

Status of Genetic Services in Puerto Rico: A Qualitative Analysis

by

Coralys Marie Carcana Barbosa

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This essay is submitted

by

Coralys Marie Carcana Barbosa

on

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and approved by

Essay Advisor: Andrea Durst, MS, DrPH, LCGC, Assistant Professor of Human Genetics, Associate Director of the Genetic Counseling Program, Director of the MPH in Public Health Genetics Program, School of Public Health, University of Pittsburgh

Essay Reader: Thistle Elias, DrPH, MPA, Assistant Professor, Department of Behavioral and Community Health Sciences, School of Public Health, University of Pittsburgh

Essay Reader: Alissa Terry, ScM, CGC, Genetic Services Outreach Coordinator and Telegenetics Navigator for the New York Mid-Atlantic Caribbean Regional Genetics Network

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Abstract

Genetic services (GS) serve as an important resource in diagnosing and advising individuals with risk factors for genetic conditions. However, serious gaps remain in providing needed genetic services in Puerto Rico (PR), along with many other areas. The New York Mid-Atlantic Caribbean (NYMAC) Regional Genetics Networks (RGN) aims to collaborate with key stakeholders to create access to quality care and appropriate genetic services. This needs assessment aims to describe the current status of genetic services in PR to identify barriers and potential solutions. NYMAC gathered Puerto Rican stakeholders based on their relation to genetics needs and services and held virtual recorded meetings to gain a better understanding of genetic services in PR and its impact on public health professionals and patients. Herein, we summarize and analyze four meeting transcripts between NYMAC and stakeholders in Puerto Rico using a grounded thematic qualitative analysis. The results identified themes for barriers and solutions for health care professionals, patients, families, and professionals that work with them. These results suggest that expanding access and availability to genetic services, telemedicine and interpreter services can begin overcoming the barriers that exist. Challenges and limitations exist when integrating and improving GS in PR. The public health importance of this needs assessment is that it highlights health disparities that exist in the PR population and describes potential solutions.

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Preface

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Abbreviations

ACA – Affordable Care Act
APHL – Association of Public Health Laboratories
CCHD – Critical Congenital Heart Defects
CDC – Center for Disease Control and Prevention
CSHCN – Child with Special Health Care Needs
DoH – Department of Health
FMAP – Federal Matching Assistance Percentage
GS – Genetic Services
HRSA – Health Resources and Services Administration
MIECHV – Maternal, Infant, and Early Childhood Home Visiting
NBS – Newborn Screening
NYMAC – New York Mid-Atlantic Caribbean
PR – Puerto Rico
RGN – Regional Genetics Network
SERN – Southeast Regional Network
SES – Socioeconomic status
U.S. – United States of America
USVI – United States Virgin Islands

1.0 Introduction

Genetic services (GS) are an important resource in diagnosing and guiding individuals with risk factors for genetic conditions. GS have been especially useful in identifying a diagnosis and providing care that best supports the quality of life for an individual. However, barriers exist that limit access to GS, such as lack of genetic knowledge, access to genetic providers, medical coverage, long wait times, increased average new patient caseloads, and language barriers (Cornel and Van El, 2017; Maiese et al. 2019). These barriers appear more prominently in Puerto Rico (PR), where there are few genetic providers, constrained medical coverage and limited genetic knowledge that strongly affects this underserved community.

PR is a United States (U.S.) territory, with a population size of almost 3.2 million individuals (U.S. Census Bureau, 2019). However, there are a total of five genetic professionals in the entire island: four geneticists and one genetic counselor (NPIdb.org; MedicosPR.com; Personal Communication, 2022). Although not everyone may need GS, the number of genetic professionals currently available is not adequate to best support or provide for families and patients, especially with such a large population. In Puerto Rico, there is approximately 1 genetic provider per 640,000 population. In a workforce study from the professional organizations of genetic counselors, it has been estimated that the number of GS providers needed in a population is between 1 per 75,000 and 1 per 100,000 (Hoskovec et al., 2017). In comparison, the entire U.S. has a population of over 328 million people and 4,700 genetic counselors and 1,240 geneticists (U.S. Census Bureau, 2019; Office, 2020). Although there are still a limited number of genetic professionals, the genetic professionals per person ratio in the U.S. is significantly more proportional, with the U.S. Government Accountability Office (GAO) saying states averaged 7 genetic counselors per

500,000 people in 2019 and 2 medical geneticists per 500,000 people in 2020 (Office, 2020), or about 1 GS provider per 55,556 persons, although this ratio varies greatly across the country. The insufficient genetic professionals in PR exacerbates the disparity in accessing GS.

Disparities in terms of access to GSs are not the only ones that exist in PR. Health disparities are more notable in populations with low income, racially/ethnically minoritized populations, and more Spanish-speaking populations, all of which are found in PR. As of 2019, the median household income in PR is \$20,539, which is three times lower than the median household income of \$62,843 in the U.S. (U.S. Census Bureau, 2019). The income levels in PR impact an individual or family's ability to pay for anything except necessities. Families will prioritize food and shelter, so any costs surrounding health will negatively impact a family. In addition, PR is mainly a Spanish-speaking country with 98.7% identifying as Hispanic or Latino and where 93.4% of the population speaks Spanish, and the rate of speaking English "very well" or only English is 20.4% (U.S. Census Bureau Data, 2019). Also, the racial composition of the PR population is 65.9% white, 11.7% black, 0.4% other race and 5.3% as two or more races (U.S. Census Bureau, 2019), but this may underestimate diversity because PR's cultural standards cause a large proportion of individuals to report their race as white, despite skin tone, because of the underlying disdain for dark or black skin (PR Title V, 2019). Disparities in health care are often affected by systemic inequalities that exist surrounding race and ethnicity. Despite a generalized denial of racial prejudice and discrimination on the island, racism and racist acts still manifest through sly comments, racial jokes, and underrepresentation in media outlets (PR Title V, 2019). These implicit inequalities can still implicitly reinforce disparities.

The New York Mid-Atlantic Caribbean (NYMAC) regional genetics network (RGN) has recently started working with PR to reduce these disparities and increase genetic access to patients

and families. NYMAC's work with PR first began as of June 2020; before that, their sister RGN, Southeast Regional Network (SERN) worked with PR for years, though specific details on that collaboration are limited (National Coordinating Center for the Regional Genetics Network, 2021). NYMAC, as an organization, aims to collaborate with individuals with genetic conditions, their families, advocates, and healthcare professionals to ensure that individuals with heritable conditions and their families can access quality care and genetic expertise. NYMAC hopes to accomplish that goal by supporting a regional infrastructure for GSs, providing education and resources to families and providers, and facilitating the use of telehealth in genetics (NYMAC, 2021). The specific PR team within NYMAC is composed of 10-15 key stakeholders to assess the specific needs and barriers of PR and navigate how to implement NYMAC-funded projects to improve access to GSs.

Through their work, NYMAC has established the base level of GSs in PR to include four geneticists, one genetic counselor, no telegenetics and no graduate programs specific to genetic providers (Personal Communication NYMAC, 2021). NYMAC aims to improve access by increasing the number of available genetics providers over time, potentially using telehealth avenues, begin to advocate for recognition of the profession of Genetic Counseling in PR, and build at least one collaborative program where students from PR can train to become genetic providers or genetic assistants (Personal Communication NYMAC, 2021).

This project attempts to further the scope of this limited understanding of genetic services in PR by conducting meetings with PR stakeholders. These meetings were conducted by the NYMAC Regional Genetics Network. These stakeholders have been identified based on their connections to GSs they work with, starting with private and public geneticists, followed by public health providers, personnel from the PR Department of Health (DoH), educators in the universities

in PR, organizations that support families with special health care needs, and lastly, families and parent support groups. Individual conversations with each stakeholder helped in understanding their own perspectives and their reach in the PR community. Though each meeting met with a different stakeholder, the purpose behind these meetings was to better understand an individual's knowledge of GS, the process of receiving GS and the impact that process has had on an individual with or at risk for genetic conditions. This needs assessment utilizes these meetings and conversations with important PR stakeholders to understand and assess the GSs in PR by conducting a grounding qualitative analysis.

1.1 Specific Aims

Aim 1: To identify the current status of genetic needs in Puerto Rico according to Puerto Rican stakeholders.

Aim 2: To summarize and analyze four meeting transcripts between NYMAC and stakeholders in Puerto Rico using a grounded thematic qualitative analysis.

Aim 3: To propose potential methods to address genetic services needs in Puerto Rico based on the results of the qualitative data set.

2.0 Literature Review

This literature review will begin by defining and looking into genetic needs, services and professionals and providing an example of GSs offered in PR. Next, we will focus more on PR, especially in its relationship with the United States as a territory, the influence of the PR culture on its people's perspectives on health care approach and health care access. Then, describing previous research involving engaging PR patients in recruitment and education and challenges faced. The last section will focus on the public health significance of this research focusing on disparities and barriers in underserved communities, especially Spanish-speaking communities, and in implementing medical genetics, limited public health efforts and gaps of knowledge in the research.

2.1 Genetic Services and Needs in Puerto Rico

2.1.1 Defining Genetic Services and Needs

Genetics refers to the information contained in our genes and DNA composition that is passed down through generations within families. Despite not having a universal definition, for this paper GS focuses on methods, technology, and ways to better understand the genetics and hereditary risks for genetic conditions. Traditionally, these services include medical services like genetic testing, diagnosis of genetic conditions, genetic counseling, and treatments for individuals who are at risk or have genetic conditions (Innovations in Service Delivery in the Age of

Genomics: Workshop Summary, 2009), although some also conceptualize GS more broadly to include genetics education and family support organizations. In addition, genetic needs are defined as complications or challenges faced that can benefit from seeking certain GSs. This definition allows individuals with genetic conditions and providers to identify what GSs can aid a patient to improve their current quality of life or recognize where certain areas are weakened because of a lack of those GSs.

2.1.2 Defining Genetic Professionals

Genetic professionals consist of medical providers who offer GS for patients with or at risk of having a genetic condition. These professionals are categorized by those who have received formal training, certificate and/or licensure in genetics, which includes medical geneticists, genetic counselors, and genetic nurses, and other health care providers who offer genetics in their care (Innovations in Service Delivery in the Age of Genomics: Workshop Summary, 2009). Genetic professionals without any formal training create a challenge in determining exactly how many and what type of health care providers offer GS as part of their practice and how effective the quality of care they offer is (Innovations in Service Delivery in the Age of Genomics: Workshop Summary, 2009). This challenge essentially acts as a data gap as it provides no specific way to count or characterize all the medical providers who may be offering GSs (Innovations in Service Delivery in the Age of Genomics: Workshop Summary, 2009). Due to that limitation, genetic professionals in this paper will mainly concentrate on healthcare providers with formal education in genetics. Now that these main terms have been defined, it is important to discuss what specific GSs are offered in PR.

2.1.3 PR Newborn Screening Program

Newborn Screening (NBS) Programs recognize genetic conditions that can potentially impact a child health or survival in the long-term (Newborn Screening Portal, 2021). This public health program conducts certain tests for specific genetic conditions that may not be apparent at birth. Newborns undergo three different kinds of examinations that occurs 24 – 48 hours after birth. The first is a heel prick test, where a newborn’s heel is pricked to collect a few drops of blood and sent to a lab for further analysis (Kaneshiro and Zieve, 2021). The second is a hearing test, where a medical provider puts either a small earpiece in the newborn’s ear or electrodes on the newborn’s head while asleep (Kaneshiro and Zieve, 2021). The third test is a pulse oximetry test, where a medical provider places a sensor on the newborn’s skin attaching it to an oximeter, which measures the newborn’s oxygen levels to test for critical congenital heart defects (CCHDs) (Kaneshiro and Zieve, 2021). While there is a specific and established recommended list of genetic conditions for states to screen, ultimately the state or territory decides which conditions to include in their NBS program (Recommended Uniform Screening Panel, 2020).

The NBS in PR was funded in 1987 and has expanded to 50 genetic conditions as of 2020 (States Puerto Rico, 2020). This program has since served over 99% of newborns to provide appropriate laboratory screening and clinical follow-up as needed (States Puerto Rico, 2020). In fact, all newborns in PR are mandated by the PR government law as of 1987 and renewed again at 2015 to be tested for NBS and does not offer an opt-out policy (States Puerto Rico, 2020). In addition, the fee charged to each patient for submitting a sample to the NBS panel for analysis is about \$105 (States Puerto Rico, 2020; “Para Hospitales...,” n.d). NBS testing and sampling are sent to the Pediatric University Hospital at the San Juan Medical Center. This indicates that any newborn across the entire island has their sample sent to one hospital that conducts analysis for

genetic conditions. That center holds responsibility to contact patients, pediatricians, or corresponding specialists for follow-up of an abnormal result (“Para Hospitales...,” de la Salud, n.d). The NBS in PR has faced complications recently that impact its ability to provide the best quality and timely results for patients.

NBS services were complicated by Hurricane Maria, a Category 5 hurricane, which hit PR in 2017 leaving devastation across its path, destroying houses, schools, and roads, and leaving the people without water, electricity, or homes (HRSA Maternal & Child Health, 2019; Welton et al., 2020). According to a webinar by Dr. Rivera-Sánchez, the associate director of the PR NBS program, the program and hospital faced from minor to more critical impacts. Due to the cement-based infrastructure of the NBS building and facilities, they suffered minor damages. They had unstable power and internet, less available personnel, one occasionally functioning telephone line and some water entered the building (Rivera-Sánchez, 2018). In addition, each step in NBS experienced its own challenges. The sample collection and delivery to NBS laboratory were negatively affected. During the hurricane, most of the 69 hospitals in the island had no electricity or fuel for generators, communication within hospitals was lost, and many of the roads experienced obstruction (Rivera-Sánchez, 2018). The NBS program reached out to hospitals to inform them of their operational status and accepted all samples as they arrived. Also, hospitals which were no longer sending samples were reported to the CDC Hurricane Response Division to assess their status (Rivera-Sánchez, 2018). The hurricane also impaired sample analysis and reporting of results. Reduced personnel, manual reporting due to an inability to use computers for reporting, and an increase in the number of repetitions for tests due to aged sampling or inadequate storage are a few of the reasons for impaired sample analysis and reporting (Rivera-Sánchez, 2018). Follow-up to patients and providers proved to be the most difficult. There was no communication

at all throughout the island, a significant increase in the number of repeated tests, extensive delays and 24 presumptively positive screen cases identified in the first weeks following Maria (Rivera-Sánchez, 2018). NBS professionals had to find various strategies to contact families, including through social media, police, volunteers, text messages sent day and night, or personal visitation (Rivera-Sánchez, 2018). While it is fortunate that PR NBS received support from various organizations, such as Perkin Elmer, New York (NY) NBS, Association of Public Health Laboratories (APHL), CDC, and NewSTEPs, PR recognized a need for revising emergency plans and preparing a backup plan to better provide services for patients (Rivera-Sánchez, 2018). Although Hurricane Maria negatively affected an important GS in PR, there are other challenges to improving GS.

2.1.4 Challenges to Improve Genetic Services

Challenges and limitations exist when integrating and improving GS in PR. A study led in 2009 about expanding newborn screening in PR and the U.S. Virgin Islands (USVI) conducted in-person or phone interviews with NBS program directors to gain a better understanding of the knowledge, needs and barriers needed to successfully implement an expanded NBS (Morales et al., 2009). They determined that about 60% of program directors in PR and USVI did not believe there are enough trained genetic professionals (Morales et al., 2009). They believe one potential reason for that is because of the cost of living, as it is not enough to attract clinicians to complete their practice on each respective island(s) (Morales et al., 2009). Other reasons include culture, language, and geography as there are greater differences between the mainland USA and PR. In respect to medical intervention, about 15% of participants indicated they did not have both access to metabolic pathways information to be used for patients with an inborn error of metabolism and

specific formula and dietary intervention information for conditions (Morales et al., 2009). Furthermore, language acts as another barrier to implementing NBS in PR. The study's data shows that about 65% of participants agreed that language differences are an issue for finding and collecting informational materials about NBS (64%), for reading the sample collection card (59%) and when establishing communication with other NBS program (59%) (Morales et al., 2009). The latter is especially significant as communication with other NBS programs can aid PR in receiving resources as well as informational and educative materials from another state's NBS program, especially if they have similar conditions on their own panel. Lastly, 51% of participants agreed that insurance and financial status can hinder the program's success. (Morales et al., 2009).

Other challenges exist in improving GS that are not necessarily specific to PR. There is a lack of knowledge of GSs overall. A focus group study by Beene-Harris et al. (2006), stated that almost all their participants mention a lack of knowledge for GS and other resources. These participants faced difficulty in obtaining information on their genetic condition and where to go for and following a diagnosis, although they were unable to determine differences between conditions (Delikurt et al., 2015; Beene-Harris et al, 2006). Also, non-genetic healthcare providers lack knowledge of genetics, risk, and conditions. Healthcare providers have rated their genetic knowledge level as limited, and a lack of knowledge in genetic conditions was associated with difficulty for providers to provide appropriate referral, treatment, and management for a patient (Aalfs et al., 2003; Beene-Harris et al, 2006; Delikurt et al., 2015). Healthcare providers also recognize a need for a genetic workforce to meet the need for an advancing genetic technology and field (Beene-Harris et al, 2006; Delikurt et al., 2015). While these are more generalized challenges with improvement in GS, to gain a better understanding of limitations in PR, it is

important to acknowledge the unique influences surrounding Puerto Rico, including influence from the continental U.S., cultural beliefs and views, and the healthcare system.

2.2 Puerto Rican Culture

2.2.1 U.S. Territory/Commonwealth

PR currently serves as a U.S. territory or commonwealth. Those terms are often used interchangeably; however, they do not necessarily have the same definition. PR became a U.S. territory after the Spanish War and the Treaty of Paris in 1898. During this time, a territory could be “incorporated as an integral part of the United States by conquest or treat, without being incorporated as an integral part of the United States but remaining in the subordinate status of a possession or dependency” (Magruder, 1953). It was not until July 3, 1952, following the final ratification by the constitutional convention and Congress approval, did the Governor of Puerto Rico proclaimed the establishment of the Commonwealth of PR under the constitution (Northrop, 1952). The use of the commonwealth occurred after Congress approved PR’s constitution; however, the allowance of local power did not reduce the federal control or involvement over PR because of the change from the territory status (The Meaning of “Commonwealth”, 2021). In fact, the term “commonwealth” was defined as ““a state which is free of superior authority in the management of its own local affairs, but which is linked to the [USA] and hence is a part of its political system in a manner compatible with its federal structure’ and ‘which does not have an independence or separate existence.’” (Northrop, 1952).

Due to PR's commonwealth status, several liberties and limitations exist. The Puerto Rican government is established as a self-government in most local matters as a tri-partite form, with an elected governor, two-house legislature, and a judiciary branch (Northrop, 1952). Puerto Ricans are exempt from the federal income tax but must pay into Social Security ("Bria 17 4 c...", n.d.). Puerto Ricans are American citizens who can reside in the U.S. and serve in the military ("Bria 17 4 c...", n.d.). Although Puerto Ricans are citizens, they are not able to vote in presidential and congressional elections. The only formal representation in the Washington D.C. is a non-voting, Resident Commissioner to the U.S. House of Representatives, currently held by Jenniffer González. The PR government is largely financed by federal grants of an annual budget of more than \$10 billion ("Bria 17 4 c...", n.d.). The U.S. has authoritative controls over: interstate trade, foreign relations and commerce, customs administration, control of air, land and sea, immigration/emigration, nationality and citizenship, currency, maritime laws, military service, military bases, declarations of war, constitutionality of laws, jurisdictions and legal procedures, treaties, radio and television, highways, and postal system (Government, n.d.). Despite major controls by the U.S., there are still recognitions of Puerto Rico's own identity, such as by the International Olympic Committee, who views PR's National Olympic Committee as a separate entity from that of the U.S. (Bermudez, 2021).

While many of PR's citizens also see PR as a separate entity, even more have voted for PR to become a state and this conflict has led to resistance towards the change of status from commonwealth to state. On November 3, 2020, the PR Statehood Referendum was on the Puerto Rican ballot as a legislatively referred statute, which asked voters: "Should [PR] be immediately admitted into the Union as a state" (Puerto Rico Statehood Referendum, 2020). Results indicate that approximately 52.5% of voters answered "Yes" to statehood, and 47.5% voted "No" on

statehood (Puerto Rico Statehood Referendum, 2020). Those who support the statehood want to reduce discrimination and colonial mindset and increase equality in financial and political support (Puerto Rico Statehood Referendum, 2020). Those who oppose statehood want to maintain their independence from the U.S., preserve the Puerto Rican nationality and viewed the ballot as a misuse of public funds and political corruption (Puerto Rico Statehood Referendum, 2020). This controversy on statehood has led to the continued status of a commonwealth. Not only are there political complications, but Puerto Rico is also very proud of its culture, and culture has a major influence and implication on health care.

2.2.2 Culture and its Influence on Healthcare Approach

Cultural identity remains important for Puerto Ricans and influences their attitudes towards healthcare. Culture is strongly rooted in elements of language, important familial ties, and religion, most of which can be tied its colonial background. Although English and Spanish are both the official languages of PR, Spanish is the primary one, where 98.7% identifying as Hispanic or Latino (U.S Census Bureau, 2019). The Spanish language remains a reminder of the Spanish colonization that occurred from as early as the 15th century. However, the Spanish language has been infused with terms and phrases from the original Taíno natives that resided in PR prior to Spain's conquest. Once the U.S. claimed PR, English was introduced and created friction in education. U.S. authorities insisted on making English the primary language in schools and instruction ("Beyond Language," n.d.). However, a strong resistance to the policy allowed the change to Spanish as the basic academic language, and English the secondary language to be studied by all citizens ("Beyond Language," n.d.). Currently, there is much variation in the level of English proficiency, from fluency to incorporation of English words in Spanish (often referred

to as “Spanglish”) to solely Spanish speakers (“Beyond Language,” n.d.). In fact, 93.4% of the population speaks Spanish, and the rate of speaking English “very well” or only English is 20.4% (U.S. Census Bureau Data, 2019).

Familial ties contribute an important portion of Puerto Rican culture. Given the American citizenship granted to Puerto Ricans, they can freely travel between PR and the continental U.S. without restrictions. This liberty allows Puerto Ricans to preserve strong family connections (“familismo”) and social network, native language, and proud nationality identity (Torres et al., 2008). In doing so, it allows the cultural representation of community to surpass national and geographical lines (Torres et al., 2008). In many cases, there may be multiple generations living within the same household or same neighborhoods. Older members are respected and sought for their wisdom and support. Grandparents are also important as they help with childcare responsibilities while parents are working or in times of crises.

In addition, within family structures, there are expectations and roles, especially divided by gender. In PR and Hispanic culture overall, “machismo” and “marianismo” has raised issues. A man shows characteristics of dominance and control on females, households, and their emotions (Bird, 1982). However, a dichotomy does exist as machismo remains prevalent, but men are also expected to show signs of chivalry, such as holding a door open or relinquishing their seat on public transportation for a woman. On the other hand, women still have an expectation of being responsible for household affairs, child-rearing and taking care of their husbands. These characteristics and beliefs are also passed down through the generations. Although as times change, these views have become more fluid and less rigid, they still remain within society. As gender roles change, Puerto Ricans will continue to restructure their social interactions and communication patterns, while still maintaining the importance of family. Understanding familial

relationships aids in understanding the culture and how to present information appropriate for Puerto Ricans. In addition, since living arrangements and family bonds are so close, this can be beneficial and act as a cultural diffusion of resources for health promotion programs (Torres, 2008).

Lastly, religion impacts Puerto Rican culture. Most Puerto Ricans are Christian with about 56% identifying as Catholic and 33% are Protestants (Krogstad et al., 2017). Due to the indigenous presence that remains, spiritism still serves as an important community support system. Spiritism is based on the belief of reincarnation and in the power of individuals (Hohmann et al., 1990). The belief is that fluids are spiritual emanations that surround that body, that can result in mental and physical illness (Hohmann et al., 1990). This practice encourages spiritual and herbal work instead of medical aid to alleviate physical and mental illnesses. Puerto Ricans use spiritism as a folk psychotherapy, an outlet for the anxiety due to economic and interpersonal problems and as community support for chronic schizophrenia (Hohmann et al., 1990). Though not everyone adheres to this belief, it is important to acknowledge that many Puerto Ricans still have a connection with the supernatural and nature. Many Puerto Ricans prefer taking natural and herbal medicinal before relying on pharmaceutical prescriptions. When treating Puerto Ricans, a holistic treatment has the best approach. Providing a spiritualist counselor, or religious counselor, treats emotionally related problems, while also treating physical illnesses.

2.2.3 Health Care Access

Understanding culture and its influences highlights struggles in access to health care. A few notable access barriers include insurance and language. In PR, about 93.2% are insured, which leaves about 6.8% of individuals uninsured (U.S. Census Bureau, 2020). Of those insured, 61.3%

have public health insurance coverage, whereas 38.9% have private coverage (U.S. Census Bureau, 2020). Of those with public health insurance coverage, 24.7% are covered by Medicare alone, 46.8% are covered by Medicaid, and 1.5% are covered by VA health care coverage (U.S. Census Bureau, 2020). Despite the high insurance coverage, insurance does not cover everything. The Medicaid program in PR differs from the U.S. states. The PR DoH is the single state agency and has a cooperative agreement with the PR Health Insurance Administration. Each territory is provided base funding, for about \$5.4 billion in Medicaid funding to PR (Medicaid.gov, 2019). However, unlike the 50 states, the federal matching assistance percentage (FMAP) rate is applied until the Medicaid and Affordable Care Act (ACA) funds are used, rather than matching (Medicaid.gov, 2019). In other words, PR receives a block grant from Medicaid and ACA, and once those funds are used, PR must use its own funds to cover the cost (MACPAC, 2021). Since the funds covered are not enough, this leads to families having high co-pays as insurance is not able to cover care in a way that sufficiently supports the Puerto Rican population.

Insurance and costs are still a problem for other reasons. Given the large discrepancy in English proficiency, seeking services outside of Puerto Rico can be a major challenge. If insurance is not in a patient or family's primary language, miscommunication and confusion are prominent. This can cause insurance to not cover a cost, due to patients' not understanding the information that was covered. In addition, since there is one genetic counselor and four geneticists on the island, many individuals may need to travel to the U.S. to seek GSs. Patients and families need to cover the expenses of traveling. Insurance may vary by state and thus could work differently or not cover the same services as those offered in PR. On top of that, if a patient does not speak English well, language can create issues on what is understood, what results say, and what the next steps for service are.

According to the Title V State Application, for some families with children diagnosed with genetic conditions, accessing a genetic laboratory may be difficult due to the high costs. Some laboratory panels may have too high of an insurance deductible for a family to pay, which can cause laboratory tests to be delayed while the family gets the money. Furthermore, most of these specialized laboratory tests are done outside PR, which increases the time for results to arrive. In the 2015 PR Child with Special Health Care Needs (CSHCN) Survey, almost 20% of families with a child in need of a genetic or metabolic laboratory could not access the tests needed. Of that value, about 66% could not access due to high costs and almost 30% because the insurance plan did not cover the cost (*HRSA Maternal...*, 2019). These barriers exist in access to health care, and specifically for GS.

2.3 Previous Research in Engaging Puerto Rican Patients

Research on GS in PR is quite limited. Much content available in the literature review does not focus on GS, especially in PR. Research focusing on genetics conditions or genetic ancestry exist, but not on genetic services and testing. Despite the limited research, previous research has been done on effectively recruiting Puerto Rican participants and the education of genetics that is offered.

2.3.1 Recruitment Strategies

When trying to engage with Puerto Rican patients, it is valuable to keep in mind appropriate strategies to encourage recruitment and engagement. One study elaborated on important

considerations for recruiting women in PR to cancer genetic studies. Their goal was to enhance cancer research training and outreach programs by being culturally appropriate to the audience (August et al., 2011). Over the course of the study, multiple sociocultural factors were noted, and four themes were noted: be respectful, establish personal relationships, build networks, and present yourself professionally. Verbal communications were accompanied by nonverbal cues, including emotional expression, physical proximity, light touching, and expressions of affection. Partnerships with key communities' advocates were an essential component in the recruitment strategies as they can deliver information about the study in a way that is acceptable, appropriate, and understood to the community. In addition, participants were more willing to call about participating in a study when suggested by a trusted member of the community or a friend than by posters and flyers. Respect and professionalism are closely tied when interacting with the community, specifically by formal language and attire. Using the formal language of "usted" rather than "tu" in the Spanish language, was important in conversations with a person of authority or high social status. Regardless of SES, how one presents themselves with their attire is an important custom for Puerto Ricans. Lastly, it was found that introducing oneself as a researcher rather than student was prioritized as it aids the participants in establishing trust, confidence, and credibility to the study. Given the study's focus on engaging with Puerto Ricans, these considerations are especially useful in maintaining an appropriate and respectful engagement with stakeholders. This level of respect encourages and helps foster relationships to be more long-standing (August et al., 2011).

2.3.2 Education

When aiming to engage with Puerto Rican patients, it is important to keep in mind the education in genetics that patients may have or what strategies should be taken to encourage genetic education. In PR, medical genetics courses are offered at all levels of higher training, including undergraduate and medical schools (Cruz-Correa et al., 2016). Clinical genetic services are primarily offered in the capital, San Juan, through the Pediatric University Hospital and concentrated on diagnosing congenital defects, inborn errors of metabolism and craniofacial abnormalities (Cruz-Correa et al., 2016). Despite the educational training offered, various genetic conditions, especially rare conditions, often are not completely known or understood in the depth needed by trained professionals.

Despite formal education, many Puerto Rican citizens are not as well-versed or fluent in genetic literacy. When a group of women were asked about genetic testing on cancer genetics, the most frequently cited concern was whether the test would be painful, which demonstrates the minimal knowledge about genetic testing these women had (Vadaparampil et al., 2011). Data from National Health Interview Survey between 2000 and 2005 claim that less than 33% of Puerto Rican women have heard of genetic testing for inherited cancer susceptibility, especially when compared to almost 50% of all Whites (Vadaparampil et al., 2011). Thus, knowledge and literacy of genetics to the general population is low and providing culturally appropriate education can improve understanding and importance to the definition of genetics and benefits of genetic testing.

2.4 Public Health Significance

2.4.1 Disparities in Puerto Rico

According to the CDC, health disparities is defined as a preventable difference in the burden of disease, injury, violence, or other opportunities to achieve optimal health that are experienced by socially underserved populations (CDC, 2020). Health disparities result from a multitude of factors, from individual, such as race or ethnicity, gender, education, and disability, to interpersonal, such as SES, poverty, and environment. Health disparities are directly linked to the historic and present unequal distribution of resources (CDC, 2020). Although health disparities are apparent and exist in several Spanish-speaking countries; this section will focus on specifically on genetic disparities in PR.

Some disparities that exist for access to GS in PR include lack of genetic professionals, language, financial support, and education, some of which have already been explained in previous sections. As discussed, there are few genetic professionals on the island; five, in fact. There is one genetic counselor and four geneticists. Although not everyone in the 3.2 million population will need a genetic professional, there are not enough genetic professionals currently available to adequately provide care to patients and families. As stated above, a workforce study from the professional organization of genetic counselors estimates that the number of GS providers needed in a population is approximately 1 GS per 75,000 – 100,000 people (Hoskovec et al., 2017), but PR has approximately 1 genetic provider per 640,000 people and that's combining genetic counselors and geneticists. If genetic providers were to stand alone; there would be 1 geneticist per 800,000 people and 1 genetic counselor per 3.2 million people. That value is significantly higher than what studies suggest being the optimal level of provider-to-patient load. When

compared to the GAO, the U.S. states average 7 genetic counselors per 500,000 people in 2019 and 2 medical geneticists per 500,000 people in 2020 (Office, 2020). Although still a limited number of genetic professionals in the U.S., the disparity is significantly more exacerbated in PR.

Language is another major disparity in PR. Despite English and Spanish being the official languages, Spanish is the primary language. Thus, if GS or information is received in English, patients will have a harder time understanding their genetic situation or the terms genetic providers are saying. In addition, if insurance content and material is in English, getting insurance access can lead to miscommunication and confusion. In addition, a study looking at impact of NBS, a type of genetic test, shows that about 60-65% of participants agreed that language differences were an issue for finding and collecting informational material about NBS, reading the sample collection card and in communication with other NBS programs (Morales et al., 2009). As mentioned above, lack in communication with other NBS programs is notable since it helps PR receive resources and educational materials from another NBS program, especially if they have similar genetic conditions on their panel.

Financial support has also been a notable disparity in PR. Though a majority of Puerto Ricans are insured, public insurance through Medicaid and ACA may still lead to high co-pays or insurance deductibles since federal funds do not cover all costs. In addition, since not all genetic services can be obtained on the island, many patients and families may need to travel to the U.S. On top of traveling expenses, insurance can vary by state/territory, so costs and services covered under the insurance may vary. Lastly, certain genetic tests, such as those for a child in need of a genetic or metabolic laboratory, may not be covered by insurance plans (*HRSA Maternal...*, 2019). These financial barriers exist in access to health care, and specifically for GS.

Education about genetics and its services are also limited for the public. Many Puerto Rican citizens do not have a high genetic literacy. As mentioned above, Puerto Rican women have concerns about the pain for genetic testing, which highlights unfamiliarity on genetic processes, how they are conducted, and how results are obtained (Vadaparampil et al., 2011). Women are in addition unfamiliar with inheritance, susceptibility, and probability of having a specific genetic condition and the specific processes of genetic testing. Thus, knowledge and literacy of genetics to the general population is relatively low, and a way to address this disparity is by providing culturally appropriate education to improve understanding of the definition of genetics and benefits of genetic testing.

Lack of genetic professionals, language, financial access, and education have all been demonstrated to be disparities within the Puerto Rican population. As public health professionals, the goal is to protect and improve the health of communities. One important method in doing that is by detecting health conditions as early as possible and responding appropriately to encourage the best quality of life. However, these disparities create further challenges for public seeking health services, specifically in the genetics field.

2.4.2 Gaps in knowledge

Much of the literature review discussed attempts to primarily focus on PR, however, that is not entirely possible. Research, studies and even websites do not often discuss GSs and what is offered in PR. The content that is available may be too difficult to find and comprehend. Thus, this presents several gaps in the knowledge. Research does exist in genetic ancestry or specific studies in relation to certain genetic conditions, including Alzheimer's, Albinism, and cancer as a notable few. Yet, the focus on genetic services and testing is limited. The most recent research found on

GSs in PR is of expanding NBS in PR and USVI from 2009. The study focused on the implementation of the expanded newborn screening panel of 29 disorders as recommended by the American College of Medical Genetics while in its early development stages. As of 2022, the NBS has expanded to 51 genetic conditions (HRSA, 2022). The genetic conditions on the panel have almost doubled in the past decade; however, research has not advanced with those changes.

Although some research exists on genetic services in other Latin American countries, there is a caveat to using those studies as it will not be fully transferable to PR. Most countries have their own health care system and own government, whereas, PR's government and healthcare system works congruently with the U.S. Studies involving the genetic studies in other U.S. territories including USVI, Guam or American Samoa could be beneficial as their government and healthcare system is intertwined with the U.S. Research on Guam and American Samoa would be particularly interesting as they each have a native language, Chamorro and Samoan, respectively, and English as their official language. However, like PR, there is limited research.

This project will be able to address some of the gaps that currently exist in the knowledge. It will provide the most-up-to date information on what the state of GS are in PR. It can further highlight current challenges and barriers that both patients of a genetic condition and providers face. It provides a better understanding as to the impact of lack of support and genetic services has had on families. NYMAC will be utilizing the data gathered from this needs assessment to describe the status of genetic services and find potential solutions to increase access to GS in PR. Ideally, this needs assessment can be used to impact governments, policies, and researchers.

3.0 Methods

3.1 Methods

The data used for this needs assessment consist of meeting transcripts between NYMAC team members and PR stakeholders. NYMAC identified stakeholders based on how they are impacted by GSs in PR, starting with private and public geneticists using previous contacts within the island (Personal Communication – NYMAC, 2021). Next, they continued with public health providers and personnel from the PR DoH by utilizing contacts from previous projects and work for the Zika Project (Personal Communication – NYMAC, 2021). Due to the close relationship between the PR DoH and the University of PR, NYMAC was easily able to connect with the University of PR (Personal Communication – NYMAC, 2021). Following seeking professional contacts, they reached out to parent support groups, such as Support for Parents of Children with Disability (APNI, by their acronym in Spanish - Apoyo a Padres de Niños Con Impedimentos) (Personal Communication – NYMAC, 2021). These support groups were able to connect NYMAC to people who serve expectant mothers and parents of young children in PR (Personal Communication – NYMAC, 2021). To assess each stakeholder, conversations were held with each contact to gain a better understanding of their perspective and the reach they have in the community (Personal Communication – NYMAC, 2021). In these conversations, NYMAC as an organization was explained to each contact and their interest levels in being a part of the PR team was evaluated (Personal Communication – NYMAC, 2021). Those who expressed interest agreed to being a part of the PR team and commitment to NYMAC’s goals (Personal Communication – NYMAC, 2021).

To gather the stakeholders for each meeting, multiple methods of contact were utilized. Due to previous interactions, each stakeholder was contacted by their preferred method. Methods for contact included texts, phone calls, WhatsApp messages and emails. The meetings held prioritized the availability of those most closely involved in the genetics field and families, and Friday afternoons best worked with people. The meetings included translators for those who English was not their primary language. The meetings were held via the Zoom application and recorded. The recordings of these meetings are the specific data set utilized for the purpose of this project.

3.2 NYMAC Description of Data and Analysis

The meetings occurred from October 2020 until September 2021. Of the 18 meetings recorded, four were identified for the purpose of this paper as those conversations focused specifically on NYMAC and PR stakeholders. The other meetings were conversations amongst the NYMAC team. The dates of the meetings occurred on October 23, 2020, March 12, 2021, April 16, 2021, and May 14, 2021. The meetings were transcribed using an automatic transcription application, Temi©. The meeting transcriptions were individually reviewed to ensure the transcription aligns with what was spoken. In addition, due to audio issues, manual translations from Spanish to English were completed to have the most thorough and complete transcription as possible.

The meeting transcriptions were analyzed using grounded theory to identify emerging themes. Grounded theory is a form of inductive qualitative analysis utilizing line-by-line analysis to discover relationships based, or “grounded” in the data (rather than utilizing a pre-established

theory) (Patton, 1980; Foley and Timonen, 2015; The Practical Guide to Grounded Theory, n.d.). The benefit of utilizing this qualitative approach is that findings are tightly connected to the data and represents real issues specific to PR as they emerge (The Practical Guide to Grounded Theory, n.d.). Transcripts are broken down into line-by-line data bits (short snippets of data) to closely analyze what each stakeholder said. Codes were derived from the data bits, by noticing patterns or similarities in what has been said by the stakeholders. This process was followed for the four meeting recordings. Names of the participants were removed to maintain their privacy; however, participants were identified by their profession.

3.3 Data Demographics

Each meeting focused on specific stakeholders and had specific focus for the discussion. Table 1-4 lists the total participants, and the breakdown of the specific role of the PR stakeholder for each meeting: 10/23/2020, 3/12/2021, 4/16/2021, and 5/14/2021, respectively. It is important to note that participants with the same role in the meetings are the same individual. A total of 14 PR stakeholders have attended the meetings of which three work in the genetics field, three work with the PR DoH, five work specifically with family and patients, and three work with universities and the educational system. Table 1 illustrates the total participants in each recorded meeting and Tables 2 – 5 provide the individual participant role for each individual recorded meeting. Certain professions were combined into three categories: genetics provider, public health professional and provider. Those who have the role of genetic counselors, private geneticists and public health geneticists were assigned a “Genetics Provider” role. Those whose roles are Coordinator of Hereditary Disease Division and MIECHV Program Coordinator were assigned a “Public Health

Professional” role. Lastly, those whose roles were registered nurse, Director of Pediatrics Department, and Pediatrics Specialist and Dean of School of Medicine were assigned a “Provider” role.

Table 1 Total Participants in Each Meeting Recording

Date	Total Participants
October 23, 2020	9
March 12, 2021	15
April 16, 2021	8
May 14, 2021	14

Table 2 October 23, 2020, Participant Role Information

Participant Role	Total Participant(s)
NYMAC Team Member	3
Family Voices Member	2
Translator	1
Public Health Professional	1
Genetics Provider	2
Total	9

Table 3 March 12, 2021, Participant Role Information

Participant Role	Total Participant(s)
NYMAC Team Member	2
Family Voices Member	2
Translator	1
Public Health Professional	2
Genetics Provider	2
Provider	2
Parent	1
Family Representative	1
Independent Consultant	1
University of Pittsburgh Graduate Student	1
Total	15

Table 4 April 16, 2021, Participant Role Information

Participant Role	Total Participant(s)
NYMAC Team Members	2
Family Voices Members	2
Genetics Provider	2
Provider	2
Total	8

Table 5 May 14, 2021, Participant Role Information

Participant Role	Total Participant(s)
NYMAC Team Member	4
Family Voices Member	4
Public Health Professional	2
Early Intervention Program	1
Family Representative	1
Provider	1
University of Pittsburgh Graduate Student	1
Total	14

4.0 Results

Based on the meeting transcripts, at least 18 codes in total were identified from the data directly. These codes depicted themes identified from the analysis of the meetings with PR stakeholders. They were divided in two groups: codes based on conversations with health care professionals (Table 6) and codes based on conversations with patients, families, and professionals whose work focused on patients and families (Table 7). On conversations specific to health care professionals, one major focus on is on genetic professionals in PR, including themes about where they are located, the need for them, and challenges with incorporating more genetic professionals into PR, specifically how insurance plays a role. Other themes centered around differences in processes and GS between the continental US and PR, specific types of patients who are seen by a genetic professional, and ideas or interventions for change and improvements to increase GS. On conversations specific to families and patient needs, there were multiple themes including emotions, both positive and negative, that parents identified with, barriers to accessing of genetics appointment and provider, potential solutions to barriers, and challenges to the solutions. Another theme for patients and families focuses on ways to prepare for genetic appointments and seeking support. In addition, another major theme was surrounding the deaf community and potential solutions that addressed the specific needs of this community including a need for interpreters. Lastly, an interesting trend was the consistent use of the word “important” in the conversations with patients and families.

Table 6 Codes for Conversations with Healthcare Professionals

Codes	Definition
GPInPR	Genetic professionals within PR
GPlocate	Where genetic professionals are located within PR
GPNeed	Mention or discussion of needing genetic professionals
GPNeed-CH	Challenges with having genetic professional in PR
GPNeed-CH:In	Challenges with having genetic professionals in PR that focus on insurance
DiffUS	Comments and Discussions where genetic services or processes are different from the U.S.
PatientSeen	Comments and discussion of types of patients who see a genetic professional
ChangeIdeas	Comments and discussions of ideas or interventions for change and improvements in increasing genetic services

Table 7 Codes for Conversations with Patients/Families/Professionals

Codes	Definitions
NEGEmo	Negatively connotated emotions parents used, mentioned, and felt, including anger, fear, blame, guilt, worry, concern, etc.
POSEmo	Positively connotated emotions parents used, mentioned, and felt, including joy, thankful, pleased, excited, etc.
Barriers	Comments and discussions to having a genetic appointment or seeing a genetic professional in PR
Barries-Deaf	Comments and discussions on barriers, especially associated with patients and communities with hearing impediments.
Solutions	Comments and discussions on potential solutions to address the barriers in access to a genetics appointment or seeing a genetics professional in PR.
Solutions-Deaf	Comments and discussions on potential solutions to address the barriers in access to a genetics appointment or seeing genetic professionals, especially as it relates to deaf patients and communities, including interpreters
Solutions-Block	Comments and discussions on potential challenges or roadblocks to solutions proposed
Prep	Conversations surrounding what patients and families need to be prepared for genetic appointments
SeekSupport	Comments and discussions of patients and families seeking or getting through organizations and advocacy groups
IMPORTANT!	Use of important variations of the word, including importance and critical

4.1 Themes in Discussion Between Health Care Professionals

As a described above, several themes emerged from conversations between healthcare professionals during the meetings. Providers recognize the lack of genetic professionals in PR, but challenges arise in addressing that challenge. One participant, a genetics provider, shared:

As a matter of fact, today, I received about six or seven different patients with different conditions that I wish I had a genetic counselor with me because it takes so much time.... The work that genetic counselors do is so good.

The genetics provider notes how busy they are by having so many patients, each with a different genetic condition they need to attend to. Because of the variety of patients, they are meeting with, they each take their own time to be as thorough as needed, but they are simply overworked. A genetic counselor would be helpful to patients by providing counsel and options as needed, but even more helpful to the geneticist, who must take over some of the genetic counseling responsibilities given the limited number of providers available on the island. This participant also explains further challenges:

If we have a genetic counselor, we need space. And the only place where I have space for a genetic counselor is in Mayaguez. But then most of my patients in Mayaguez are basically, um, uh, autistic patients.

Although, participants recognize that an increasing workload would benefit from having more genetic professionals, specifically genetic counselors, with no place for them to have an office or practice, increasing genetic professionals would prove to be only so helpful. In addition, current genetic professionals are primarily seeing patients for autism; however, there are countless

other genetic conditions that exist that are not being seen. Extending genetic professionals not only can address the workload but can also help in seeing patients of various genetic conditions.

Another important challenge for genetic professionals includes insurance coverage. One participant stated that:

Genetic counseling is not recognized by insurance companies in Puerto Rico, uh, or paid, or as a profession in Puerto Rico.

This highlights the issue that even if genetic professionals, specifically genetic counselors, were to increase in the island, they would not receive insurance support. This also proves to be a challenge for patients as they may not be able to cover the costs of GS for their condition. One participant explains:

Public health insurance ... some of them can cover the basic, but not all of them. If the patient cannot afford a visit, usually they have to come to [public health geneticists] for the visit, or if they can convince their, their health insurance to cover the visit or if the [private geneticists] have any kind of, uh, agreement with the insurances that they will depend on what they decide.

Public health insurance only covers so much, which may make things difficult for a patient trying to access GS or go to a visit. Although the genetic providers attempt to find solutions to make expenses easier for them, there is still only so much it can cover, especially as genetic counselors are not not recognized by insurance companies as providers in PR. In addition, if patients need to travel outside of PR to receive GS, would the insurance be able to cover that visit or is the insurance even applicable to other states. It poses as an important question and barrier if insurance does not work

Discussions in the meetings investigate ideas that can provide some sort of solution for the challenges faced. Two potential solutions were mainly discussed: collaboration with university partners and telemedicine. One genetics provider affiliated with a US University explained their visit to PR:

We took our students to Puerto Rico last year and established a relationship there that we're hoping to continue in terms of being able to, um, promote genetic counseling services on the island, but to become involved... ..longer term in educational collaboration, either with students from Puerto Rico that wants to be trained as genetic counselors.

This collaboration would foster relationships between genetic providers in PR and those in the continental U.S., to increase GSs. Students in the program would be able to learn about the issues PR faces and be encouraged to continue their practice either directly in PR or be willing to take in patients from PR.

In addition to collaboration, telemedicine was voiced to provide a solution for patients and providers alike. Telemedicine did exist initially in PR; however, it is no longer in practice. One participant elaborates:

We used to have that service through the Jennifer Lopez foundation some years ago and helped a lot of patients to be seen faster and to reach a diagnosis a lot easier.... They will not have to be waiting for six or nine months to be seen by [public health geneticist]. And if they are really positive for a genetic disorder, then I can give the follow-up in an easier way.

Telemedicine has been seen as truly beneficial in the goal of seeing patients more efficiently; however, barriers still exist with that, as another participant describes:

Telemedicine is yes, definitely a potential solution. Um, I think the biggest barrier is, you know, getting that funded. Certainly, it can start with grant funding, but if genetic counselors still aren't recognized by the, um, insurance payers and other people, then it's hard too hard to keep something like that going.

Telemedicine has shown a history of being a major support for providers, but issues, like funding, especially given the constraints established with genetic counseling not recognized by insurance, make telemedicine only so helpful. Grant funding can initially begin the telemedicine program, but without a way to sustain it long-term, it can just as easily fall apart again. This quote specifically highlights how one complication and challenge can further exacerbate the barriers that already exist within PR.

4.2 Themes Between Patients/Families

Several notable themes were prominent in conversations with patients, families and those who work with them. One important theme focused on the emotions parents verbally expressed when having a child with genetic conditions. Some key negative emotions described include fear, loss, and guilt. Parents expressed the fear of losing their child because of their genetic condition. One parent had their child's birthday fall on the day of the meeting, and described it as:

Today, he is 16 years old.... Can't believe it Because sometimes there are moments, we worry because he may not be here with us, but it is special that he is here.

They truly believe that every moment with their child is a gift, because at any moment, the child may no longer be here. The fear of losing your children is a common fear for many parents but given the extra care and worries a parent with a CSHCN has, the fear is increased.

Another important emotion expressed by a parent is confusion. One parent explains:

We have all the professionals, but we don't know how to ask everything. And they are the ones that know. And the family sometimes who do not know or who don't feel they don't know how to ask for the support...

This quote identifies ways that patients and parents feel lost and confused. Parents and patients do not know how to ask their medical providers their concerns or questions. They believe that because providers are trained, they have the knowledge and information they need, but are not sure the best way to ask them or seek support. When they do ask providers, they may explain it in technical terms the patients and families do not understand. Families can get lost and confused as they try to navigate the next steps for a patient with a genetic condition or what services they even need. This quote identifies struggles that they personally faced, but many families share.

Another more negative emotion parents expressed was guilt. One parent explained it as:

[I've] been feeling for so long, for months, I thought it was my fault. I thought it was something that I did wrong. Something maybe that they didn't take the right vitamins during my prenatal. Nobody told me, nobody explained to me.

Nobody took the time to explain that it is not the parent's doings for this situation or that the situation could not have been prevented; that there is a gene or chromosomal change, which caused their child's condition. This lack of communication and knowledge on genetics combined impacted this parent, but unfortunately, she is not alone.

However, not all emotions during the meeting were negative. One major positive emotion was gratitude. Parents were thankful that something was being done to address their concerns, and that they were a part of an organization that is actively working on creating a change that benefits the lives of themselves and their children. One mother commented

I want to give a special thank you to [NYMAC and Family Voices personnel] to work with you in this project. It is important to me. As a mom, I thank you so much. All the work that we are doing to help children with special needs.

This quote represents how grateful parents are for the organization, for being included in the project and continuing the work that they are doing in recognizing the needs and barriers Puerto Rican families face. As a parent who has a child with special needs, this project is close to their heart, and it means a lot to them to be a part of it to ensure a better quality of life for their child.

Another major theme discussed were barriers Puerto Rican patients and families faced. Some barriers were listed as:

Unfortunately, like the services of the geneticists, sometimes they are not enough. In the community, providers, the cost of the tests, which tests, and which are the ones that doctors are suggesting and the insurance and so they need test the patients and for many families, the transportation.

This quote summarizes the list of barriers identified. The geneticists, providers, and the services they offer are sometimes not enough to address all the needs. In addition to not having enough genetic professionals, the specific tests needed, the tests suggested by medical providers, and the costs of the tests can be additional challenges for patients to receive services or a diagnosis. Insurance was a notable barrier by families. Insurance may not be able to cover the tests, and if it does, deductibles may be too high. The situation can also be that insurance does cover the exams, but to get the insurance to pay for it, you need to provide specific reasons, or jump through hoops, which further complicate a stressful situation. And lastly, transportation was identified as an issue. Even if patients can get a genetics appointment, they may still face the challenges of getting there. Many of the genetic clinics are in San Juan, the capital. For parents who do not live near San Juan, that would entail requesting a day off work or finding public or private transportation to get there if possible.

A major conversation point was on barriers for the deaf community. One individual proposed:

The doctor can call me for babies that have a hearing defect, and they cannot hear. It's a very important they communicate with families that are deaf.

If a patient has a hearing impediment, receiving a phone call from the doctor would not be suitable. Finding solutions for this community must focus on being accessible and appropriate. For some individuals, they were unfamiliar with or were unaware of barriers that exist for the deaf community, and the conversation was enlightening and provided a new perspective they may not have realized previously.

However, the discussion not only included barriers, but also potential solutions to overcome the barriers faced. The question focused on what solutions parents could come up with

that focused best on the challenges they personally face. For transportation, some solutions voiced were:

Maybe we can get some collaboration to get doctors to send them. Maybe the families can go to pediatric centers in the different regions of the island to have maybe an appointment doctor that is there and different locations.

Thus, having providers be spread out throughout the seven regions of PR, so that they can find a center or clinic closer to them that has what they need, was identified as a possible solution. Having a closer center also encourages more frequent care that is more accessible to families. In addition, collaboration with doctors can further the reach and network of genetic providers. By being able to communicate with other doctors, they may be able to find other avenues that better serve the patients and families.

Another helpful solution is telemedicine. As one of the public health professionals states:

We have telemedicine on the pediatric centers in the island.

Telemedicine is still widely used and scattered throughout the island to attend to the needs of patients. Telemedicine alleviates transportation struggles as families will not need to worry about finding the means of getting there or requesting an entire day off work. Telemedicine also helps doctors spend more time focusing on patients, rather than running around a hospital floor to see everyone.

One solution that focused directly on the deaf community was having interpreters. A public health professional mentioned that:

We would like to be able to have resources for people that work with sign language We already have a sign interpreter in some centers.

An interpreter acts as a bridge for patients and families that need to be able to understand what the provider is saying. This can be very beneficial for the deaf community, and people who speak another language. This service can also be very helpful for patients in PR, who currently need to go to the U.S. to receive some services.

Despite all those potential solutions, many of them faced some hesitation and potential challenges to implementation. For transportation, the solution of collaboration between doctors may be a challenge. One participant explained:

We don't have good collaboration with geneticists, so it's not happening yet....

I would like to mention that the access from the genetic tests coming from the lab. Not all the labs can do the tests. And the geneticists ask you for the tests [they] have. They have these problems sometimes because they don't have access to the lab that can do these tests.

Currently, there is not a good collaboration between geneticists and non-genetics providers. This may likely be due to the fact that there are only four geneticists on the island. While ideally collaboration would be the goal, PR's current situation does not allow for that. However, potential collaborations can occur with other states or slowly as more geneticists and genetic counselors come to PR. In addition, genetic tests are very specialized. Not every lab in PR has the genetic tests needed nor the staff who knows how to use the lab equipment. This causes doctors to have limited lab and test access and the need to find means of getting labs done elsewhere. This creates a further complication for families.

Despite the many benefits with telemedicine, it still has some limitations, especially if provided by genetic counselors. As described by one of the public health professionals:

Counseling is not as regulated at this time. So, I am not sure how to give that service since it is long distance. So that with certification, because every doctor that is going to give telemedicine services needs to have a certification.

Currently, genetic counseling is not well regulated in PR, and people, both patients and professionals alike, are unfamiliar with the process and what services can be accepted. In addition, for a provider to offer telemedicine, they must have a certification that approves them to provide telemedicine services. If a patient is seeking services, and the office does not offer those services, they would need to find a different location, often further away to get the services; still leaving transportation as a barrier.

In addition, there are some challenges with offering interpreters as a solution for access to the deaf community. As described by a participant:

So, because of staffing, we are trying to have a company that can help us with this signing interpreters in all of [the pediatric centers] It's really important for me to note this, because not everybody knows about this [signing interpreters service], and we said to them, "It's new! It's new!"

This quote indicates that there are not enough staff members currently to have signing interpreters in every pediatric center. So, while this service does exist, only certain locations offer it, simply because they do not have the personnel to offer it. Staffing can be caused by not enough professionals available or enough resources to afford the professionals, so having a contract with a company can help. In addition, the current addition of interpreters to pediatric centers is very new. Thus, many families and patients are unaware that this service exists.

Another major theme for families was being prepared for genetics appointment and how to do that. Patients recognize that doctors are very busy, and appointments are short, so coming

prepared with a list of questions and all their medical documents can make the appointment efficient:

From the folder, we put all the documents, how can we have them to create the folder in paper or electronic, how to have them organized in the folder for all the information they would need to take for the appointments because of the, um, because of the hurricane and pandemic.

A folder is an easy way to have all the necessary medical documents in one location, for easy access. The folders and documents can be converted electronically, as a PDF in a USB drive or in pictures on your phone, which can protect the original paper documents from natural disasters. This system encourages parents and families to empower themselves and take control of their health. It also encourages healthcare to be a collaboration between providers and patients. The use of a folder has been used by family members, and they have found this to be very important and efficient for their appointments.

Another theme is that families and patients seek support from organizations and other groups who share their struggles. The Family-to-Family Center (F2F) in PR provides direct support for families. They found out that families tend to know each other, so they meet through WhatsApp. They explain what that process has been like:

For [families] to discover all the tools to get them through if affected, and how to search for these support groups. If the family just have a baby, maybe they have no idea how this disease, so then we can help them and get the benefits, like support groups. Sometimes, these people in the mourning, they get isolated and that is an important process, and they need support.

Through this organization, people with similar situations can find a place and people to share their experiences with; the good and the bad. Someone who has been in the situation longer can help new patients and families in providing advice on the best providers, the process, what to do, and what questions to ask. If someone has faced recent loss, people may feel isolated and without resources, and support groups work on rectifying that. Creating support groups provides so many benefits to new and veteran patients with genetic conditions. Support groups can also become advocacy groups as people join, not only for support but to work on creating a change that better fits the health care needs of their community or their condition.

Throughout many of the themes identified, and in particular in the meetings with families and patients (March 12, 2021), language about the “importance” of many topics were frequent (mentioned dozens of times). Many points throughout this meeting were repeated to be “important” or expressed in some variation of the word. This emphasized how so many of these issues are important to families and patients. On the other hand, it can leave families feeling disheartened by all the challenges they face and overwhelmed by finding a way to prioritize just some of the important items, or just trying to keep track of all of them.

5.0 Discussion

This needs assessment highlights the current climate around the status of GS in PR, including challenges and barriers that arise for professionals, patients and families. Based on professionals' conversations, GSs are still very limited in PR, with a total of five genetic professionals to conduct all GS that exist. Barriers include available space to allow genetic professionals to practice, and insurance failure in recognizing genetic counselors, thus, making it difficult for patients to cover the costs of GS. Insurance and funding also create a barrier in establishing a telemedicine program. However, increasing collaboration with universities and others can foster relationships and aid in increasing GS and genetic professionals.

Based on conversations with families, patients and professionals, several factors of GS and its barriers can impact a family with special health care needs. Parents and family members may be feeling several emotions as they navigate having a child or family member with a genetic condition, both negative like fear, loss, and guilt, and positive such as excitement and gratitude. Patients and families face multiple barriers to accessing genetic services, including not enough genetic professionals, very specific tests and the cost for those tests, insurance, and transportation. Though some solutions were presented, such as collaboration between genetic providers and telemedicine, they too have their own constraints such as lack of a network, regulation of telemedicine for genetic counseling and providers needing certification for telemedicine. There was also an emphasis on barriers with the deaf community and a potential solution to address them; as a patient may not be able to hear the phone ring, let alone understand the information the doctor is trying to tell them. Other important themes include ways for patients and families to be prepared for their genetics appointments and providing support through organizations and support groups.

These conversations help give a better sense as to the current state of GS in PR from both a healthcare professional and patient/family lens. Through the themes, we can also note some major similarities between the two groups to be a lack of resources, specifically not having many genetic professionals on the island. Insurance being an issue for access was a major concern on both groups, as well as telemedicine being a viable solution, but the administration and execution of it has constraints.

This is not the first look into the status of GS in a Latin American country. A study published in 2012 focused on the genetic testing and services in Argentina. The barriers they face are like those listed in PR: availability and access of genetic services, genetic counselors are not a recognized or registered profession, high workloads, and genetic tests are difficult to obtain. Barriers that Argentina faces that are not determined based on this study are: ban on abortion for fetal indications leads to private market for prenatal screening with no quality control, genetic services in the public system are provided free of charge (though academic labs may charge fee), and no national guidelines or recommendations for the provision of medical genetic services. Some of the barriers listed were bureaucratic as each province has their own set of policies and budget, which lead to solutions being difficult to administer throughout the country (Penchaszadeh, 2013).

Another study looked at the clinical cancer GS for Latinos living in both the U.S. and Latin America. The barriers to genetic testing identified for US Latinos were cost or out-of-pocket expenses for testing, learning about future and familial cancer risk, fear of testing, competing life concerns and adverse psychological consequences. Medical genetic services in Latin America are mostly provided through public health services, but the service is often uneven and concentrated on urban areas. Barriers include fragmented health systems, deficient education in genetics, lack of explicit policies, patients' lack of knowledge, difficult accessing services, and resistance of

patients to seek genetic testing. These studies identify barriers similar to results from this project and illustrate GS barriers are prominent across Latin American countries (Cruz-Correa et al., 2016).

Limitations exist in this project that can impact the findings included. The meeting transcripts were recorded through zoom, but issues with video and audio quality affects what can be transcribed. It was sometimes very hard to fully capture words and conversations of what was being said during the meetings. This makes it incredibly difficult for the transcription, both for the application and the person reviewing transcription. Potential reasons behind the poor audio are partly due to the translators' audio interfering with the actual speaker, but also low internet connection, especially since PR can have a very spotty connection on the island. In addition, the automatic transcription application, Temi, does not transcribe in Spanish, only English. Thus, certain conversations missed were unable to be captured, even after manual translations and transcription. Although everything during the meeting that the interpreter said is translated, the interpreter may not have captured every word said, potentially due missing important details, context, or names. In addition, because of translations, tone behind transcription will be lost in the process. Emotions can be identified by what is spoken, but nonverbal cues, body language and tone are important to identify emotions in sections, where it may be significant. This situation was worsened given that the transcriptions were of translated discussions.

Qualitative methods are used to 'dig deep' into human perspectives within a particular context rather than to be broadly generalizable. These qualitative findings then help us understand the lived experience of 14 Puerto Rican stakeholders, sharing their expertise to better inform an understudied topic. Given that only a total of 14 PR stakeholders who participated in the meetings,

this is a very small sample size and it's not intended to be representative of the population. These are important limitations that affect this needs assessment.

Recommendations based on this needs assessment would be to encourage genetic providers, especially genetic counselors to practice in PR or provide education for current healthcare professionals in PR to be versed in genetics, labs, and services. One way of doing that is creating a policy that allows insurance to recognize genetic counseling as a profession; this helps both providers and families by providing financial incentive and covering costs of GS. Another way is by fostering collaborations between continental U.S. genetic professionals and those in PR. Funding should be focused on providing interpreter services and expanding hospitals throughout PR whose labs offer genetic tests, allowing testing to be more accessible for patients living throughout the island. These recommendations are similar to what was determined in a previous study that focused on clinical cancer genetic services in Latin America. They offer three main recommendations to promote access to clinical cancer genetics in Latin America: training of health professionals in genetic testing and counseling, educating the general public about genetic counseling and testing, and infrastructure development (Cruz-Correa et al., 2016).

Given the results gathered from this needs assessment, NYMAC plans on utilizing this information to gain clarity on the current state of GS in PR. NYMAC's goal is to ensure that individuals with heritable conditions and their families have access to quality care and appropriate genetic expertise. This information allows NYMAC to understand where PR is facing genetic disparities and focus on addressing them. NYMAC plans on supporting a regional infrastructure for GS, providing education and resources to families and providers, and facilitating the use of telehealth in genetics (NYMAC, 2022). Currently, NYMAC offers a wide array of educational and telegenetics resources, an online directory of genetics clinics, and an annual conference where

regional stakeholders share on a wide range of topics. With this project, NYMAC will be able to understand the needs and barriers specific to PR and implement funded projects, especially based on the recommendations provided, to improve access of GS.

The importance of this needs assessment is that it highlights health disparities that exist in the PR population. PR does not have enough resources to support the population in providing GS. However, the continental US also faces challenges with GS system and delivery (Innovations in Service Delivery in the Age of Genomics: Workshop Summary, 2009). Although interventions used in the continental U.S. are not always achievable or translatable to other locations, such as PR, the interventions could be a good steppingstone. This qualitative analysis focused on conversations with key PR stakeholders to illustrate these problems, but it can also explore what items can be used to move forward. This needs assessment highlights the limited public health efforts that currently exist to address GS in PR. Efforts in public health should prioritize building a bridge that aims at overcoming these barriers, but, unfortunately, that is not always the case. Future research and literature are needed on this topic as several gaps in knowledge still exist. Future research can investigate how social constructs of race and ethnicity play a role in receiving GS or in insurance, which can impact accessing GS.

Appendix A IRB Approval



Office of Research Protections
Human Research Protection Office

Hieber Building, Suite 106
3500 Fifth Avenue
Pittsburgh, PA 15213
412-383-1480
www.hrpo.pitt.edu

MEMORANDUM

TO: Coralys M. Carcana Barbosa, BS

FROM: Human Research Protection (HRP)

DATE: March 30, 2022

SUBJECT: IRB# 2203008: Status of Genetic Services in Puerto Rico: A Qualitative Analysis

The above-referenced project has been reviewed by the Institutional Review Board. Based on the information provided, this project has been characterized as being an activity that does not meet the formal definition of research, according to the federal regulations at 45 CFR 46.102(l)

That is, the proposed activity is not a systematic investigation, including research development, testing, and evaluation, designed to develop or contribute to generalizable knowledge. Should the scope of this project change such that the definition of research is subsequently met, the investigator must notify the IRB immediately.

Given this determination, you may now begin your project.

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