Genetics Services in Latin America: A Descriptive Study of Availability and Utilization of Genetics in Healthcare

Ryan J. German[1], Erin Atkinson[2], Eric A. Storch[3], Claudia Soler-Alfonso[4], Sonia Margarit[5], Philip J. Lupo [6,7,8], Stacey Pereira[9]

1) Baylor College of Medicine, Genetic Counseling Program 2) Texas Children's Hospital 3) Baylor College of Medicine, Department of Psychiatry and Behavioral Sciences 4) Cook Children's Genetics Center 5) Center for Genetics and Genomics, Universidad del Desarrollo 6) Baylor College of Medicine, Department of Pediatrics, Section of Hematology-Oncology7) Emory University School of Medicine, Department of Pediatrics, 8) Children's Healthcare of Atlanta, Aflac Cancer and Blood Disorders Center, 9) Baylor College of Medicine, Center for Medical Ethics & Health Policy

Introduction

Genetic services are expanding globally, but access remains limited in low-resource regions such as Latin America (LATAM) and have not been well described in recent medical literature. Understanding current service availability, barriers, and facilitators is critical to guide capacity building and improve patient care.

We defined genetic service as:

- Order genetic testing
- Evaluating patients with genetic conditions
- Providing genetic counseling

Genetic services are seen in a wide variety of clinical specialties such as oncology, neurology, cardiology, pediatrics, and psychiatry. National programs such as population screening for high prevalence conditions or newborn screening are common instances of genetics integration into healthcare.

Objectives

Catalogue genetic service availability across LATAM

Identify barriers and facilitators to service delivery

Assess clinician-perceived adequacy and needs

Methods

Design:

- Cross-sectional, web-based survey via REDCap.
- Intended to take <10 minutes
- 25 questions with branching logic

Participants: Healthcare professionals providing genetics services in LATAM

Spanish/English Survey: Brazil included with a focus on Spanish speaking countries, based on identified knowledge gap.

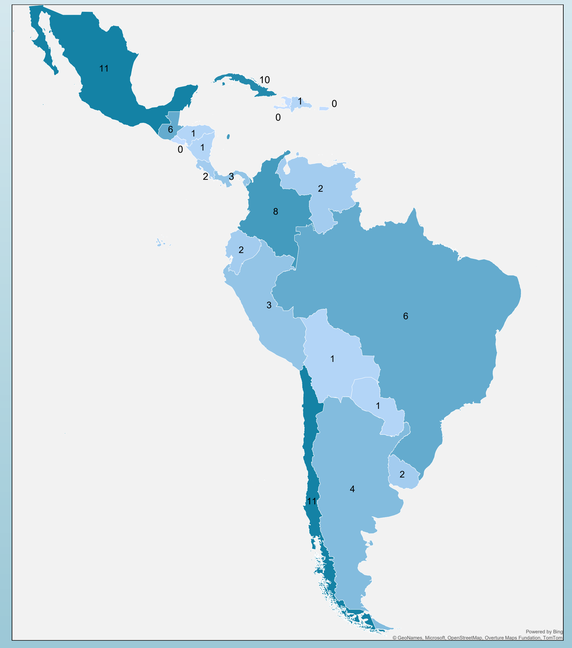
Key Survey Domains:

1. Patient Population
2. Genetic Testing
3. Barriers and Facilitators (CFIR)
4. Adequacy of Genetic Services
5. Demographic Information

Qualitative Questions: Thematic analysis used to allow for more nuance and context.

Results

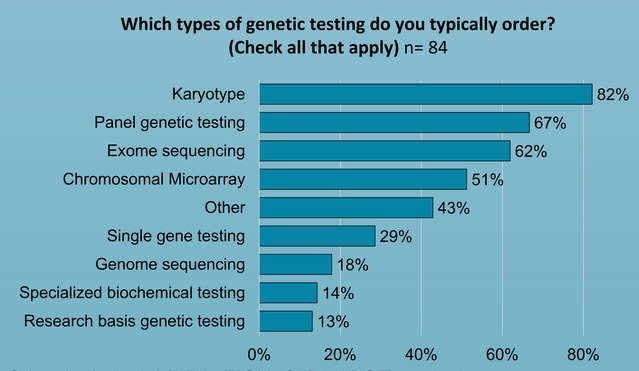
- Eighty-five respondents from 18 countries.
- **Referrals:** pediatric, cancer, adult-onset genetic conditions, pharmacogenetics, and prenatal testing.
- **Respondents:** 91% practice as a specialist in genetics



Testing Location

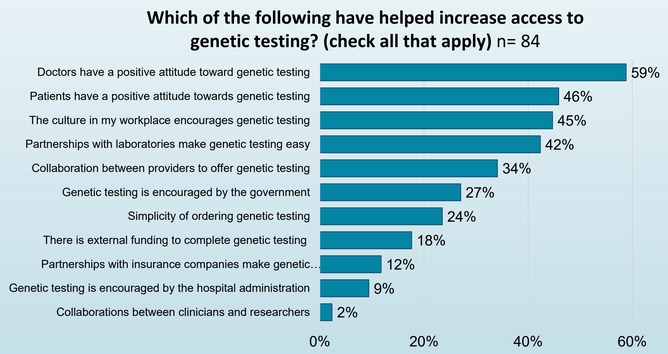
- Testing was commonly performed a commercial labs both in-country (49/84) and out-of-country (42/84). In house or hospital laboratories was selected by over a third of respondents (30/84).
- Government labs were not commonly used except in Cuba

Common Genetic Testing Orders

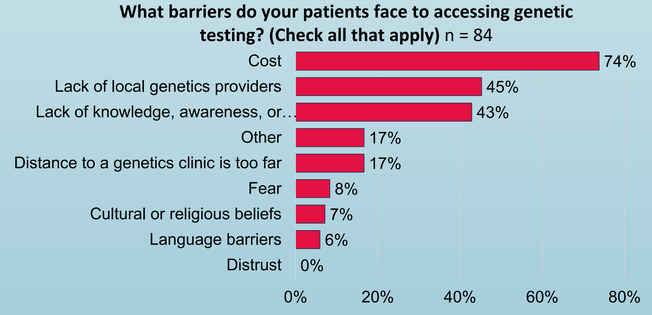


Other included: MLPA, FISH, SKY, APOE, and chromosome breakage studies

Facilitators of Genetic Services



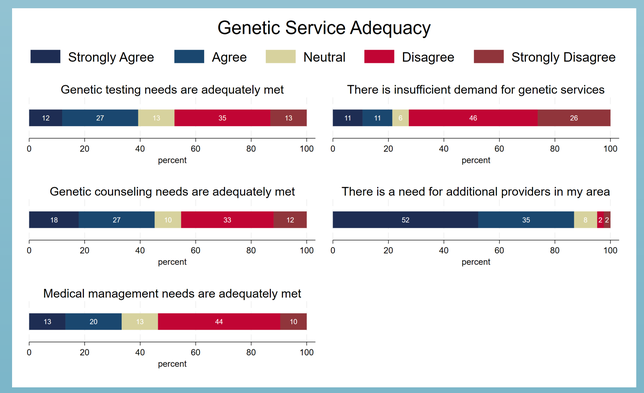
Barriers to Genetic Services



Perceived Needs and Adequacy

Thematic Analysis of Needs (n=69)

1. Better Access to Genetic Testing (38%)
2. Need for Genetics Providers (26%)
3. Specialty-specific Genetic Evaluation (20%)
4. Government & Public Recognition (9%)
5. Training &Technology for Local Genetic Services (7%)



45% disagree that genetic testing needs are adequate
45% disagree that genetic counseling needs are adequate
55% disagree that medical management needs are adequate
72% disagreed that there was insufficient demand
87% agreed that there is a need for additional genetics providers

Conclusions

Key Takeaways

- Genetic services are available in Latin America
- Cost and accessibility barriers similar to USA
- Support for genetic services in Latin America

- Local capacity building through international collaboration and training can support genetic services.
- Public and government recognition are necessary improve access to genetic services.
- Genetic services in Latin America show substantial growth but face persistent barriers.

Future Directions

In future studies we propose:

- Better coverage of non-geneticists who provide genetic services.
- Country specific studies to address the nuanced geographic and systemic differences between nations.
- Qualitative study of patient experiences with genetic services in Latin America

Needs Assessment
What interventions can be used to improve access to genetic services in each local or national health system?

Access in Rural Areas
Are genetic services available outside of regional or national capitals?

Economic Evaluation
Is in-country testing or regional partnerships to lower testing cost economically viable?

Acknowledgements

This research was completed as a part of a master's thesis at the Baylor College of Medicine Genetic Counseling Program. Research reported in this publication was supported by the National Institute of Mental Health (U01MH125062). The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.