We recognize that across LATAM, healthcare access is variable depending on factors such as geography, economic development, political stability, and national priorities. At this time, genetics services are available to some extent in every country which had a respondent, though there are myriad differences between them, not all of which could be captured in this study.

Collaboration across borders can aid in developing local infrastructure and effective implementation of genetic services. Some concrete examples of this in low-resource settings are: (1) leveraging funding opportunities from the World Health Organization (WHO) and Rare Disease International (RDI) as well as guidance on research frameworks and practices; (2) regional meetings focused on action that bring together key stakeholders to address local barriers and advocate for the resolution of access issue; (3) pilot programs that demonstrate cost-effectiveness and clinical benefits and incentivize further adoption and investment by local governments; (4) existing training opportunities that serve as templates for developing accessible and appropriate genetics education programs. International societies of genetics professionals such as the Latin American Professional Society for Genetic Counseling (SPLAGen) (Diaz Caro and Simone 2024), and the Latin-American Network of Human Genetics (RELAGH) (Rojas-Martínez et al. 2014) are a keystone to developing efficacious local training and infrastructure for genetic counselors, clinical geneticists, and laboratory staff. This professional community nurtures and supports local professionals who understand the context, language, and needs of LATAM communities.

Future directions

Future studies may include country-specific assessments or surveys to investigate the landscape of genetic services in regions not well captured in this study. Additionally, the utilization of genetic services in healthcare in less urbanized areas, outside regional capitals, remains unstudied in many countries. Indeed, many of our respondents noted that they provide services in the national or provincial capital, although almost half of respondents offer telemedicine appointments, which may extend their reach to more rural areas. Lastly, a study of the economic viability, benefits and limitations of performing genetic testing in-country versus out-of-country could prove valuable in guiding the development of local genetics infrastructure such as training programs, laboratories, and/or genetic testing partnerships within LATAM. Such a study may be able to identify necessary sample volumes and reimbursement levels that would make local genetic testing a viable option in a variety of scenarios with the goal of reducing cost, thus lowering a major barrier to genetic services identified in this study.

Overall, building local capacity is essential to increasing access to genetic services in LATAM. We describe the existing landscape with the goal of awareness and recognition of genetic services and provide context for future studies. Public recognition and government buy-in is crucial to advancing access to genetic services within the context of each country and their healthcare system. As genetic technologies continue to advance, it is crucial to support a sustainable and locally driven future for accessible genetic services in LATAM. Whether a patient has an inherited cancer predisposition, rare genetic disease, congenital anomaly, or adult-onset neurological disease; local genetic testing and evaluation should be an attainable option regardless of geography. By implementing genetic services that leverage local facilitators, systemic barriers can be overcome, diagnostic delays can be reduced, and unnecessary costs can be avoided; at the same time improving patient health outcomes and drive long-term savings for the healthcare system.

Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/s12687-025-00832-0.

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Author contributions Conceptualization: R.J.G.; Data curation: R.J.G.; Formal analysis: R.J.G, E.A., P.J.L.; Resources: S.P., E.A.S., S.M.; Methodology: S.P, P.J.L, E.A.S., E.A.; Supervision: S.P; Visualization: R.J.G, P.J.L.;Writing-original draft: R.J.G.; Writing-review & editing: R.J.G., S.P., E.A., E.A.S, S.M., C.S., P.J.L.

Data availability The data obtained and reported in this study are available on a case-by-case basis to maintain the privacy of the respondents. Requests may be submitted to the corresponding author.

Declarations

Ethics This project was approved by the Baylor College of Medicine Institutional Review Board (Protocol Number: H-55131). This project was granted a waiver of documentation of written consent and informed consent was indicated by the respondent's continuation beyond the introduction and screener questions.

Conflict of interest Dr. Storch reports receiving research funding to his institution from the Ream Foundation, International OCD Foundation, and National Institutes of Health. He was a consultant for Brainsway and Biohaven Pharmaceuticals in the past 12 months. He owns stock less than $5000 in NView (for distribution of the Y-BOCS and CY-BOCS). He receives book royalties from Elsevier, Wiley, Oxford, American Psychological Association, Guildford, Springer, Routledge, and Jessica Kingsley.

References

Abacan M, Alsubaie L, Barlow-Stewart K, Caanen B, Cordier C, Courtney E, Davoine E, Edwards J, Elackatt NJ, Gardiner K, Guan Y, Huang L, Malmgren CI, Kejriwal S, Kim HJ, Lambert D, Lantigua-Cruz P, Lee JMH, Lodahl M, Wicklund C (2019) The global state of the genetic counseling profession. Eur J Hum Genet 27(2):183–197. https://doi.org/10.1038/s41431-018-0252-x

Allen CG, Hunt KJ, McMahon LL, Thornhill C, Jackson A, Clark JT, Kirchoff K, Garrison KL, Foil K, Malphrus L, Norman S, Ramos PS, Perritt K, Brown C, Lenert L, Judge DP (2024) Using implementation science to evaluate a population-wide genomic screening program: findings from the first 20,000 in our DNA SC participants. Am J Hum Genet 0002–0001. https://doi.org/10.1016/j.ajhg.2024.01.004

Blazer KR, Chavarri-Guerra Y, Garza CV, Nehoray B, Mohar A, Daneri-Navarro A, Toro Ad, Aguilar D, Arteaga J, Álvarez RM, Mejia R, Herzog J, Castillo D, Fernandez M, Weitzel JN (2021) Development and pilot implementation of the genomic risk assessment for cancer implementation and sustainment (GRACIAS) intervention in Mexico. JCO Global Oncol 7(7). https://doi.org/10.1200/GO.20.00587

Bonilla C, Sortica A, Schuler-Faccini V, Matijasevich L, A., Scheffer MC (2022) Medical geneticists, genetic diseases and services in Brazil in the age of personalized medicine. Personalized Med 19(6):549–563. https://doi.org/10.2217/pme-2021-0153

Bucio D, Ormond KE, Hernandez D, Bustamante CD, Lopez Pineda A (2019) A genetic counseling needs assessment of Mexico. Mol Genet Genomic Med 7(5):e668. https://doi.org/10.1002/mgg3.668

Ceyhan-Birsoy O, Murry JB, Machini K, Lebo MS, Yu TW, Fayer S, Genetti CA, Schwartz TS, Agrawal PB, Parad RB, Holm IA, McGuire AL, Green RC, Rehm HL, Beggs AH (2019) Interpretation of genomic sequencing results in healthy and ill newborns: results from the babyseq project. Am J Hum Genet 104(1):76–93. https://doi.org/10.1016/j.ajhg.2018.11.016

Chen F, Ahimaz P, Wang K, Chung WK, Ta C, Weng C, Liu C (2023) Phenotype-driven molecular genetic test recommendation for diagnosing pediatric rare disorders. *Research Square*,, rs.3.rs–3593490. https://doi.org/10.21203/rs.3.rs-3593490/v1

Daggett A, Abdollahi S, Hashemzadeh M (2023) The effect of Language concordance on health care relationship trust score. Springer Science and Business Media LLC. https://doi.org/10.7759/cureus.39530

Damschroder LJ, Lowery JC (2013) Evaluation of a large-scale weight management program using the consolidated framework for implementation research (CFIR). Implement Sci 8(1):51–51. https://doi.org/10.1186/1748-5908-8-51

De Castro M, Restrepo CM (2015) Genetics and genomic medicine in Colombia. Mol Genet Genomic Med 3(2):84–91. https://doi.org/10.1002/mgg3.139

De Lima FT, De Lima MA, Correia PS, Honjo RS, Maia RE, Kyosen SO, Melo DG (2024) Teaching and training of human resources for genetics and genomics in Brazil. Springer Science and Business Media LLC. https://doi.org/10.1007/s12687-024-00726-7

De Rosa MA, Bernardi MT, Kleppe S, Walz K (2024) Hearing loss: genetic testing, current advances and the situation in Latin America. Genes 15(2):178. https://doi.org/10.3390/genes15020178

De Souza CFM, Tonon T, Silva TO, Bachega TASS (2025) Newborn screening in Brazil: realities and challenges. J Community Genet. https://doi.org/10.1007/s12687-024-00762-3

Diaz A, Castillo Tafur JC, Lin Y, Echenique DB, Drake B, Choubey AS, Mejia A, Gonzalez MH (2024) Education, language, and cultural concordance influence patient-physician communication in orthopaedics. Ovid Technologies. https://doi.org/10.2106/jbjs.24.00167. (Wolters Kluwer Health)

Diaz Caro D, Simone L (2024) The role of the Latin American professional society of genetic counseling (SPLAGen): advancing genetic counseling in Latin America. Genet Med Open 101870. https://doi.org/10.1016/j.gimo.2024.101870

Dimmock D, Caylor S, Waldman B, Benson W, Ashburner C, Carmichael JL, Carroll J, Cham E, Chowdhury S, Cleary J, D'harlingue A, Doshi A, Ellsworth K, Galarreta CI, Hobbs C, Houtchens K, Hunt J, Joe P, Joseph M, Farnaes L (2021) Project baby bear: rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. Am J Hum Genet. https://doi.org/10.1016/j.ajhg.2021.05.008

Félix TM, Oliveira BMD, Horovitz DDG (2024) Building a national policy for rare disease in Brazil. Springer Sci Bus Media LLC. https://doi.org/10.1007/s12687-024-00732-9

Fernández-Ramires R, Morales-Pison S, Rucatti GG, Echeverría C, San Martín E, Cammarata F, Salas-Burgos A, Adorno-Farias D, González-Arriagada WA, Benavides C, Espinosa-Parrilla Y, Zapata D, Norese G, Lázaro C, González S, Pujana MA, Sullcahuaman Y, Margarit S (2024a) Cancer genetic counseling in chile: addressing barriers, confronting challenges, and seizing opportunities in an underserved Latin American community. Genet Med Open 101898. https://doi.org/10.1016/j.gimo.2024.101898

Fernández-Ramires R, Morales-Pison S, Rucatti GG, Echeverría C, San Martín E, Cammarata-Scalisi F, Salas-Burgos A, Adorno-Farias D, González-Arriagada WA, Espinosa-Parrilla Y, Zapata-Contreras D, Norese G, Lázaro C, González S, Pujana MA, Sullcahuaman Y, Margarit S (2024b) Cancer genetic counseling in chile: addressing barriers, confronting challenges, and seizing opportunities in an underserved Latin American community. Genet Med Open 2(Suppl 2):101898. https://doi.org/10.1016/j.gimo.2024.101898

Giugliani R, Castillo Taucher S, Hafez S, Oliveira JB, Rico-Restrepo M, Rozenfeld P, Zarante I, Gonzaga-Jauregui C (2022) Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. Front Genet. https://doi.org/10.3389/fgene.2022.1053559

Giugliani R, De Oliveira BM, Guaraná BB, Tonon T, Trapp FB, Costa D, Silva FMD, Baldo LP, Riegel-Giugliani G, Barbosa M, Daher F, A., De Souza C (2025) Redefining the approach to rare diseases: the experience of Casa Dos Raros in Brazil. Springer Science and Business Media LLC. F. Mhttps://doi.org/10.1007/s12687-025-00771-w

Guio H, Poterico JA, Levano KS, Cornejo-Olivas M, Mazzetti P, Manassero-Morales G, Ugarte-Gil MF, Acevedo-Vásquez E, Dueñas-Roque M, Piscoya A, Fujita R, Sanchez C, Casavilca-Zambrano S, Jaramillo-Valverde L, Sullcahuaman-Allende Y, Iglesias-Pedraz JM, Abarca-Barriga H (2018) Genetics and genomics in Peru: clinical and research perspective. Mol Genet Genom Med 6(6):873–886. https://doi.org/10.1002/mgg3.533

Hackl M, Cook L, Wetherill L, Walsh LE, Delk P, De León R, Carbonell J, Vicioso RC, Hodges PD (2023) Readiness for Parkinson's disease genetic testing and counseling in patients and their relatives in urban settings in the Dominican Republic. NPJ Parkinsons Dis 9(1):126. https://doi.org/10.1038/s41531-023-00569-y

Hamilton JG, Shuk E, Arniella G, González CJ, Gold GS, Gany F, Robson ME, Hay JL (2016) Genetic testing awareness and attitudes among Latinos. Public Health Genomics 19(1):34–46. https://doi.org/10.1159/000441552

Hann KEJ, Freeman M, Fraser L, Waller J, Sanderson SC, Rahman B, Side L, Gessler S, Lanceley A (2017) Awareness, knowledge, perceptions, and attitudes towards genetic testing for cancer risk among ethnic minority groups: a systematic review. BMC Public Health 17(1):503. https://doi.org/10.1186/s12889-017-4375-8

Harris PA, Taylor R, Thielke R, Payne J, Gonzalez N, Conde JG (2009) Research electronic data capture (REDCap)—a metadata-driven methodology and workflow process for providing translational research informatics support. J Biomed Inform 42(2):377–381. https://doi.org/10.1016/j.jbi.2008.08.010

Harris PA, Taylor R, Minor BL, Elliott V, Fernandez M, O'Neal L, McLeod L, Delacqua G, Delacqua F, Kirby J, Duda SN (2019) The REDCap consortium: Building an international community of software platform partners. J Biomed Inform 95:103208. https://doi.org/10.1016/j.jbi.2019.103208

Hendricks-Sturrup RM, Linsky A, Lu CY, Vassy JL (2020) Genomic testing is best integrated into clinical practice when it is actionable. Personalized Med 17(1):5–8. https://doi.org/10.2217/pme-2019-0106

Horovitz DDG, Félix TM, Ferraz VE, d. F (2024) Medical genetics in Brazil in the 21st century: A thriving specialty and its incorporation in public health policies. Genes 15(8):973. https://doi.org/10.3390/genes15080973

Jenkins BD, Fischer CG, Polito CA, Maiese DR, Keehn AS, Lyon M, Edick MJ, Taylor MRG, Andersson HC, Bodurtha JN, Blitzer MG, Muenke M, Watson MS (2021) The 2019 US medical genetics workforce: a focus on clinical genetics. Genet Med 23(8):1458–1464. https://doi.org/10.1038/s41436-021-01162-5

Kim N, Kong S, Yoo J, Kim D, Seo SH, Kim J (2022) Current issues, challenges, and future perspectives of genetic counseling in Korea. Ann Lab Med 42(3):314–320. https://doi.org/10.3343/alm.2022.42.3.314

Larrandaburu M, Matte U, Noble A, Olivera Z, Sanseverino MTV, Nacul L, Schuler-Faccini L (2015) Ethics, genetics and public policies in Uruguay: newborn and infant screening as a paradigm. J Community Genet 6(3):241–249. https://doi.org/10.1007/s12687-015-0236-2

MacFarland SP, Maese L, Rednam SP, Kamihara J, Perrino MR, Nichols KE, Brodeur GM, Schiffman JD, Plon SE, Diller LR, Malkin D, Porter CC, Villani A (2022) Collaboration to promote research and improve clinical care in the evolving field of childhood cancer predisposition. Cancer Prev Res (Philadelphia Pa) 15(10):645–652. https://doi.org/10.1158/1940-6207.CAPR-22-0215

Margarit SB, Alvarado M, Alvarez K, Lay-Son G (2013) Medical genetics and genetic counseling in Chile. J Genet Couns 22(6):869–874. https://doi.org/10.1007/s10897-013-9607-1

Migliavacca MP, Sobreira J, Bermeo D, Gomes M, Alencar D, Sussuchi L, Souza CA, Silva JS, Kroll JE, Burger M, Guarischi-sousa R, Villela D, Yamamoto GL, Milanezi F, Horigoshi N, Cesar RG, De Carvalho WB, Honjo RS, Bertola DR, Sobreira N (2024) Whole genome sequencing as a first-tier diagnostic test for infants in neonatal intensive care units: A pilot study in Brazil. Wiley. https://doi.org/10.1002/ajmg.a.63544

Ormond KE, Laurino MY, Barlow-Stewart K, Wessels T, Macaulay S, Austin J, Middleton A (2018) Genetic counseling globally: where are we now? Am J Med Genet C Semin Med Genet 178(1):98–107. https://doi.org/10.1002/ajmg.c.31607

Ormond KE, Abad PJ, Macleod R, Nishigaki M, Wessels T (2024) The global status of genetic counseling in 2023: what's changed in the past 5 years? Elsevier BV. https://doi.org/10.1016/j.gimo.2024.101887

Paz y Miño C, Sacoto G, Leone PE (2015) Genetics and genomic medicine in ecuador. Mol Genet Genomic Med 4(1):9–17. https://doi.org/10.1002/mgg3.192

Penchaszadeh VB (2013) Genetic testing and services in Argentina. J Community Genet 4(3):343–354. https://doi.org/10.1007/s12687-012-0093-1

Powell S, Artigas M, Borovova I, Gadiya P, Hsu A, Kaur R, Kidd L, Rosenfeld D, Saeed MM, Scarelli E, Youssef MW (2024) Magenta: a multinational patient survey assessing the awareness, perceptions and unmet needs in genetic testing and counselling among patients with breast cancer. Front Oncol 14:1380349. https://doi.org/10.3389/fonc.2024.1380349

Pritchard D, Goodman C, Nadauld LD (2022) Clinical utility of genomic testing in cancer care. JCO Precision Oncol 6:e2100349. https://doi.org/10.1200/PO.21.00349

Ramirez AG, Chalela P, Gallion KJ, Muñoz E, Holden AE, Burhansstipanov L, Smith SA, Wong-Kim E, Wyatt SW, Suarez L (2015) Attitudes toward breast cancer genetic testing in five special population groups. J Health Disparities Res Pract 8(4):124–135. https://pmc.ncbi.nlm.nih.gov/articles/PMC4739821/

Rao SK, Blakely LJ, Small K, Schleicher S, Natalie RD (2024) Building efficiency and scaling with a remote genetic counseling program. Oncology 38(1):20–25. https://doi.org/10.46883/2024.25921011

Raspa M, Moultrie R, Toth D, Haque SN (2021) Barriers and facilitators to genetic service delivery models: scoping review. Interact J Med Res 10(1):e23523. https://doi.org/10.2196/23523

Roblejo Balbuena H, Marcheco Teruel B (2017) Genetics and genomic medicine in Cuba. Mol Genet Genom Med 5(3):196–201. https://doi.org/10.1002/mgg3.299

Rodas-Pérez C, Clarke A, Powell J, Thorogood M (2015) Challenges for providing genetic counselling in Colombian genetic clinics: the viewpoint of the physicians providing genetic consultations. J Community Genet 6(3):301–311. https://doi.org/10.1007/s12687-015-0237-1

Rodrigues G, Poletto E, Vairo E, F. P., Baldo G (2024) Basic and translational research in rare diseases in low- and middle-income countries: challenges and solutions. Springer Science and Business Media LLC. https://doi.org/10.1007/s12687-024-00759-y

Rojas-Martínez A, Giraldo-Ríos A, Jiménez-Arce G, de Vargas AF, Giugliani R (2014) RELAGH - the challenge of having a scientific network in Latin america: an account from the presidents. Genet Mol Biology 37(1 Suppl):305–309. https://doi.org/10.1590/s1415-47572014000200017

Szigety KM, Crowley TB, Gaiser KB, Chen EY, Priestley JRC, Williams LS, Rangu SA, Wright CM, Adusumalli P, Ahrens-Nicklas RC, Calderon B, Cuddapah SR, Edmondson A, Ficicioglu C, Ganetzky R, Kalish JM, Krantz ID, McDonald-McGinn

DM, Medne L, Sheppard SE (2022) Clinical effectiveness of telemedicine-based pediatric genetics care. Pediatrics 150(1):e2021054520. https://doi.org/10.1542/peds.2021-054520

Thong M, See-Toh Y, Hassan J, Ali J (2018) Medical genetics in developing countries in the Asia-Pacific region: challenges and opportunities. Genet Med 20(10):1114–1121. https://doi.org/10.1038/s41436-018-0135-0

Toland AE, Forman A, Couch FJ, Culver JO, Eccles DM, Foulkes WD, Hogervorst FBL, Houdayer C, Levy-Lahad E, Monteiro AN, Neuhausen SL, Plon SE, Sharan SK, Spurdle AB, Szabo C, Brody LC (2018) Clinical testing of BRCA1 and BRCA2: a worldwide snapshot of technological practices. NPJ Genom Med. https://doi.org/10.1038/s41525-018-0046-7

Torre-Hernandez CADL, Guedez Y, Pineda-Bernal L, Ojeda HA, Guevara-Guerra YA (2018) Los servicios de genética médica En Venezuela. Rev Panam Salud Publica 42:e78. https://doi.org/10.26633/RPSP.2018.78

Unim B, Pitini E, Lagerberg T, Adamo G, De Vito C, Marzuillo C, Villari P (2019) Current genetic service delivery models for the provision of genetic testing in europe: a systematic review of the literature. Front Genet 10:552. https://doi.org/10.3389/fgene.2019.00552

Vishnopolska SA, Turjanski AG, Herrera Piñero M, Groisman B, Liascovich R, Chiesa A, Marti MA (2018) Genetics and genomic medicine in Argentina. Mol Genet Genomic Med 6(4):481–491. https://doi.org/10.1002/mgg3.455

Wainstock D, Katz A (2023) Advancing rare disease policy in Latin America: a call to action. Elsevier BV. https://doi.org/10.1016/j.lana.2023.100434

Wang S, Liu K, Yang H, Ma J (2022) A cost-effectiveness analysis of screening strategies involving non-invasive prenatal testing for trisomy 21. Front Media SA. https://doi.org/10.3389/fpubh.2022.870543