Implementation and Evaluation of a Genetic Services Referral Phone Line in the New York-Mid-Atlantic Consortium (NYMAC) Region

by

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Abstract

The NYMAC Regional Genetics Network implemented the Genetic Services Referral Phone Line in August 2018 with the goal of improving access to genetic services for medically underserved populations. During the first year, the phone line was under-utilized despite multiple marketing efforts to increase awareness amongst individuals and healthcare providers.

NYMAC staff are tasked with maintaining a regional genetics service directory and staffing the phone line. Between August 2018 and February 2020, phone line utilization was tracked and analyzed based on phone line call volume, number of callers who were referred to relevant genetic services or support resources, and number of individuals who contacted phone line staff via email. Online activity related to digital and email marketing campaigns was also tracked. Preliminary qualitative interviews were conducted to gauge healthcare provider perceptions of genetics and the phone line implementation process.

The phone line received 67 calls from 51 unique callers and 6 email inquiries between August 2018 and February 2020. A total of 16 phone and email inquiries were handled by NYMAC staff, providing those inquiring with relevant information for referrals or support resources. The initial digital marketing campaign delivered over 10 million impressions in targeted medically underserved areas throughout Maryland and Delaware and generated over 30,000 website clicks.

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The provider email campaign reached more than 48,000 healthcare providers in the region and generated 131 website clicks. A subsequent targeted digital marketing campaign launched in New York in January 2020 generated over 5,000 website clicks. Provider interviews identified common themes in perceived patient and provider barriers, as well as perceived utility of the NYMAC phone line and potential future resources.

As the phone line continues to receive sparse calls, additional analysis of call volume and marketing reach is needed in order to guide future implementation efforts. Future studies regarding individual or healthcare provider attitudes and beliefs about decreasing barriers and improving access to genetic services may be warranted. Improving access to genetic services in medically underserved areas is of public health significance, and lessons learned from this project will inform future NYMAC efforts to reach individuals in need of genetic services.

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Preface

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Abbreviations

AAMC	Association of American Medical Colleges
ABMG	American Board of Medical Genetics and Genomics
ACMG	American College of Medical Genetics and Genomics
APHMG	Association of Professors of Human and Medical Genetics
BCM	Baylor College of Medicine
CME	Continuing Medical Education
DO	Doctor of Osteopathic Medicine
DTC	Direct to Consumer
EHR	Electronic Health Record
EMR	Electronic Medical Record
F2F	Family to Family
FV	Family Voices
GARD	Genetic and Rare Diseases Information Center
GC	Genetic Counselor
GCA	Genetic Counseling Assistant
GP	General Practitioner
GSB	Genetic Services Branch
GTC	Genetics Track Curriculum (Baylor College of Medicine)
HIV	Human Immunodeficiency Virus
HRSA	Health Resources & Services Administration
MCHB	Maternal and Child Health Bureau
MD	Doctor of Medicine
MS	Multiple Sclerosis
MUA/Ps	Medically Underserved Areas and Populations
NCC	National Coordinating Center
NCHAM	National Center for Hearing Assessment and Management
NCHPEG	National Coalition for Health Professional Education in Genetics
NERGN	New England Regional Genetics Network
NORD	National Organization for Rare Disorders
NSGC	National Society of Genetic Counselors
NP	Nurse Practitioner
NRHA	National Rural Health Association
NYMAC	New York-Mid-Atlantic Consortium
NYSDOH	New York State Department of Health
P2P	Parent to Parent
PA	Physician Assistant
PCP	Primary Care Provider
RAINN	Rape, Abuse and Incest National Network
RC	Regional Collaborative Group
RGN	Regional Genetics Network
SERN	Southeast Regional Genetics Network
WSRGN	Western States Regional Genetics Network
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1.0 Introduction

With an increasing public health focus on reducing health disparities, improving access to genetic services has been a widely discussed issue in recent years. Prior needs assessments and surveys of rural populations in various U.S. states have identified perceived barriers to genetic services. Both individuals and healthcare providers identify lack of awareness of genetics services and physical location of genetic services as barriers to care¹, as well as access to genetic counselors.² While strides have been made in developing alternate service delivery models for genetic services³, the issue remains that individuals need to be made aware of such services, and may need assistance in identifying relevant services. This can be particularly challenging in Medically Underserved Areas and Populations (MUA/Ps), areas or populations within the U.S. designated by the Health Resources & Services Administration (HRSA) as lacking adequate primary care services, with high percentages of the population aged 65 or older and high levels of poverty and infant mortality.⁴

The New York-Mid-Atlantic Consortium (NYMAC) Regional Genetics Network aims to improve access to and awareness of relevant genetic services in the NYMAC region. In order to attempt to address some of these barriers to services, including within MUA/Ps, the NYMAC Regional Genetics Network implemented the Genetic Services Referral Phone Line in August 2018. The phone line was created to assist individuals and healthcare providers with locating the nearest genetic services provider based on zip code and the type of services needed. Initial phone line marketing campaigns targeted individuals in medically underserved areas in Maryland, and later Delaware as well as New York, with additional email campaigns targeted towards primary care providers. To date, phone line utilization has been much less than anticipated, despite efforts to increase awareness of the phone line among providers and the public.

This project focused on detailing the implementation process for the NYMAC Regional Genetics Network Genetic Services Referral Phone line, as well as conducting an evaluation of utilization and effectiveness of the phone line. Results of this project will serve to provide information about the strengths and limitations of phone line implementation efforts. Reaching medically underserved populations and connecting these individuals with relevant genetics services is an important step towards addressing the public health need to improve access to healthcare services. Results will guide future NYMAC efforts, and potentially those of other Regional Genetics Networks or genetic service providers.

1.1 Specific Aims

Aim 1: To describe the implementation process of the NYMAC Genetic Services Referral Phone Line.

Aim 2: To assess low call volume to the NYMAC Genetic Services Referral Phone Line.

Aim 3: To evaluate the effect of targeted marketing campaigns on NYMAC website activity and utilization of the NYMAC Genetic Services Referral Phone Line.

Aim 4: To identify potential limitations of the NYMAC Genetics Services Referral Phone Line implementation process through utilization evaluation and qualitative interviews.

2.0 Literature Review

2.1 Access to Healthcare Services

2.1.1 Disparities in Healthcare Services

According to Healthy People 2020, health disparities are differences in health between individuals or groups that are "closely linked with social, economic, and/or environmental disadvantages."⁵ These disparities exist across racial and ethnic groups, special populations, gender, disability status, age, socioeconomic status, and location.⁶ Different geographic locations within the U.S. have different levels of morbidity and mortality, with health disparities being prominent in both rural and urban areas, each with unique challenges related to healthcare.⁶ According to the National Rural Health Association (NRHA), individuals living in rural areas have decreased access to physicians in comparison to individuals in urban areas, and on average, also have a larger elderly population (aged 65 and older) and lower socioeconomic status.⁷

Medically Underserved Areas and Populations (MUA/Ps) are areas or populations within the U.S. designated by the Health Resources & Services Administration (HRSA) as lacking adequate primary care services, with high percentages of the population aged 65 or older and high levels of poverty and infant mortality.⁴ Based on the NRHA description of the rural healthcare climate,⁷ many areas designated as MUA/Ps may also be considered rural, with some health disparities and barriers potentially attributed to workforce shortages^{8,9}.

A survey of rural hospital CEOs identified physician shortage and workforce needs, with an average of over 75% of these CEOs nationwide identifying an overall physician shortage in their hospitals.⁸ Notably, 100% of the rural CEOs surveyed in the New England to Virginia region indicated an overall physician shortage.⁸ Shortages in at least two primary care specialties (including family medicine, internal medicine, and pediatrics) were identified by 70.3% of CEOs nationwide, with a higher rate of these shortages in the New England to Virginia region (83.9%).⁸

2.1.2 Genetic Service Provider Workforce Shortages

Workforce shortages also exist within genetics services, with a 2018 workforce study suggesting that as genetic testing becomes less expensive and more widely available, the demand for genetic services and testing will increase; however, the ability for the genetics providers nationwide to meet this demand is limited by a workforce shortage of genetics providers, specifically genetic counselors (GCs).⁹ Using a goal rate of one full-time equivalent (FTE) GC per 75,000 persons, their model indicated that the shortage of GCs working in direct patient care may not be resolved until the year 2030, taking into account the inevitable growth of the profession as well as the changing nature of healthcare systems.⁹ With these results, Hoskovec et al. concluded that to address the demand for genetic services and shortage of genetics service providers, focus must be put on expanding genetic counseling training programs, exploring ways to involve non-genetics providers in genetics services, and integrating tools to increase efficiency of practicing GCs.⁹

A 2003 workforce analysis of American Board of Medical Genetics (ABMG) certified medical geneticists in the United States was completed with a 55% response rate.¹⁰ Weighing their findings for the living and active medical geneticist population at the time (n=1377), Cooksey et al. found that approximately 70% of MD geneticists provided direct patient care, with those providing patient care spending approximately half of their working time on activities related to

patient care.¹⁰ Citing a previously published ratio at the time of approximately 3.5 MD clinical geneticists per 1 million people in the U.S, the analysis showed variation across regions of the United States, with some states averaging 2.0 or less geneticists per 1 million people, and some states with no clinical geneticists, suggesting an insufficient supply of medical geneticists in 17 states acting as a potential barrier to access to genetics services.¹⁰

2.1.3 Barriers to Accessing Genetics Services

Studies aimed at better understanding the healthcare needs of medically underserved populations, including genetics-related needs, can be instrumental in addressing potential barriers to accessing services. In addition to workforce shortages,⁹⁻¹¹ barriers to access to genetics services for individuals living in MUA/Ps, including rural areas, as well as how to address these barriers have also been widely studied in recent years. As a follow-up to Cooksey et al.¹⁰, a 2015 needs assessment identified several genetic service components which could benefit from resource development.¹¹ This needs assessment surveyed 924 genetics professionals and providers to gather additional information about aspects of genetic services which could be addressed to improve access, which included appointment wait times, underserved populations, and workforce shortages.¹¹ Of those surveyed, 43.5% reported geneticist job openings at their institution, with 100 medical geneticist openings and 200 GC openings reported across institutions.¹¹ The assessment reported increased appointment wait times between the Cooksey et al. study and the 2015 needs assessment; wait times of over 3 months for a nonemergency new patient appointment were reported by 10% of respondents in 2003, and increased to over 30% of respondents in 2015.¹¹ Estimating an updated workforce ratio of 2 clinical geneticists per 1 million people, the assessment suggested that increased patient referrals and wait times, paired with a shortage of genetic service

healthcare providers greatly reduces patient access to genetic services.¹¹ This needs assessment also identified that individuals living in rural areas do not access genetic services, nor do individuals who are uninsured or do not speak English.¹¹ Distance to services, lack of genetic service providers, and primary care provider recognition and awareness were among identified barriers.¹¹

A needs assessment completed by the Michigan Department of Community Health between 2000-2002, targeted individuals with genetic conditions, parents of children with unique health concerns, a sickle cell anemia parent support group, a Native American student group, and genetics service providers to identify barriers to access to genetic services for these populations, as well as potential ways to address the identified barriers.¹ Focus group participants identified lack of knowledge and awareness of genetic services, by both individuals and providers, as a barrier, as well as lack of awareness of personal risk and concern for discrimination.¹ Additional concerns included workforce shortages, referral coordination, cost, insurance, and distance to genetic services.¹ Individuals or families affected by genetic or other health concerns reported relying heavily on internet resources for information, while individuals who were unaffected reported relying on their healthcare providers for information they needed about genetics.¹ Suggestions to improve access to information about genetics and genetic conditions included a 24hour hotline and centralized source of necessary information, with focus groups also suggesting increased efforts in implementing telemedicine and additional outreach clinics to improve access to genetics services for underserved populations.¹

A recent survey of individuals living in rural Illinois communities captured identified gaps in knowledge, attitudes, and perceived barriers to access to genetic testing in relation to cancer genetics.² Individuals surveyed lived in "isolated communities," or towns with populations of approximately 1500 persons which were 20 or more miles away from a larger town (defined as having a population of over 5000 persons).² 69% of these individuals were aware that genetic services were available in the context of cancer genetics, while 46% were aware that genetic services are available for other, non-cancer conditions.² In addition, a little less than half of those surveyed noted that not having GCs or genetic services close to their location was a barrier to services.²

Barriers to genetic services have been identified in racial and ethnic minority groups as well.¹² In an analysis by Suther and Kiros, national representative sample data were obtained and analyzed to identify and predict potential barriers to genetic testing as they relate to racial and ethnic healthcare disparities.¹² In comparison to 63.5% of non-Hispanic white individuals, 43.8% and 48.7% of Black and Latino individuals, respectively, knew someone having a child "born with a genetic defect."¹² This analysis also showed an association between race/ethnicity and genetic testing knowledge, with the odd of Black and Latino individuals having appropriate knowledge was 28% and 52% lower, respectively, than non-Hispanic white individuals.¹² Individuals identifying as Black or Latino were also more likely to be concerned about misuse of genetic testing, with this concern being 66% and 58% higher in Blacks and Latinos, respectively, in comparison to non-Hispanic whites.¹² Overall, data analysis suggested that minority groups are less likely to utilize genetic services due to barriers which include lack of knowledge, as well as lack of physician-provided information, health insurance coverage, and distrust of healthcare providers and the medical system.¹²

A 2012 study interviewed 120 at-risk Latina women in the New York City area with a suggestive personal and/or family history of cancer and no prior genetic counseling or testing to assess perceived barriers and perceptions of genetic counseling for *BRCA1/2*-related indications.¹³

More than half of the women interviewed had relatively little or almost no awareness of the availability of genetic counseling for hereditary conditions or cancer, and nearly half of those interviewed had little awareness about genetic counseling for breast and ovarian cancer.¹³ Over one third of participants identified competing health or life concerns as a barrier to pursuing *BRCA*-related genetic counseling, with the majority of those interviewed sharing logistical concerns, including uncertainty of where to go for genetic counseling, time constraints, and insurance coverage.¹³

In a study completed by Mountcastle-Shah and Holtzman, 60 primary care providers across four specialties (pediatrics, obstetrics, family medicine, and internal medicine) were interviewed regarding perceived barriers of integration of genetics into their practice.¹⁴ Respondents discussed lack of demonstrated clinical utility of testing as the most common barrier, with 60% of providers stating they would show some hesitation to order a genetic testing without an adequate understanding of how it would affect patient care.¹⁴ Additional barriers identified included clinical validity, cost, and concern for loss of insurance coverage.¹⁴

Barriers to genetic services as perceived by primary care providers were further explored in a 2003 review completed by Suther and Goodson.¹⁵ Across 18 publications, the most common barrier identified was provider lack of knowledge of genetics and genetic testing and counseling.¹⁵ Suther and Goodson comment that while this is the most commonly cited barrier, it may be based on the potentially problematic assumption that providers' lack of knowledge is "undesirable," suggesting that rather, the perceived lack of knowledge should be explored further to determine if this lack of knowledge translates to uncertainty surrounding genetic testing.¹⁵ Other commonly identified barriers included lack of or outdated family history information, lack of referral guidelines, decreased confidence amongst providers, and insurance or financial concerns. A 2015 review by Delikurt et al. also identified barriers to patient referral to genetic services.¹⁶ Reviewing nine publications, barriers related to both individuals and healthcare providers were identified.¹⁶ Individual barriers to genetic services identified by Beene-Harris et al.¹ were summarized, including lack of personal risk awareness, lack of knowledge of family medical history, and lack of knowledge of available genetic services, the latter of which was reported in two additional publications.¹⁶ In regard to healthcare providers, perceived barriers to patient access included lack of adequate family history and lack of provider knowledge of genetics and awareness of genetic services.¹⁶ Additional barriers related to healthcare providers were lack of awareness of patient risk, referral coordination issues, and genetic workforce shortages.¹⁶

From the genetic service provider standpoint, similar barriers to access to genetics services have been identified. In a survey of 28 genetic service providers from across the U.S. (GCs and medical geneticists), 72% of respondents noted that pursuing cancer genetic counseling was not an apparent priority for patients.¹⁷ 52% of providers cited patient concerns regarding insurability, and 44% also noted lack of insurance coverage as perceived patient barriers to cancer genetic counseling services.¹⁷ Nearly half (48%) of the survey respondents reported distance to genetics services as a perceived patient barrier, and 36% included lack of knowledge about or understanding of genetic counseling by both patients and other healthcare providers as an additional patient barrier.¹⁷ Additional perceived barriers included patients being discouraged by family members, fear of results and their impact on a patient's family or employment, and lack of time.¹⁷ This study's respondents also provided suggestions for addressing these potential barriers, with the majority (70%) suggesting increased provider education.¹⁷

In a study interviewing 6 GCs working in rural areas, the GCs shared that they see diverse populations, including elderly populations and individuals from "economically diverse" backgrounds and individuals with rare conditions.¹⁸ They identified that the distance patients must travel to an appointment, lack of awareness of genetic conditions, the availability of genetic services, providers lacking knowledge of when to make an appropriate referral, and lack of clinical geneticists are potential barriers to patients accessing genetics services.¹⁸ GCs practicing in rural areas have also identified several factors which may decrease some barriers to genetics services for rural and underserved populations, including professional resources and support systems, adapting clinic schedules and availability and adding outreach clinic locations to meet the needs of the unique population they serve, and implementing alternate service delivery models.¹⁸

2.1.4 Alternate Service Delivery Models

With increased demand for genetic services and genetic testing in a genetics workforce shortage climate, implementation and evaluation of alternative service delivery models is necessary in order to improve access to genetics services.¹⁹ Traditional models of genetic counseling, which are often in-person and often require a significant amount of time pre-, during, and post-session, may not support increased patient access and/or GC efficiency, and thus, many GCs have adopted additional alternate service delivery models.^{3,19} Multiple service delivery models have been developed as a way to address the various identified barriers to genetic services, including telegenetics and telephone genetic counseling options to improve access for individuals located a far distance from genetics services, genetic counseling assistants to assist with decreasing wait times for genetic counseling appointments, and group genetic counseling and use of genetic counseling extenders to address the workforce shortage of GCs.³

To assess the support for and utility of group genetic counseling, a 2017 study described the experiences and successes of group genetic counseling in a prenatal setting.²⁰ In the study, 172

women referred for prenatal genetic counseling following positive serum screening for Down syndrome were systematically assigned to an individual genetic counseling appointment (37.8%) or group genetic counseling appointment (62.2%).²⁰ Group genetic counseling sessions were made up of 2-6 patients, as well as their partners, and one GC, with participants having the option to stay for an individual meeting with the GC following the group session.²⁰ Using pre- and postcounseling questionnaires, Cloutier et al. were able to asses patient anxiety, knowledge, and satisfaction.²⁰ This study found a significant decrease in patient anxiety and decisional conflict, as well as an increase in perceived personal control and knowledge scores for all study participants who received genetic counseling, with similar patient satisfaction in both service delivery models.²⁰ Significant differences were not shown between study groups in assessments of perceived personal control, decisional conflict, or knowledge; however, those who had received individual genetic counseling were found to have significantly decreased anxiety as compared to those who received group counseling and the GC spent less time per group patient compared with time spent with individual patients.²⁰ These findings suggest that group counseling may be an acceptable approach to providing genetic counseling to patients; however, 85% of group genetic counseling patients elected to stay after the group session for an individual discussion with the GC, suggesting the importance of the availability of individualized counseling.²⁰ This study highlights that group genetic counseling is feasible and can increase the time efficiency of GCs, which can increase access by allowing more patients to be seen by a genetic service provider, but limitations do need to be acknowledged.²⁰

While not a formal service delivery model, the addition of genetic counseling assistants (GCAs) to genetics practices has been suggested as another potential solution to increasing patient access and clinic efficiency, as described in a 2018 study by Hnatiuk et al.²¹ Following a survey

of 271 GCs, including those who had worked with GCAs previously and those who had not, GCAs were determined to primarily perform clerical (93%) or administrative tasks (83%) and data entry, with a smaller number of GCs (\leq 20%) reporting working with GCAs carrying out more clinically involved tasks, such as test coordination, letter or report writing, and calling out test results.²¹ Using open ended questions, study participants were able to comment on GC and clinic efficiency, stating that GCA integration into their institutions allowed GCs to spend more time on tasks for which they were specifically trained, which allowed more patients to be seen and overall, increasing patient access to genetic services.²¹ This assessment advises that GCAs are not a single solution to increasing patient access; however, adoption of other alternate service delivery models in addition to GCA integration can maximize this efficiency and increase access.²¹

In a 2013 report by the National Society of Genetic Counselors (NSGC) Service Delivery Model Task Force, 54.7% of survey respondents reported using only traditional genetic counseling models in practice; however, the remainder of respondents reported adoption of at least one alternate service delivery models, including telephone genetic counseling, group genetic counseling sessions, and telegenetics.¹⁹ Telegenetics, or telemedicine, is typically understood to be a genetics services consultation completed via video and audio connection, providing a benefit for individuals who may live in an areas without genetics services available, or who are unable to travel to an area with genetics services available.²² Among a survey of self-identified telegenetics programs in seven states within the New York-Mid-Atlantic region and Washington, D.C., respondents reported increased patient access through decreased wait times, increases in the number of patients seen per month, and a geographic reach of over 200 miles.²² These survey respondents reported that telegenetics services were particularly successful in reaching individuals who otherwise may not have been able to access genetics services.²² A 2015 study by Buchanan et al. describes a random trial to assess telegenetics services in cancer genetic counseling.²³ In the trial, 162 patient participants were randomized to receive genetic counseling at a local rural oncology clinic or via telegenetics, with subsequent assessments of cost, attendance, and patient satisfaction.²³ Telegenetics consultations were found to be more cost efficient, with a per-patient cost that was \$138.14 less than in-person genetic counseling.²³ Post-appointment surveys showed no significant difference in patient satisfaction between those who had telegenetics appointments and those who did not; however, study participants were more likely to attend an in person appointment (89%) versus a telegenetics appointment (79%).²³ These findings suggest that telegenetics may be a feasible and cost-effective way for clinics to increase access to genetic counseling; however, it should be noted that telegenetics may still not be an appropriate solution to reach all underserved populations.²³

Alternate service delivery models, including telegenetics, will likely be imperative to linking more individuals with genetics service providers in the future, by helping to decrease barriers affecting genetic service utilization by patients and healthcare providers.³ It will be important; however, to better understand how to increase awareness of the availability of such service delivery models. The increasing availability and successful integration of alternate service delivery models will not only address barriers to individuals accessing genetics services, but may also lend to more streamlined integration of genetics into primary care practice in the future.³

2.2 Genetics in a Primary Care Setting

2.2.1 The Role of the Primary Care Provider

A primary care provider (PCP) has been defined as a provider "who provides, coordinates, or helps a patient access a range of health care services."²⁴ According to the Institute of Medicine (IOM) Committee on the Future of Primary Care, primary care encompasses care provided by providers who "are accountable for addressing a large majority of personal health care needs, developing a sustained partnership with patients, and practicing in the context of family and community."²⁵ Primary care has extended beyond duties of doctors of medicine (MDs) and doctors of osteopathic medicine (DOs) practicing in family or internal medicine, pediatrics, and obstetrics and gynecology to also include non-physician clinicians, including nurse practitioners (NPs) and physician assistants (PAs).²⁵

PCPs serve as a central point of contact for their patients, providing long-term care for a variety of medical concerns and promoting overall health and wellness, while also providing a link to the broader health care system.²⁶ PCPs regularly use referrals to connect their patients to specialty care to address more complex or specific healthcare needs,²⁶ with genetics being included in such specialty care. With the increasing demand for genetic services and the workforce shortage of genetics healthcare providers, the integration of genetics into primary care is becoming an important component of healthcare.²⁷ PCPs and other key informants within one health system have acknowledged that it is a PCPs duty to facilitate relevant genetic care for their patients, with one PCP stating their role as identifying genetic conditions and placing an appropriate referral.²⁷

2.2.2 Genetics Education for Primary Care Providers

Medical education is typically structured with both didactic coursework and clinical experiences and skill development, with didactic courses emphasizing basic biomedical concepts, as well as keeping current with advances in the field, such as chronic illness, geriatrics, and population health.²⁸ Medical school curriculum is governed by each specific institution, with general competencies used to guide curriculum development, but no standardized coursework implemented across institutions.²⁸ Similar competencies have been developed for PAs²⁹ and NPs^{29,30}. This likely accounts for differences in the amount and type of genetics education a PCP in training may experience. According to the Association of American Medical Colleges (AAMC), approximately 19% of medical schools' curricula in 2017-2018 included an independent genetics course.³¹ More commonly, genetics has been increasingly taught as part of an integrated course, incorporated with other topics in which genetic conditions may be relevant.³¹ However, it has been estimated that genetics makes up an average of 2% of the didactic content at AAMC member institutions.³²

The Core Competency Working Group of the National Coalition for Health Professional Education in Genetics (NCHPEG) was established in the early-2000s to address potential gaps in genetics education for all types of healthcare providers.³³ These core competencies include knowledge of basic genetics concepts, ability to gather and assess family medical history, capacity to provide patients with education and relevant resources, and recognition of perspectives which influence genetic services and testing.³³ While the competencies were created as a resource to encourage providers to address gaps in genetics knowledge, the NCHPEG recommends that, at a minimum, all healthcare providers should be able to recognize the limitations of their genetic

knowledge, understand the implications of genetics, and know when to refer their patients to an appropriate genetics provider and how to facilitate such referrals.³³

Similarly, the Association of Professors of Human and Medical Genetics (APHMG) have developed a core curriculum for competency-based genetics education in medical schools.³⁴ Taking into account the changing landscape of genomic medicine as a whole, the core curriculum incorporates emerging topics of which all medical school graduates should have knowledge.³⁴ The APHMG curriculum were developed as a framework that will allow medical schools to incorporate genetics education in a variety of ways, including standalone or integrated coursework.³⁴

Assessment of historical and current genetics integration within medical education has consistently shown that PCPs find their genetics education to be lacking.^{32,35–37} A 2008 study among individuals in family medicine residency programs showed that residents had learned about genetics early in their medical education, and typically in the context of rare diseases, making it difficult for these individuals to find relevance for genetics in everyday clinical practice.³⁶ It was also noted that during residency, genetics was primarily encountered in pediatrics and obstetrics, so these specialties become more associated with genetics.³⁶ Suggestions have been made that medical education should broaden the scope of genetics integration, framing genetics beyond the context of rare conditions, with more consistency between institutions.²⁷ It has also been suggested that concepts should be taught in the context of clinical cases and that genetics should remain a separate course in medical school curriculum in order to increase provider knowledge.³⁷

In a survey of 112 medical genetics course directors at medical schools in the United States and Canada, Plunkett-Rondeau et al. determined that 75% of schools taught the majority of their genetics curriculum in the first year of medical school, with only 26% reporting formal genetics education in the last two years of medical school.³² In this survey, genetics was found to be a standalone course at 25% of schools, with it being more common for genetics to be integrated with other science content, such as biochemistry, and 7% of schools integrating genetics content with clinically relevant topics.³² Over half of respondents (60%) reported using the APHMG core curriculum to guide curriculum content and development at their institution.³²

A 2012 report by Baylor College of Medicine (BCM) details efforts to increase genetics exposure for their medical students within their institution.³⁸ Utilizing an existing "track" system, in which medical students can personalize their education, BCM has established the Genetics Track Curriculum (GTC).³⁸ Establishment of GTC resulted in the addition of several genetics electives to the BCM curriculum, and while the GTC is aimed at medical students who wish to gain more knowledge and experience specifically with genetics and genomics, courses are available to any medical student.³⁸ GTC courses span across all four years of medical school and allows students to build upon foundational genetics concepts, enhance their clinical experience, and gain a better understanding of clinical diagnostics and patient experience.³⁸ Dhar et al. suggest that development of this specialized curriculum within BCM can not only enhance the educational experience of those who wish to become medical geneticists, but can also provide a greater breadth of education so that those training in other specialties, such as primary care or internal medicine, can be equipped to provide better patient care and personalized medicine in their practice.³⁸

In a recent special report, Hyland, Garber, & Dasgupta detail genetics-related efforts in medical education.³⁹ In addition to medical schools' efforts to engage students, further integrate genomics into curriculum, and expand genetics education across all years of medical school, the report describes initiatives by professional organizations, including APHMG, Association of Pathology Chairs Undergraduate Training in Genomics working group, and ACMG.³⁹ These organizations have all developed and continue to work on developing teaching resources and

educational activities, peer-reviewed resources, adaptable lesson plans, and extracurricular programs for medical students, including ACMG sponsored student interest groups (SIGs) and a summer program which allows medical students to gain clinical genetics experience.³⁹ Hyland, Garber, & Dasgupta acknowledge the availability of genetics- and genomics-focused continuing medical education (CME) opportunities for healthcare providers; however, to further address gaps in genetics education, they suggest enlisting the help of "genomics champions," or individuals with genomics knowledge within their specific practice specialty who can educate and coach others.³⁹

2.2.3 Primary Care Providers' Perceptions of Genetics

As genetics becomes an increasingly prevalent topic in today's society, and a more integrated part of contemporary healthcare, it is important to address how the topic of genetics fits into the role of the primary care provider. There has been widespread interest in ascertaining providers' perceptions of genetics, with multiple research approaches being used to elicit provider understanding, concerns, and suggestions regarding integration of genetics into primary care practice.^{40–44} Concerns remain about PCP readiness to integrate genetics into their practice, as PCPs have self-identified themselves as having limited genetics knowledge and educational background.^{35,40}

A pervasive concern amongst PCPs is that they do not know enough about genetics and/or genetic testing, and that they do not feel prepared to answer questions that patients may have about these topics. A study published by Harding et al. in 2018 focuses on PCPs' experiences with genetics following 10 interviews and 5 focus groups with PCPs and other key informants in rural and urban locations within Canada.⁴⁰ In regard to their experiences with genetics in practice, both

rural and urban PCPs shared having more genetics exposure in the prenatal and cancer settings, and considered genetic information to be beneficial.⁴⁰ PCP participants had experience ordering genetic testing for common indications; however, concerns arose regarding further integration of genetics into future primary care practice without appropriate support for PCPs.⁴⁰ It was noted that PCPs' lack of genetics knowledge may impact decision-making regarding genetics referrals for their patients, with a need for more education, resources, and PCP support.⁴⁰ Participating PCPs suggested a need for further continuing education options, including updates via email and mail, print resources such as pamphlets, and an online genetic condition database.⁴⁰ Those in rural locations also expressed interest in having easy access to a genetics expert via phone or email.⁴⁰ Providers reportedly acknowledge that genetics is becoming part of primary care practice, but feel that keeping up to date with genetics knowledge is "tricky," and additional barriers influence PCP reports that they are not prepared for their potential roles in regard to integrating genetics in their practice.⁴⁰ Concerns were raised that genetics may be outside of the scope of primary care practice, with identified barriers including the availability of funding to establish PCP resources, location and travel limitations, lack of understanding and knowledge of particular conditions and test result implications, and patient apprehension.⁴⁰ Of note, those in rural locations also described a phenomenon in which individuals living in rural areas "learn to manage without" certain healthcare services, and when these services become available to them, the perceived utility may be low.⁴⁰

An Australian needs assessment completed by Metcalfe et al. identified similar provider concerns during focus group interviews with 39 general practitioners (GPs) from a variety of clinical specialties and geographical areas.³⁵ GP participants shared that often they discuss genetics with patients only in specific high risk scenarios, such as advanced maternal age in pregnancy or suggestive ethnic background or family history, with one GP concerned that patients often present

knowing more information about a particular genetic condition than the GP does.³⁵ GPs raised concerns about patient psychosocial responses related to genetic testing, as well as GP ability to explain concepts in lay language or appropriately convey risks.³⁵ GP educational concerns were an additional theme amongst focus group participants, with GP knowledge level found to be dependent on the timeline of their medical education.³⁵ Providers noted difficulties in keeping up to date with genetic developments, and that additional knowledge was often learned in context of clinical experiences.³⁵ It was noted that efforts surrounding GP genetics education would likely improve knowledge, with GPs suggesting distance education opportunities, case-based seminars, electronic and print resources, as well as a telephone hotline/advisory service as potential sources of information related to genetics.³⁵

Concerns repeatedly arise that providers do not receive adequate genetics education in their training, with a study by Lopes-Júnior et al. reporting that 57.4% of PCPs and nurses surveyed (n=54) did not feel that their undergraduate genetics education was relevant to their clinical practice.⁴⁵ Of those surveyed, 72.2% were aware of the importance of genetic counseling; however, 62.9% were unaware of institutions that offered genetic counseling services.⁴⁵ The majority of PCPs and nurses surveyed had participated in the care of an individual with a genetic condition, but 77.8% shared being unprepared to care for these individuals.⁴⁵ 33.3% of respondents were unsure of how and where to refer patients and over 90% of respondents indicated they had not had any training in genetic screening or genetic counseling, with only 27.7% of those surveyed demonstrating appropriate genetic counseling knowledge.⁴⁵

This perceived lack of knowledge has been identified as a barrier to patients receiving genetics services, impacting PCPs' ability to recognize appropriate patients to refer to genetics⁴¹, as well as their ability to identify resources that can assist them.⁴³ In a qualitative study of 51 PCPs,

Carroll et al. identified limited exposure to personalized medicine, with most having experience in the realm of hereditary breast cancer or the prenatal setting.⁴¹ Lack of knowledge was a recurring theme in interviews, with providers expressing anxiety and apprehension regarding discussion of personalized medicine with patients.⁴¹ Some PCPs also shared that their attitudes and perceptions were shaped by personal experience, with personal feelings influencing discussions with patients.⁴¹ PCP participants in this study also identified uncertainty in when to make referrals and lack of relationships with genetics professionals as barriers.⁴¹ The study identified PCP needs related to personalized medicine, including more education and information about screening management, treatment, referral guidelines, available genetic tests and information about cancer genetics, and genetic testing benefits.⁴¹ PCPs also identified resource needs, such as point-of-care tools, updated and reliable information sources, educational sessions, and resources within clinic, such as a go-to person or resource who could answer their questions related to genetics-related care for their patients.⁴¹

In an online survey of 88 PCPs recruited from the American Academy of Pediatrics' Quality Improvement Innovation Networks, Rinke et al. aimed to identify PCP current practices and attitudes surrounding genetics.⁴³ The majority of respondents reported ordering genetic testing zero to three times annually, with an average of 4.8 patients per year referred for genetics evaluation.⁴³ Of those surveyed, 89% reported having access to a genetics professional, with 75% reporting these professionals are located 30 miles or less from their practice.⁴³ Less than half of PCPs felt competent providing genetics care to patients, with 94% respondents indicating that an increased understanding of genetics would allow them to more effectively integrate genetics into practice.⁴³ 59% of respondents reported that they did not have adequate resources to determine when to order an appropriate genetic test, and 42% of respondents in the study were aware of ≤ 1

national resources for genetic services and information. Rinke et al. suggest that provider education, leveraging existing provider resources, and increasing access to genetic services could improve the diagnosis and care of children with genetic conditions.⁴³

Providers have identified physical barriers to access to genetic services for their patients as well, stating that they have no established relationships with GCs or genetics providers to refer to^{41,44} or lack GCs in the area in which they practice⁴⁴. Of note, Diamonstein et al. found that of the providers in their study who reported no GCs in their area, one-third of these providers had GCs practicing within 10 miles of their practice location, indicating the importance of increased education and making providers aware of genetic services and GCs in their area.⁴⁴ Additional barriers that have been identified by PCPs include concern for patients' mental health, insurance discrimination, and cost.⁴²

In addition to barriers attributed to genetic knowledge and availability of genetics service providers, PCPs have also acknowledged struggles related to "direct to consumer (DTC)" genetic testing⁴¹, or genetic testing which can be initiated by a patient/consumer with or without prior consultation with a healthcare provider or genetics specialist. PCPs have shared that they feel they have very little knowledge about DTC testing, and have gone as far as to describe it as "scary."⁴¹ In a study completed by DTC genetic testing company 23andMe, 130 PCPs were surveyed regarding attitudes and perceptions of DTC testing.⁴⁶ Just over one quarter of PCPs surveyed felt comfortable with their knowledge and ability to help patients understand DTC genetic test results prior to completing DTC genetic testing themselves as part of the study.⁴⁶ With patients taking on a more active role in their healthcare and medical decision-making, DTC approaches for genetics, as well as other aspects of healthcare, will provide unique challenges to the changing landscape of primary care.⁴⁷

2.3 Hotline Availability and Evaluation Studies

Telephone call centers or hotlines have been long established as resources to connect individuals with services in a variety of settings, including healthcare, consumer services, and counseling. For example, the Rape, Abuse and Incest National Network (RAINN) has successfully implemented both phone and internet-based hotlines with the goal to provide support, advice, and advocacy to victims of sexual assault.⁴⁸ Staffed by trained volunteers with services similar to inperson rape crisis center services, evaluation of these hotline services has shown that RAINN has been able to effectively extend their services to areas and communities which may otherwise lack in-person services.⁴⁸ This service model may also be successful due to the added layer of anonymity for callers/users.⁴⁸

Studies evaluating the implementation and utilization of hotlines in any setting are critical to understanding the effectiveness of the service.^{49–51} The Department of Veterans Affairs have detailed the implementation of a suicide hotline for veterans, describing the challenges of targeting veterans, a population which is largely adult males, and which does not fit the typical hotline caller demographic.⁴⁹ This Department of Veterans Affairs hotline was promoted using targeted advertising campaigns, including posters on public transit vehicles, and call data correlated with an increase in referrals to the department's suicide prevention group following hotline implementation.⁴⁹

Hotline evaluation projects can also help in identifying limitations and areas of improvement for underutilized hotlines.^{51,52} The Heath Care Safety Hotline was implemented in 2014 and designed as a way for consumers to report patient safety concerns in a healthcare setting.⁵² The hotline and an accompanying website were operational for 17 months, implemented within two healthcare organizations and staffed by research team members.⁵² The availability of

the hotline was marketed through brochures, hospital websites, and pamphlets provided at time of patient discharge; however, the hotline was widely underutilized, with a limited number of calls.⁵² Evaluation of hotline webpage traffic showed that more individuals were accessing the website than utilizing the website or hotline to make safety reports.⁵² Schneider et al. note that the outreach was expanded and outreach strategies were refined over the 17 month period; however, the report volume remained "suboptimal."⁵² The group who developed the phone line was able to acknowledge the potential of the phone line, and highlighted that the healthcare systems were able to use the available data to address the concerns of those who did utilize the reporting system.⁵² The study suggests that the implemented hotline prototype can be adapted and can be scalable to other settings, and suggests future efforts be directed at evaluation of outreach strategies and online platforms as well as sharing hotline materials within a public domain to increase the use of the developed prototype and materials.⁵²

Modifications in hotline implementation have been shown to increase utilization by target audiences.⁵³ A German-based medical group developed a hotline for individuals with multiple sclerosis (MS) and caregivers of individuals with MS to obtain additional information and resources about palliative care.⁵¹ This hotline was advertised in a print magazine targeted to the MS community, as well as on pamphlets shared with MS caregivers and healthcare providers; however, the hotline received only 18 calls during its pilot year.⁵¹ The group implementing the phone line acknowledged the low call volume, but believed that the fact their hotline was being used at all suggested its potential to better serve the MS community.⁵¹ Follow-up assessment of this same hotline two years after its implementation and expansion to more areas of the country showed an increased call volume, with the hotline receiving an additional 222 calls over a 27 month period, suggesting the hotline is a valuable resource for this target group.⁵³

Hotlines are not only designed as a resource for patients and consumers, but can also be targeted to healthcare providers, such as the *Uliza!* HIV hotline based out of Kenya.⁵⁰ This hotline was created to provide consultation services to healthcare providers in rural, under-resourced areas of Kenya with a high burden of HIV.⁵⁰ Available 24-hours per day, 7-days per week, the *Uliza!* hotline was staffed by consulting expert HIV physicians equipped with internet resources and a specialist support network and aimed to take advantage of the widespread cell phone use by providers in remote areas.⁵⁰ The hotline was presented to healthcare providers and nurses from study sites at a publicity meeting, and the *Uliza!* group relied on word of mouth, text message promotions, and continuous medical education sessions to further advertise the availability of the hotline.⁵⁰

Following implementation, the *Uliza!* group completed an evaluation study, assessing the utilization of the hotline as well as potential barriers.⁵⁰ Using a form to capture demographics, nature of caller questions, staff responses, and resources utilized during the calls, the group was able to assess hotline utilization for the first year of the hotline implementation.⁵⁰ Surveys of hotline users and non-users at study sites were completed at multiple time points to obtain feedback, identify barriers to usage, limitations, and future expansion opportunities.⁵⁰ During its first year, the hotline received 296 calls from 79 healthcare providers, with surveys indicating that the hotline was helpful in improving quality of care.⁵⁰ Those that used the hotline noted that poor cell phone coverage, slow staff response times, and access to alternate information acted as barriers and impacted their hotline use.⁵⁰ The *Uliza!* group found that those who had not used the hotline indicated they did not know about the hotline availability, did not have a cell phone/coverage, did not have questions that required assistance, and had access to alternate resources.⁵⁰ Most calls to the hotline came from study sites with high patient volume and more experience with HIV

services.⁵⁰ Despite barriers identified by users and nonusers, the hotline evaluation by Karari et al. suggest potential utility for a telephone based consultation service for resource-limited areas, which can have an important role in increasing healthcare provider access to relevant patient care information.⁵⁰

Within the scope of genetics, current national telephone resources are available from Genetic Alliance, a non-profit advocacy organization focusing on individuals and families impacted by genetic conditions⁵⁴; the Genetics and Rare Disease Information Center (GARD), a National Institutes of Health sponsored program focused on providing information about genetic conditions⁵⁵; and the National Organization for Rare Disorders (NORD), an organization focused on supporting those impacted by rare disorders, as well as organizations dedicated to such individuals and families⁵⁶. These groups all have publicly available phone lines available for individuals to call and get information about specific genetic conditions or support resources, without providing medical advice.^{54–56} The availability of such hotlines suggest the potential value of phone lines specific to genetics-related questions and concerns; however, there is little information available about the utility of a phone line dedicated to connecting individuals with genetic services, rather than disease-specific information.

2.4 Regional Genetics Networks

2.4.1 History

In an effort to work towards improving the health of children and families through the intersect of genetics, newborn screening, and public health across the United States, the Maternal

and Child Health Bureau (MCHB) of HRSA, Genetic Services Branch (GSB) provided funding for the establishment of a National Coordinating Center (NCC) and seven Genetics and Newborn Screening Regional Collaborative Groups (RCs) in the United States and United States territories in 2004.^{57 58} These RCs were established in order to meet a need for the availability of a centralized group of experts to assist states in the diagnostic and follow-up protocols for state-coordinated newborn screening programs.⁵⁹ The ultimate goal of the RCs was "to enhance and support the genetics and newborn screening capacity of the States" through training and educational strategies and assisting in improving the infrastructure of newborn screening and follow-up services and improve access to genetics services and expertise.⁶⁰

In 2017 the RCs were renamed Regional Genetics Networks (RGNs)⁶¹, and the goals of these RGNs broadened with the most recent funding cycle (June 2017-May 2020), with RGNs tasked with the following:

- 1) By May 2018, each RGN will provide services to at least 250 individuals or families within the geographic area served by the RGN
- 2) By May 2020, each RGN will provide services to at least 1,500 individuals within the geographic area served by the RGN.
- *3)* By May 2020, increase by 20 percent the number of medically underserved patients served by each RGN.
- 4) By May 2020, increase by 20 percent the percentage of clinical sites that use telehealth/telemedicine to provide genetic services.
- 5) By May 2020, increase by 20 percent the number of medically underserved patients receiving genetic services through telemedicine visits.
- 6) By May 2020, increase by 20 percent the number of primary care providers using RGN resources.⁶²

These RGNs continue to be coordinated by the NCC, which functions to support the RGNs through infrastructure development and evaluation, quality improvement efforts, financing and technical assistance, and engagement of RGNs and partners.⁶¹ In partnership with ACMG, the NCC and RGNs collaboratively develop educational resources and work towards improving access to genetics services for medically underserved populations.⁶³

Under the goals outlined by the NCC, the various RGNs across the United States have developed a variety of resources and projects to work toward increasing access to genetic services. For example, the Western States RGN (WSRGN) has developed projects to support telegenetics implementation and training for healthcare providers, established a mentorship program for minority GCs, created resources related to state newborn screening, and funded outreach clinics in underserved populations in Alaska, Hawaii, and Guam.⁶⁴ The New England Regional Genetics Network (NERGN) has developed a variety of patient and provider educational resources, as well as a web-based services directory for those who visit their website to find genetic services, telemedicine locations, family organizations, and public health programs.⁶⁵ The Southeast Regional Genetics Network (SERN) has a variety of resources available, and a similar web-based directory.⁶⁶ The NCC website also hosts a resource repository, in which the NCC, all RGNs, and other partners can submit and publicly share resources developed for a variety of purposes and audiences, including fact sheets for patients and healthcare providers, educational guides, and webinars and educational modules.⁶⁷

2.4.2 NYMAC Regional Genetics Network

One of the seven national RGNs, the New York-Mid-Atlantic Consortium (NYMAC) Regional Genetics Network (Figure 1) serves Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia.⁶⁸ NYMAC is based out of the Wadsworth Center, New York State Department of Health.⁶⁹ NYMAC focuses on a variety of projects, with a unifying goal of improving access to genetic services, specifically for medically underserved populations.⁷⁰ NYMAC also aims to improve access by increasing the number of individuals served overall by the NYMAC RGN, increasing the number of PCPs utilizing NYMAC resources, and increasing telemedicine use in clinical sites and in medically underserved populations.⁷⁰ NYMAC also works closely with family-led organizations to support their efforts and act as a resource. These organizations include state specific Parent-to-Parent (P2P) groups,⁷¹ as well as Family-to-Family (F2F) or Family Voices (FV) affiliates,⁷² family-led groups in which one parent or family provides information, resources, and emotional support to families of and children with special healthcare needs.



Figure 1. NYMAC Regional Genetics Network Logo

Many of the individuals served by the NYMAC RGN live within a HRSA-designated MUA/P.⁴ A regional needs assessment showed that many individuals in the NYMAC region live below the poverty level, with those individuals from five of the region's states falling below the national average poverty level. Many individuals in the NYAMC region also live in rural areas, living in areas with limited healthcare resources and/or living a further distance from specialized medical care, which provides an additional barrier to obtaining necessary genetic services within the region (NYMAC, unpublished data, 2015).

Further regional barriers were identified in a 2017 study completed by NYMAC and the National Center for Hearing Assessment and Management (NCHAM) that surveyed 266 PCPs across the NYMAC region to assess knowledge and use of genetic services in the context of children who are deaf or hard of hearing.⁷³ Findings of this study were consistent with similar studies of provider perceptions of and barriers to genetics services^{41,42}, with 43% of respondents

indicating lack of knowledge and 39% of respondents indicating logistics of referring to genetics as barriers to genetic services for their patients.⁷³

To address the barriers to genetic services identified in both published and internal studies, NYMAC implemented the a Genetic Services Referral Phone Line in 2018 to assist patients and providers in facilitating genetics appointments or referrals.⁷⁴ In conjunction with ACMG, NYMAC staff compiled a clinical services directory and developed a clinical service location map.⁷⁵ The directory, created by NYMAC to be used internally, and later incorporated into a national ACMG directory (https://clinics.acmg.net/), includes information about each clinic site's location and genetic services.⁷⁵ Trained phone line staff use these resources to provide phone line callers with relevant nearby clinic information based on the caller's location and needs.⁷⁵

By assisting callers in identifying the nearest genetic services location to them, including telemedicine options, NYMAC hopes to work towards increased access to genetic services in the region.⁷⁴ In addition to the Genetic Services Referral Phone Line implementation efforts, NYMAC has a variety of workgroups addressing healthcare access and insurance coverage, newborn screening, primary care education, and public health assessments.⁷⁶ In order to continue working towards improved access through provider education, NYMAC's website provides resources for healthcare providers, include summary documents outlining guidelines and recommendations for making genetics referrals.⁷⁷

There is currently limited information available about the success of efforts to connect medically underserved populations with relevant genetic services. Development of a genetic services referral phone line resource in the NYMAC region has the potential to increase awareness of and connect populations in need with relevant genetic services. Implementation of this phone line and evaluation of the utilization can provide information for quality improvement of this effort within the NYMAC Region and similar future efforts aimed at increasing access to genetic services. Evaluation of phone line implementation can also provide insight as to types of resources which may or may not effectively reach medically underserved populations or healthcare providers, and how to adapt or develop interventions and resources for future use.

3.0 Manuscript

3.1 Background

3.1.1 Access to Healthcare and Genetic Services

In a current healthcare climate where disparities and barriers to relevant healthcare services exist,^{5,6} a public health focus on strategies for increasing access to healthcare services for underserved populations is crucial. According to the U.S. Health Resources & Services Administration (HRSA), Medically Underserved Areas and Populations (MUA/Ps) are designated as areas or populations which lack adequate primary care services, with high percentages of the population aged 65 or older and high levels of poverty and infant mortality.⁴ This type of designation may correlate with National Rural Health Association (NRHA) findings that individuals in rural areas have decreased access to physicians, lower socioeconomic status, and on average, a larger elderly population,⁷ suggesting that a focus on increasing access should be given to these populations.

Workforce shortages in healthcare related fields, in part, may contribute to barriers to access of healthcare services. Assessments of rural healthcare systems show the extent of such shortages, with 100% of rural hospital CEOS in the New England to Virginia region citing an overall physician shortage, with the majority citing shortages in at least two primary care specialties.⁸ However, workforce shortages are not limited to primary care. A 2018 workforce study of the genetic counseling profession has suggested that the shortage of genetic counselors (GCs) working in direct patient care may not resolve until the year 2030,⁹ with a 2003 workforce

analysis of clinical geneticists suggesting an insufficient supply of medical geneticists in 17 states across the U.S, with some states averaging 2.0 or less geneticist per 1 million people.¹⁰ In addition to shortages of GCs and other genetics professionals, multiple studies have found that additional barriers exist regarding access to genetic services for underserved and rural populations, such as racial and ethnic disparities^{12,13}, logistical barriers (i.e. distance to genetics service location, referral or insurance coordination, etc.) and knowledge barriers (i.e. lack of awareness of availability of genetic services, lack of knowledge of what constitutes an appropriate referral, etc.).^{1,2,14–18}

Initial efforts to address workforce shortages and barriers in hopes of increasing access to genetic services involve adoption of alternative services delivery models, including group genetic counseling, telegenetics, and genetic counseling assistants or extenders.^{3,19–21} Telegenetics services have been found to be successful in reaching individuals who may not have otherwise been able to access genetic services, and telegenetics program adopters have reported increases in the number of patients served, and from a wider geographic reach.²²

3.1.2 Genetics and Primary Care

With an increasing demand for genetic services and a shortage of genetics healthcare providers, integration of genetics into primary care is becoming increasingly prevalent.²⁷ As healthcare providers who are responsible for providing and coordinating a variety of healthcare services for their patients, primary care providers (PCPs) play a role in facilitating relevant genetic care for their patients.^{24,27} Prior studies have surveyed or interviewed PCPs regarding their perception of genetic services and the role of genetics in primary care. PCPs have acknowledged

their responsibility to identify patients in need of genetic services, and place appropriate referrals, while also raising concerns about their own genetics knowledge.^{27,35,40}

In regard to medical education for PCPs in training, genetics is being increasingly included in curriculum development; however, no standardized genetics coursework is currently being implemented across all institutions.^{28,31} Professional groups including The National Coalition for Health Professional Education in Genetics (NCHPEG) and Association of Professors of Human and Medical Genetics (APHMG) have worked to establish competencies that can be used to address potential gaps in healthcare provider genetics education; however, PCPs and other healthcare providers must recognize the potential limitations of their knowledge, while having and understanding of the implications of genetic assessment and knowing when and how to refer patients appropriately to genetic services.^{33,34} Assessments of genetics integration in medical education suggest that PCPs identify weaknesses in their genetics education, citing difficulties finding relevance of genetics in daily practice, and associating genetics with rare or very specific health conditions.^{36,37}

PCPs often agree that their role is to assist in coordinating referrals to genetics; however, in addition to lack of knowledge, PCPs raise multiple concerns about integrating genetics into primary care practice, including being unable to keep up with current developments in the field; not having enough knowledge about available genetic testing options and their utility and/or implications for patient management; inability to recognize patients who may benefit from a genetics referral; and lack of relationships or awareness of relevant genetics healthcare providers in the area in which they practice. ^{40–42,44} While identifying PCP concerns and perceptions of genetics and associated barriers to access, multiple studies have also prompted suggestions from PCPs about resources which would make for a smoother integration of genetics into primary care

practice, including referral and risk assessment guidelines, information specific to genetic testing and associated benefits, a go-to individual or resource available for questions related to genetics care for patients, and a telephone hotline to provider information.^{35,36,40,41}

3.1.3 Telephone Hotlines

Telephone call centers or hotlines have long been used as resources to connect individuals with services in a variety of settings. Hotlines managed by groups such as the Rape, Abuse and Incest National Network (RAINN) and the Department of Veterans Affairs have been implemented with the goal of providing support and advice to individuals who may lack nearby in-person services or may be part of a demographic less likely to pursue in-persons services.^{48,49}

Hotlines have been designed and implemented to target individuals, patients, consumers, as well as healthcare providers. Following implementation of hotline services, evaluation of utilization, limitations, and areas for improvement can be fundamental to ensuring that such telephone lines are meeting their goals and providing intended services or resources. Hotline evaluation projects can be important in recognizing the need for modifications to effectively reach target groups, with long-term and follow-up utilization studies providing valuable information about strategies which may or may not effectively increase hotline utilization by a target demographic.^{51–53} While efforts to provide increased access and resources to target demographics may not be as effective as intended, lessons can be learned from these types of studies. For example, one study of an under-utilized provider resource hotline in Africa was able to evaluate call data and survey those who did or did not utilize their phone line to identify barriers, limitations, and future expansion opportunities.⁵⁰

3.1.4 Regional Genetics Networks and NYMAC

As part of efforts to improve the health of children and families in the United States by connecting genetics, newborn screening, and public health, the Maternal and Child Health Bureau (MCHB) of HRSA, Genetic Services Branch (GSB) provides funding for a National Coordinating Center (NCC) and seven Regional Genetics Networks (RGNs).^{57,58} In partnership with the American College of Medical Genetics and Genomics (ACMG), the NCC and RGNs collaboratively develop educational resources, and work towards improving access to genetics services for MUA/Ps.⁶³ In particular, a 2015 needs assessment conducted jointly by ACMG and the NCC identified barriers associated with access to genetics services, including distance, lack of available genetics service providers, and lack of awareness.¹¹

One of the seven national RGNs, the New York-Mid-Atlantic Consortium (NYMAC) Regional Genetics Network serves Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia.⁶⁸ NYMAC focuses on a variety of projects, with a unifying goal of improving access to genetic services, in particular for MUA/Ps in the region.⁷⁰ In order to address previously identified barriers to genetic services, NYMAC implemented the Genetic Services Referral Phone Line in 2019. Targeted towards both individuals and healthcare providers, trained NYMAC staff utilize a regional clinical services directory to provide phone line callers with referral and contact information for genetic services near the caller's location, including telemedicine sites.⁷⁴

To date, phone line utilization has been much less than anticipated, despite efforts to increase awareness of the phone line through marketing campaigns. With limited information of the success of prior efforts to increase access to genetic services for MUA/Ps, this assessment serves to detail the implementation process for the NYMAC Genetic Services Referral Phone Line

and evaluate phone line utilization, as well as strengths and limitations of implementation efforts, through evaluation of website and call data, marketing campaign reach, and healthcare provider perceptions of barriers to genetic services. This quality improvement assessment will provide baseline information regarding the effectiveness of a hotline aimed at assisting callers with locating appropriate genetic services and act as a guide for future NYMAC efforts to increase access to genetic services for MUA/Ps, and potentially future efforts of other RGNs.

3.1.5 NYMAC Genetic Services Referral Phone Line Implementation

3.1.5.1 Needs Assessment

Following a regional needs assessment, NYMAC identified areas in which support resources could be integrated in order to address barriers to access to genetic services. Areas which could be addressed included increased awareness and resources for PCPs, increased education regarding available telegenetics services and assistance in implementation of telegenetics services across the region, and increased outreach to inform and connect individuals residing in the region with available genetic services (NYMAC, unpublished data, 2015). In order to increase access to genetic services for individuals within the region, particularly MUA/Ps, NYMAC developed and implemented the Genetic Services Referral Phone Line in 2018. The phone line was designed to be used as resource for both patients and healthcare providers, with established goals of assisting callers in identifying relevant genetic services or family-led organizations in their area, increasing the amount of patients from MUA/Ps who are appropriately referred to genetic services, and improving overall access to genetic services in the region. The NYMAC Genetic Services Referral Phone Line implementation process and goals are outlined in Figure 2.

			ral Phone Line Imple in medically unders			
INPUTS	OUT	PUTS		OUTCOMES		
What we invest	Activities What we do	Participants Who we reach	Short Learning	Medium Action	Long Conditions	
Funding (HRSA) Time Staff: Project manager(s)/coordinat ors, GCs, Pitt graduate student workers, physicians, NYMAC steering committee Marketing assistance (OpAd media, MMS) Phone line technology and support (Verizon inContact)	Implement Genetic Services Referral Phone line Build NYMAC regional clinical services directory Utilize marketing campaigns (mail, social media, email) Raise awareness (presentations, publications)	Patients/Consumers Healthcare providers Family-led Organizations/Groups	Compile NYMAC region clinical genetics services directory Implement NYMAC Genetic Services Referral Phone Line Increase awareness of NYMAC/Phone Line	Act as a resource for patients and healthcare providers Contribute regional clinical genetics services directory data to ACMG for national database Increase amount of patients from medically underserved areas who are appropriately referred to genetic services	Improve access to relevant genetic services	
ASSUMPTIONS			ENVIRONMENT			
 Providers have knowledge of when to refer to genetics Genetics is a relevant concern for patients and healthcare providers Individuals are willing to call phone line/receive information via telephone Patients/Consumers respond to social media advertising 				Social determinants of healt Healthcare/practice structur Phone line staffing availabilit	e	

Figure 2. NYMAC Phone Line Implementation Logic Model

3.1.5.2 Clinical Genetic Services Directory

NYMAC, under the coordination of the NCC and ACMG, maintains a regional clinical genetic services directory. Originally maintained internally via Google Maps, the directory was manually updated by staff members when new clinic information was made available to NYAMC. Upon the announcement that ACMG and the NCC would be working with the RGNs to update a national clinical genetics service directory hosted on the ACMG website, NYMAC staff contacted genetic services locations in the region via telephone and/or email in order to obtain updates to current directory listings. These update efforts were made in order to compile the most up to date

clinical genetic service location information in the internal directory, which would then be used by ACMG and the NCC to update the online directory.⁷⁵

When phone line implementation began in August 2018, an email announcement was sent to a current NYMAC regional email list, including healthcare providers, family-led organizations, and other partners, and post cards were sent to healthcare provider offices in the NYMAC region. This advertising served a dual purpose, introducing the phone line to individuals and healthcare providers in the region, and also leading individuals affiliated with genetic services locations to contact NYMAC in order to have their clinic added to the directory or to ensure their clinic directory listing was up to date.

In December 2019, the NYMAC regional clinical services directory began integration into a national clinical genetic services directory publicly available via the ACMG website. Visitors to the ACMG directory can search for genetic services nationwide using location-based search fields or clinic name and an interactive map. This directory was not used for locating genetic services during phone line implementation; however, following completion and official launch of the national directory, the online ACMG directory, rather than the NYMAC regional directory, will be used to locate relevant clinical genetics services for phone line callers. In addition to search functionality, the ACMG directory website page includes online forms which clinic locations can fill out to submit a new clinic for inclusion into the directory. Functionality is being developed so that clinics can also submit edits to their current directory listing on their own. Figure 3 illustrates a timeline of phone line implementation activities, including clinical service directory updates, as well as efforts to increase phone line awareness, which will be further outlined in subsequent sections.

Phone Line Implementation Activity	Date	Phone Line Awareness Activity
Phone line go-live	August 2018	Phone line information sent via email/postcards to providers in NYMAC region
Phone line website URL/redirect updated	September 2018	
NYMAC internal regional clinical services directory complete	January 2019	
Standard phone line hours initiated	January 7, 2019	Digital marketing campaign launched in Maryland
"Contact us" form available online	February 2019	
	March 1, 2019	Digital marketing campaign launched in Delaware
	May 31, 2019	Maryland/Delaware marking campaign ends
	Way 51, 2015	Provider marketing email campaign launched
	June 2019	NYMAC information presentation at regional Family Voices meeting
	August 2019	NYMAC services and phone line informational article published in Exceptional Parent Magazine
NYMAC directory integration into ACMG directory begins	December 2019	
	January 6, 2020	Digital marketing campaign launched in New York
	March 29, 2020	New York marketing campaign ends

Figure 3. Phone Line Implementation Timeline

3.1.5.3 Phone Line Staffing

The phone line is housed within Verizon inContact, a cloud-based virtual contact center which allows for the phone line to be staffed remotely. The phone line was staffed part-time by Master's in Genetic Counseling graduate student workers from the University of Pittsburgh from August 2018 through May 2020. Bi-weekly meetings were held with student workers and members of the NYMAC leadership team to discuss the phone line, utilization, and staff concerns. Standard hours of operation were initiated in January 2019 and published and updated regularly on the NYMAC phone line website. The phone line was staffed two to three days per week, primarily during business hours (Monday through Friday, 8:00am-5:00pm), with occasional availability during early mornings, evenings, and weekends. Phone line staff were available to answer calls between 8-19 hours per week. During the hours that a staff member was unavailable to answer calls, a voicemail system was in place, allowing callers to leave a message.

3.1.5.4 Phone Line Website

The phone line was managed by NYMAC and coordinated through the Wadsworth Center at the New York State Department of Health (NYSDOH). The NYMAC RGN website is housed with the NYSDOH website, and a phone line specific webpage with URL redirect (wadsworth.org/nymac/referral) was added to the site in September 2018. The phone line web page included the phone line contact phone number and hours of operation, as well as a brief description of the purpose of the phone line, what to expect during the call, and why genetic services may be helpful to some individuals. A NYMAC email address for general questions was also published on the website and phone line webpage. In February 2019, a "contact us" form was added to the phone line webpage to allow individuals to contact phone line staff with questions via email directly through the website.

3.1.5.5 Phone Line Protocol

When a caller contacted the NYMAC Genetic Services Referral Phone Line, they were prompted to provide their zip code within an automated phone tree as this information was needed to find the genetic services closest to them and was requested to be collected by HRSA. No additional identifiers or medical information were recorded. After providing this initial information, the caller was connected with an available phone line staff member during staffed hours of operation. If the caller was contacting the phone line to locate genetic services, staff used the caller's zip code to locate relevant genetics service location(s) nearest to the caller, or appropriate telegenetics resources, via the regional NYMAC Clinical Services Directory and map.

Staff provided callers with referral information for relevant genetic service location(s) based on their needs by providing contact information for the relevant location. Staff were also able to provide information on appointment wait times and accepted Medicaid insurance plans. Inquiries were also accepted via email. Individuals who contacted phone line staff via the online "contact us" form were able to provide their contact information, zip code, and questions or concerns via this contact method, and phone line staff responded with requested information by either phone or email, as appropriate.

Of note, the phone line did not provide formal "referrals" to genetic services, in which care would be coordinated and an appointment would be facilitated for a caller; rather, the phone line provided contact information for relevant genetics services based on location and needs for each caller. Following receipt of this information from NYMAC, it was the caller's responsibility to contact a genetic services location or support resource to facilitate services and necessary formal referrals from their healthcare provider. For consistency with phone line nomenclature, throughout this document, usage of the word "referral" in the context of assisting a phone line caller refers to "information about genetic services nearest to them that met their stated needs," rather than a formal referral to a genetic service location.

The staff were trained to locate and provide information regarding relevant genetics services in the NYMAC region and were not trained to answer medical questions or provide advice. All callers with medical questions or questions related to specific genetic conditions were transferred or referred to GARD. Callers looking for additional support services were transferred or referred to the relevant family-led organization for their state. In cases where callers contacted the phone line outside of staffed hours of operation, they were prompted to leave a voicemail message, and asked to provide their name and contact phone number so that a staff member could return their call. This information was not recorded in any way and was only used for the purposes of returning the initial phone call. A phone line staff member addressed all voicemail messages or email inquiries within 48 business hours.

3.1.5.6 Marketing Campaigns

Following initial emails and postcards sent in 2018, NYMAC used an external marketing consultant, OpAD Media, to launch multiple advertising campaigns to increase awareness of the Genetic Services Referral Phone Line amongst individuals residing within HRSA-designated MUA/Ps. Based on prior NYMAC efforts to engage with PCPs in the state of Maryland, Maryland was selected as a pilot state for a digital marketing campaign. On January 7, 2019, online digital advertisements (Appendix B) were launched in nine targeted counties the state of Maryland designated as a MUA/P by HRSA. The marketing campaign was expanded to two counties in the state of Delaware designated as MUA/Ps on March 1, 2019 (Appendix C). The Maryland and Delaware marketing campaigns were active through May 31, 2019. Individuals from these targeted areas who interacted with advertisements on social media websites (Facebook and Instagram) were prompted to call the phone line or linked directly to the NYMAC Genetic Services Referral Phone line webpage. Location-targeted online webpage banner advertisements, or advertisements embedded within webpage margins which allow users to click the advertisement to visit a website, and search engine advertisements (Google and Bing), in which the phone line website appeared as a sponsored search result, were also utilized.

Call trends through 2019 suggested that those who contacted NYMAC via the phone line or email were primarily from the state of New York, despite no active marketing in that state. It was postulated that because the NYMAC website is housed on the NYSDOH website, the service was more likely to gain attention of individuals in New York. Because of this, an additional digital marketing campaign targeting six counties in the state of New York was launched on January 6, 2020, with an end date of March 29, 2020.

Between May 31, 2019 and June 10, 2019, an email marketing campaign targeted towards healthcare providers (Appendix D) was launched in order to increase awareness and of the phone line as a referral resource for healthcare providers. During this time, an email blast was sent three times to healthcare providers practicing in Pediatrics, Internal Medicine, Obstetrics/Gynecology, and Oncology via Medical Marketing Services, Inc., an email marketing service to healthcare professionals.

In order to raise awareness about the Genetic Services Referral Phone Line, NYMAC made a brief informational presentation at a regional Family Voices meeting in June 2019. NYMAC also contributed an article to the August 2019 issue of *Exceptional Parent* magazine in order to highlight the phone line and its services.

3.2 Methods

3.2.1 Ethical Considerations

Per the Institutional Review Board of the University of Pittsburgh, this project does not meet the formal definition of research (IRB#1807003- Appendix A). The Institutional Review Board was contacted as several time points during the project and indicated that approval was not required for any aspect of this project.

3.2.2 Call Data Collection & Analysis

Data was collected during and after all incoming and outgoing phone line calls (Table 1). Actively collected data included caller demographics and information about services sought and was collected from all callers who were willing to provide the information in order to identify the most appropriate genetic services. Passively collected data was collected from callers if it was reported during the call; however, callers were not required to provide this information and staff did not probe for this information. Following a conversation with a caller, call outcome data was also tracked.

	Data Field	Description			
		Entered in by caller prior to being connected with phone			
	Zip code	line staff member			
	Caller type	Individual Provider (Geneticist, PCP, Pediatrician, OB/GYN, Public Health Professional) Other			
Actively Collected	Type of services sought	Prenatal Genetics Pediatric Genetics Adult Genetics Cancer Genetics General Genetics Genetic Testing Other			
Information	Age of individual needing services	Under 18 18-29 30-39 40-49 50-59 60+			
	How caller found out about phone line	Google Facebook Physician(s) NYMAC website Advertisement Other			
Passively Collected	Purpose of services sought	Diagnostic Post-Diagnostic counseling Family History Other			
Information	Insurance type	Public Private Uninsured			
	Clinic(s) referred to	Documentation of which clinic(s) caller was referred to			
Call Outcome Information	Type of service(s) referred to	Genetics (In-person, Telegenetics, Telegenetics Navigation, GARD, Support Group) Not genetics related Other			

Table 1. Phone Line Data Collection Points

3.2.2.1 Call Volume & Phone Line Utilization

NYMAC website analytics were obtained starting from phone line implementation on August 2, 2018 and tracked through February 18, 2020. Phone line contact history and data reports were downloaded directly from the phone line call center online platform, Verizon inContact. Calls that were identified as test calls, NYMAC staff calls, or outgoing staff calls for non-referral related reasons were removed from data analysis. Remaining calls were coded based on if they were handled, pre-queue, or voicemails. Incoming or outbound calls in which a caller spoke with a NYMAC staff member and potentially received information about a genetic service location and/or support resource were considered "handled." All incoming calls were logged within the call center online platform; however, if a caller hung up, or did not navigate through the automated phone tree, and was never connected to a phone line staff member or voicemail system, the call was considered "pre-queue." Calls which resulted in a voicemail message were assessed individually, to determine if the call was ultimately handled. Incoming email inquiries were also assessed individually to determine the nature of the inquiry and if the email inquiry was ultimately handled. Based on the availability of caller zip code data, calls and emails were coded based on whether the caller-provided zip code was designated as a MUA/P by HRSA.

3.2.3 Marketing Campaigns

Following the digital marketing campaign in Maryland and Delaware, and at the mid-point of the New York State marketing campaign, data detailing marketing reach were obtained directly from OpAD Media. Similar data were obtained from Medical Marketing Services, Inc. following the healthcare provider email campaign. Data on marketing impressions, emails delivered, and associated NYMAC phone line calls and webpage clicks were analyzed to determine potential trends in phone line utilization.

3.2.4 Healthcare Provider Perspectives

Invitations to participate in telephone interviews (Appendix E) were sent to 12 PCPs via email. Three were PCPs practicing within the NYMAC region who had previous involvement with NYMAC and nine were PCPs practicing in the Pittsburgh, Pennsylvania area. Interested PCPs contacted the author via email and were informed about NYMAC, the phone line, and interview purposes. Formal consent was not required per the Institutional Review Board at the University of Pittsburgh because this was considered to be a quality improvement project. PCPs were made aware that agreement to participate in the interview implied consent, with the option to decline to answer questions or end the interview at any time.

A semi-structured interview guide (Appendix F) was developed to assess PCPs' experiences with referring patients to genetics, barriers they or their patients may have experienced related to a genetic referrals, and their perceptions of the NYMAC phone line and its implementation process. Interviews were conducted and recorded using Skype for Business and subsequently transcribed verbatim and de-identified by the author. Interview times ranged from approximately 13 minutes to 23 minutes., with an average length of approximately 18 minutes.

Thematic analysis was used to analyze transcribed interviews, utilizing an inductive coding method, in which themes arose directly from the interview data, rather than coding to fit within a predetermined codebook.⁷⁸ Interviews were coded independently in Microsoft Word, with all resulting codes being combined and analyzed together. Analysis was completed at the semantic level, focusing on the explicit statements and meanings present in the interview data.⁷⁸

3.3 Results

3.3.1 Website Traffic

As summarized in Table 2 and Appendix G, NYMAC website analytics were obtained starting from the date of the phone line implementation and tracked during periods of both active marketing campaigns and no marketing campaigns. Analytics from August 1, 2018 through January 6, 2019 (159 days), prior to any phone line marketing campaigns, showed 1,896 views across the entire NYMAC website. The NYMAC phone line specific webpage received a total of 232 views (12.2%), with 83.6% of those being unique views (n=194), an average of 1.2 unique views per day. Following the initial marketing campaign, analytics from January 7, 2019 through May 31, 2019 (145 days) showed 24,123 views across the entire NYMAC website, with the phone line specific webpage receiving a total of 20,638 views (85.6%), with 93.2% of these being unique views (n= 19,233), for an average of 132.6 unique views per day. From June 1, 2019 through January 5, 2020 (219 days), a period during which no marketing campaigns were active, there were 2,427 views across the entire NYMAC website. There were a total of 368 (15.16%) views of the phone line specific webpage, 85.6% of which were unique views (n=315), with an average of 1.4 unique views per day. Analytics from January 6, 2020 through February 18, 2020 (44 days), approximately midway through an active marketing campaign, showed 29,807 total NYMAC website views, and 23,752 phone line web page views (79.7%), 22,069 of which were unique views (93.0%). During this time, the phone line specific webpage received 501.6 unique views per day, on average. All additional website activity can be attributed to other areas of the NYMAC website unrelated to the phone line.

Table 2. NYMAC Website Analytics

Date Range	Active Marketing Campaign?	NYMAC Website Views	NYMAC Website Homepage Views	Phone Line Webpage Views	Unique Phone Line Webpage Views	Average Unique Webpage Views Per Day
		n	n (%)	n (%)	n (%)	n
08/01/2018 - 01/06/2019	No	1,896	540 (28.5)	232 (12.2)	194 (83.6)	1.2
01/07/2019 - 05/31/2019	Yes	24,123	685 (2.8)	20,638 (85.6)	19,233 (93.2)	132.6
06/01/2019 - 01/05/2020	No	2,427	703 (28.97)	368 (15.16)	315 (85.6)	1.4
01/06/2020 - 02/18/2020	Yes	29,807	1,510 (5.1)	23,752 (79.7)	22,069 (92.9)	501.6

3.3.2 Call Volume & Email Contact

Between August 2, 2018 and February 18, 2020, the NYMAC Genetic Services Referral Phone Line received 67 calls from 51 unique callers. Of the 67 calls, 8 (11.9%) callers spoke with a phone line staff member. Voicemails were left by 9 callers, and the remaining 50 calls (74.6%) were classified as "Pre-Queue," meaning the callers did not navigate through the phone tree and were not connected with a staff member or voicemail system. 73.1% (n=49) of calls were received during an active marketing campaign, with 26.9% (n=18) calls received outside of active marketing campaigns. More calls were made from mobile phones (n=38) than were made from landlines (n=29). NYMAC also received 6 inquires via email. The trends in inquiries are illustrated in Figure 4.

NYMAC staff members addressed inquiries from 16 people during this time period (Table 3). 100% of inquiries via phone or email were received during an active phone line marketing campaign. Of note, one caller indicated that she/he had received an email from NYMAC that prompted the call, but it was unclear if this was an email sent during the provider-targeted marketing campaign. 75% (n=12) of inquiries were made via phone, with two callers also contacting NYMAC via email in addition to their telephone call. The majority of inquiries (81.3%)

were made by individuals, or consumers/potential patients. Of those who contacted NYMAC, eight (50%) were referred to an in-person genetics or telegenetics service and one individual (6.3%) was referred to a relevant support resource. The most common reasons that the phone line was utilized included family history (18.8%), direct-to-consumer/ancestry testing information (18.8%), post-diagnostic counseling (12.5%), and diagnostic services (12.5%).

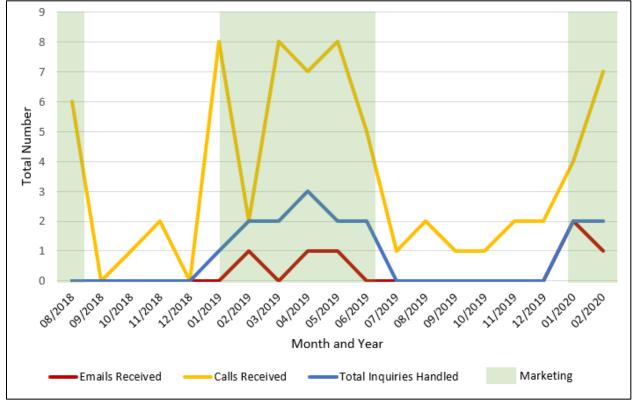


Figure 4. Total NYMAC Inquiries and Inquiries Handled

Table 3. NYMAC Inquiries Handled

Contact Type	MUA/P?	Caller Type	Type of Service Sought	Patient Age	How Caller Found Phone Line	Purpose of Service Sought	Type of Service Referred to	Call During Marketing Campaign?
Incoming Call	Yes	Individual	Pediatric Genetics	N/A	Other- Family Group	Post- diagnostic Counseling	N/A	Yes- MD
Email	No	Individual	Adult Genetics	30-39	N/A	Diagnostic	Genetics- In- Person	Yes- MD
Incoming Call	N/A	Other	Other	N/A	N/A	N/A	N/A	Yes- MD
Incoming Call	N/A	Individual	Other	N/A	N/A	Other- DTC	N/A	Yes- MD/DE
Incoming Call	Yes	Individual	Pediatric Genetics	Under 18	Other- Family Group	Family History	Genetics- In- Person	Yes- MD/DE
Email	Yes	Individual	N/A	N/A	N/A	N/A	N/A	Yes- MD/DE
Incoming Call	Yes	Individual	Other	N/A	N/A	Other- DTC	N/A	Yes- MD/DE
Outgoing Call (Voicemail follow-up)	No	Individual	Pediatric Genetics	Under 18	N/A	Post- diagnostic Counseling	Genetics- In- Person; Telegenetics	Yes- MD/DE
Incoming Call	Yes	Individual	Other	N/A	N/A	Other- DTC	N/A	Yes- MD/DE
Email	Yes	Individual	N/A	N/A	N/A	N/A	N/A	Yes- MD/DE
Incoming Call	Yes	Individual	Adult Genetics	60+	Email	Family History	Genetics- Telegenetics	Yes*
Incoming Call	No	Individual	Adult Genetics	N/A	Advertisement	Family History	Genetics- In- Person; Telegenetics	Yes- Provider Email
Email and Outgoing Call (Voicemail follow-up)	No	Other – Specialty Provider	General Genetics	40-49	Google	N/A	Genetics- In- Person	Yes- NY
Email and Outgoing Call (Voicemail follow-up)	No	Individual	Adult Genetics	40-49	Google	N/A	Genetics- In- Person	Yes- NY
Email	Yes	N/A	Genetic Testing	18-29	N/A	N/A	Genetics- Support Group	Yes- NY
Outgoing Call (Voicemail follow-up)	No	Individual	Adult Genetics	40-49	Google	Diagnostic	Genetics- In- Person	Yes- NY

Abbreviations: N/A- Not available; DTC- Direct-to-consumer genetic testing; MD- Maryland; DE- Delaware; NY- New York *caller reported receiving email from NYMAC

Calls were received during all seven days of the week, with 74.6% of calls received on Monday, Tuesday, or Thursday (n=50), which is consistent with the most common days during which staffed phone line hours were available. Approximately one quarter of calls (25.4%) were received on Sunday, Wednesday, Friday, or Saturday (n=17). Calls were received during a variety of times, with a preference for afternoon (after 12:00pm). 41.8% of calls (n=28) were received during morning hours (12:00am – 11:59am), with three calls received prior to 8:00am. 58.2% of calls (n=39) were received during afternoon hours (12:00pm – 11:59pm), with 11 calls received after 5:00pm (Figure 5). Of the 67 total calls received, 29.9% (n=20) were received during phone line hours of operation, with 56.7% (n=38) of calls received outside of hours of operation. Of note, 13.4% (n=9) of calls were received prior to regular phone line hours of operation being established.

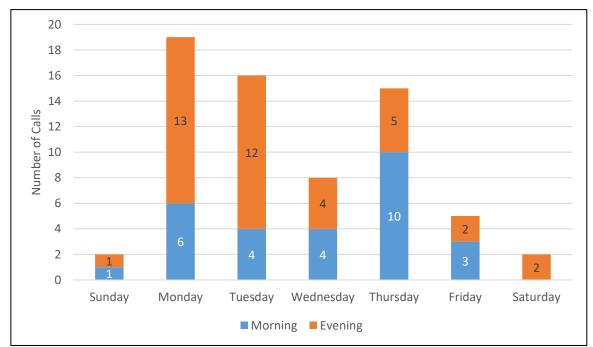


Figure 5. Call Data by Day of the Week and Time of Day

3.3.3 Marketing Campaigns

Between January 7, 2019 and May 31, 2019, social media advertisements in target MUA/Ps in Maryland and Delaware, which prompted those interacting with the advertisement to call the NYMAC Genetic Services Referral Phone Line (also known as "click to call" advertisements), were displayed 1,934,891 times, also called impressions, across Facebook and Instagram, resulting in a reach of 253,400 unique viewers. Social media advertisements that linked to the NYMAC phone line website delivered 5,450,279 impressions and resulted in 27,741 website clicks. Banner advertisements on various websites resulted in 3,967,036 impressions and 5,524 website clicks. Search engine advertisements resulted in 21,165 impressions delivered and 608 website clicks.

Between May 31, 2019 and June 10, 2019 provider-targeted marketing emails were sent out three times and reached over 48,000 PCPs (Table 4). During the first email send-out, 95.9% of emails were successfully delivered to 48,594 PCPs. Delivered messages resulted in 3,334 (6.7%) unique individuals opening the email, generating 62 unique clicks to the NYMAC phone line website. The second and third email send-outs were successfully delivered to 99.9% (n=48,458) and 99.7% (n=44,792) of PCPs, respectively. The second email send-out resulted in 3,387 (7.0%) unique opens and 52 unique website clicks. The third email send-out resulted in 1,523 (3.4%) unique opens and 17 unique website clicks. In total, the provider marketing campaign generated 131 unique clicks to the NYMAC phone line website, which account for approximately 41.6% (131 of 315 total) unique phone line website views during this time period.

Send-out	Number of Emails Sent	Delivered Emails	Unique Opens	Unique Website Clicks	
		n (%)	n (%)	n (%)	
1	50,679	48,594 (95.9)	3,334 (6.7)	62 (0.13)	
2	48,532	48,458 (99.9)	3,387 (7.0)	52 (0.11)	
3	44,908	44,792 (99.7)	1,523 (3.4)	17 (0.04)	

 Table 4. Provider Marketing Email Reach

Mid-marketing campaign data from January 6, 2020 to January 28, 2020 from the ongoing marketing campaign (January 6, 2020 through March 29, 2020) in New York State showed a total of 1,015,871 impressions across social media and internet search engines. Facebook and Instagram advertisements delivered 992,832 impressions and generated 4,655 website clicks. Google and Bing search advertisements delivered 23,039 impressions and generated 639 website clicks. Impressions from both online marketing campaigns are summarized in Table 5.

 Table 5. Marketing Impressions and Website Activity

Composing Data Danga	Social Media (Facebook, Instagram)		Internet Search (Google, Bing)		Web Banner	
Campaign Date Range	Impressions	Website Clicks	Impressions	Website Clicks	Impressions	Website Clicks
		(% impressions)		(% impressions)		(% impressions)
01/07/2019 - 05/31/2019	5,450,279	27,741	21,165	608	3,967,036	5,524
(Maryland, Delaware)	5,450,279	(0.51)	21,105	(2.87)	3,907,030	(0.14)
01/06/2020 - 03/29/2020	992,832	4,655	23,039	639	N/A	N/A
(New York)	992,032	(0.47)	23,039	(2.77)	1N/PA	1N/A
Totals	6 4 4 2 1 1 1	32,396	44 204	1,247	2 067 026	5,524
	6,443,111	(0.50)	44,204	(2.82)	3,967,036	(0.14)

3.3.4 Provider Interviews

Emailed interview invitations had a 33.3% response rate (n=4), and four providers scheduled and completed telephone interviews. One provider had previous experience working with NYMAC (Provider A), and three providers were from the Pittsburgh, Pennsylvania area and were not familiar with NYMAC or the Genetic Services Referral Phone Line (Providers B-D). All four providers identified themselves as family medicine physicians (PCPs) and described the areas

they practice in and at least a subset of the patients they serve as medically underserved and/or of low socioeconomic status, with one provider noting practicing in a rural area and one also describing their practice area as urban. Two providers reported serving a large population of African American patients, and two providers reported a large part of their patient population having health insurance coverage through Medicaid.

All providers interviewed had had some experience referring a patient for genetic services, with the most common indications being prenatal genetic screening and cancer genetics. In discussing the NYMAC phone line, implementation process, and additional barriers to genetic services, common themes and subthemes were identified and grouped into four themes of interest based on the semi-structured interview questions: patient-related barriers to genetic services, provider-related barriers to genetic services, perceptions of the NYMAC phone line and implementation, and additional patient or provider resource suggestions. Thematic analysis is summarized in Table 6.

Themes	Subthemes		
Patient-Related Barriers	Appointment wait times Genetics workforce Patient perceptions		
Provider-Related Barriers	Training/Education Awareness		
NYMAC Phone Line	Utilization Existing resources		
Resource Suggestions	Telemedicine/Telegenetics Electronic health record (EHR) integration		

Table 6. Common Themes in Provdier Interviews

3.3.4.1 Patient-Related Barriers to Genetic Services

When asked about perceived barriers to patients accessing genetic services, two PCPs interviewed identified appointment wait times as an area of concern. One provider stated:

Well access is, um, a problem, delay in having people seen. More for the adult...um, the adults...there's a time delay and people are getting, if they want evaluation for cancer genetics, they're often, um, very much in a hurry to do so. (Provider B)

Another provider shared:

Um, sometimes the amount of time that it takes for the patient to get in. Um, that's probably the biggest barrier I'd guess. Especially with pregnancy because whenever we're trying to do first trimester screening trying it has to be during certain period of time, and if a patient is coming to see me kind of toward the end of her first trimester, then we don't have too much time for that to happen. (Provider C)

The workforce shortage of genetics professionals was also a perceived patient barrier for

two PCPs, with one provider sharing:

I think we're all conscious of overuse or misuse of our specialists' time, and perhaps that's a concern. (Provider B)

The other provider related the genetics workforce shortage to their experience with individuals

without genetics expertise providing genetics services:

The other barrier that's come to...come into place really recently is a lot of unqualified folks that are offering genetic services shouldn't be doing it. Um, for instance, our breast care center, um, has set up a cancer genetics program of sorts that's really managed by a radiologist, who as far as I can tell really doesn't have a background to do it...Um, and yeah they set up a cancer genetics evaluation program, um, again, not really with a medical geneticist or genetic counselors available. Um, so that's the barrier at that end. I mean the barrier um with the heritable diseases is just availability of folks...um...nationwide. And there's a relative shortage... (Provider A)

Patient perception or knowledge of genetics and genetic services was also discussed as a

barrier by two PCPs, with one provider sharing:

And, um, but I think that the biggest problem that I bump into is really trying to get patients to understand kind of why we're doing it, and why...the value, the potential value of it. Um, sometimes the hard part for me has been making that, closing that last loop, which is, I've thought, mentioned it to the patient but how to actually get the patient to the point of that expert. Um, so that's, um, so I think that's been kind of one of the challenges that I've had. (Provider B) Another provider felt that examples of reasons for genetics referrals could be helpful information

for patients to have if they are considering seeking genetic services. The provider stated:

... I think a lot of times consumers in general aren't sure if there's somebody who would qualify for genetic testing or like kind of what sort of genetic disorders we would be looking at. Um, so I think a lot of people think of, um, you know, like pretty rare things, but I don't think a lot of people think of like, oh breast cancer or you know, colon cancer, you know, I mean, any of the more common cancers that could really be a reason for referral for genetics... (Provider C)

This provider also identified location of services, transportation, and cost as potential barriers for patients, though not barriers that the provider had personally experienced in their practice. Another provider felt that competing concerns may impact a patient's likelihood to pursue a genetics consultation:

...Maybe in the patients I see, I don't really think of genetics as being a huge need for them. There's a lot of socioeconomic factors that we work on all the time. And I...maybe I'm also underappreciating uh the role that genetics plays in their lives. (Provider D)

3.3.4.2 Provider-Related Barriers to Genetic Services

In addition to patient-specific barriers, all four PCPs interviewed identified healthcare

provider understanding and/or knowledge as a barrier to patients receiving genetic services. PCPs

noted that healthcare providers may be uncertain as to when a genetics referral is warranted. One

PCP stated:

I guess maybe a barrier that I'm imagining for the sake of your work is, um, perhaps providers don't know exactly what's appropriate. What is a really reasonable genetics referral? (Provider D)

Another PCP felt that having examples of referrals could be helpful to providers, as well as patients, stating:

...I think a lot of genetics counseling is underutilized by a lot of providers. Um, I don't think a lot of physicians know all of the information that we do have available now to be able to um, especially think about screening, moving forward based upon a patient's history, so, that's the same thing. Examples would probably be the easiest way to go about that. (Provider C)

A third provider noted that an increased understanding would help them provide guidance for their

patients, noting:

... The more, the more I guess I, as a physician, understand what happens in those visits, the more I can tell them about what to expect... Um, and so I think part of that is it's like a circle, because the more patients who go, then I get a report back and I talk to them, and I learn sort of what the expectations are and what I can tell patients... (Provider B)

The fourth PCP discussed the importance of education and awareness about resources like the

NYMAC phone line early on in a physician's training, stating:

I'm a big believer, um, when you're trying to promote change um, in getting the, um, information and the process out to people that are in their training phase...Um, so anyway, so either they don't have it burned in, so they're particularly susceptible to something that be, that would help make their life easier. So if they knew about this, um, they would be more likely to use it than somebody who's already got a way of doing things and something comes across their desk that there's this new helpline and, you know, unless they're really struggling, they're not going to take advantage of it. Um, and um, I mean, so that's one it makes their life easier, but the other thing is once you get folks in training hooked, then they're quickly out of training and it's a quick way to spread the word. (Provider A)

3.3.4.3 NYMAC Phone Line

All PCPs interviewed had generally positive perceptions of the NYMAC phone line. All

PCPs expressed that the phone line seemed like a good resource, with one provider noting the

phone line as a particularly helpful resource in the primary care setting:

I like that it is via phone, I think that's a really good idea. Um, I like that it is regional, I think that's cool also. And then, past that, it sounds like it's a 'where

should I send this person?' kind of resource, which in primary care we're always looking for things like that. (Provider D)

Two PCPs interviewed felt that the phone line was something that they would use themselves,

contacting NYMAC to find genetic services for their patient(s); however, they would not

recommend the NYMAC phone line as a resource for patient use. One PCP noted:

For our, from the patients that I take care of, um, I probably wouldn't do that. Their medical literacy and prowess in navigating the healthcare system is...I don't know, I don't know if that would go well. But I guess yeah, if I felt like someone was interested and had the resources to do that, yeah, I would absolutely do that. (Provider D)

The other two PCPs felt that the phone line could be a useful resource for their patients, but they

would not use the phone line as a resource for themselves, with one PCP stating:

Um, I think from my standpoint, um, with being from the Pittsburgh area, I mean I usually refer to the genetics counselors through [health system], um, because that's who I'm affiliated with. So, I think I can see the use of that, the utility of it if I was practicing in an area where I wasn't sure where the resources were...Um, I think from a patient standpoint, it would probably be helpful, because then it kind of puts the onus on the patient to be able to you know, find somebody that is close to them, um, but I think being in an urban area makes it kind of difficult just because we sort of know where the resources are at this point, um, already. (Provider C)

The second provider stated:

I'm a firm believer of the whole medical home concept. I would probably do it...I mean, get the list of resources for them, and vet the resources myself...So I probably wouldn't give the patient the web, the phone number, I would probably use it myself to try to find, um, what I was looking for. (Provider A)

All four providers also noted that having existing relationships with genetics professionals

or knowledge of resources in their area would impact their likelihood to use a resource like the

NYMAC phone line. One provider noted:

...Ah, well first I feel fortunate because I practice where I practice. You know, I'm in Pittsburgh and you know, one of my colleagues is very much into genetics and so I know that there's resources. (Provider B)

A second provider mentioned the same colleague:

Um, working with [physician], I feel extremely spoiled because I think me, and my colleagues, often just say to her directly "hey what do you think about this and where should we send this person?"...She's kind of like the ideal resource. But I know that's not, um, necessarily reproducible everywhere. (Provider D)

3.3.4.4 Resource Suggestions

In discussing the phone line implementation and potential utilization, all four PCPs had suggestions as to what type of resources would be potentially useful in helping them address barriers to genetic services for themselves and their patients. PCPs expressed that the most helpful resources are those that are easily accessible, with the most common suggestions relating to telemedicine/telegenetics as well as the electronic health record (EHR)/electronic medical record (EMR). Regarding the EHR, one provider noted:

Um, as a provider, I can tell you we spend like all of our time with the EMR, and there's like 10 million things to click on in a health record, and I don't know if there's a, if there's a way that you guys could integrate your services into the health record, but I guess that would be...It would be at high risk of just getting lost in all of the other buttons to click on, but...perhaps docs prefer resources coming directly from, directly from Epic or from Cerner and they don't always think about picking up the phone. (Provider D)

Another provider explained an existing EHR tool which allows providers to refer patients for various health counseling services. Thinking about integration of a resource like the NYMAC phone line, the provider stated:

Um, so, having something...not that it would have to be exactly like that, but I guess my point is that having something that was built into the EHR that may be easy for me to locate that counseling alternative has really increased the number of times I've used that service a lot. (Provider B)

In regard to telemedicine, one provider expressed that a telegenetics service resource may be more beneficial in increasing access to services, as opposed to a location service like the NYMAC phone line. When asked about their thoughts on the NYMAC phone line as it stands, the provider stated:

I think, I wish that it was more, almost more telehealth based. So, um, I think it could be more helpful if there were more kind of phone consultation services that could be available for patients instead of just kind of connecting patients to services that are close to them. Um, I think that would be something that could be more beneficial than, um, just the actual ability to get the information about where to go for genetics. (Provider C)

Another provider expressed interest in establishing telegenetics services at their practice location:

We could, you know I could sort of serve as point person for that. I don't feel capable of doing um you know full-service point of contact for most genetics things. But I also think with just a little bit of help, I probably could do some of this basic cancer genetics from my office...Um, but even for local, the local folks for genetics counselors with these fairly straightforward uh family histories and genograms, um, I could be guided in terms of any...you know, I could be guided in terms of basic information to gather when we have a conversation um and get any nuanced additional information um and then you know perhaps advice about the front-end testing. And then post-test have counseling, um you know, probably at this stage. Although, I'm getting better...I'm so involved in genetics, um, you know could be done again by telemedicine or face to face with a genetic counselor. (Provider A)

This provider also suggested using a phone line for provider consultation services, rather than

location-based services like the NYMAC phone line provides:

... Um, not just to ask a simple question for a referral, but a phone line actually where I could do a...somebody sitting there, a genetic counselor or medical geneticist, probably a medical...a genetic counselor at the front end, that could sit there and you know I could schedule, we could schedule a time together for 10-15 minutes to discuss a case, um, so that perhaps the, again, the patient wouldn't have to go there, which is some, you know, it's a distance. It's an hour or better... (Provider A)

3.4 Discussion

3.4.1 Phone Line Utilization

The goal of this project was to evaluate the implementation process and utilization of the NYMAC Genetic Services Referral Phone Line in order to identify potential strengths, limitations, and future directions for NYMAC to focus efforts in order to achieve their goal of increasing access to genetic services in the region. Analysis of the phone line utilization and marketing campaign reach were successfully completed as part of this evaluation project.

Like other phone line utilization studies⁴⁹, analytics from the NYMAC website showed that increased website activity correlated with active marketing campaigns which directed individuals to or linked individuals to the phone line webpage. The phone line webpage received substantially more views during active marketing campaigns, with an average of 132.6 unique views per day during the initial digital marketing campaign (Maryland and Delaware), in comparison to times where no marketing campaign was active, which had an average of approximately 1 view per day. Of note, data analysis was completed for the first half of the most recent digital marketing campaign (New York), and the unique phone line webpage views from January 6, 2020 through February 18, 2020 had already surpassed the total number of unique views during the prior marketing campaign (January 7, 2019 through May 31, 2019), with an average of 501.6 unique views per day. These patterns in website activity suggests that the internet may be a valuable resource to reach target populations, with digital marketing being an effective strategy in driving individuals to visit the website.

Between August 2018 and February 18, 2020, the NYMAC Genetic Services Referral Phone line was underutilized, receiving 67 calls in total. This low call volume was surprising,

given the heightened website activity observed during active marketing campaigns, as well as the findings of NYMAC's regional needs assessment (unpublished data, 2015) and prior studies indicating lack of awareness of genetic services^{1,11,16}; however, this discordance follows trends reported in other phone line evaluation studies.⁵² Also of note, the majority of callers (74.6%) were not connected to a NYMAC staff member or to the phone line voicemail service at the time of their call, which was either due to the caller hanging up and choosing not to navigate through the phone tree, or unknown technical issues with the phone line itself. The fact that the phone line did receive calls during this time frame implies that it is a resource that is perceived as useful by at least some individuals in the region; however, the lack of completed phone calls suggests that some aspect(s) of the phone line protocol continued to act as a barrier for callers. Similar healthcare resource hotlines have also experienced underutilization during initial implementation, but found increased success with an expanded implementation time line as well as geographical reach.^{51,53} The phone line was only marketed to some MUA/Ps within the NYMAC region, and therefore, additional efforts may be needed to implement the phone line on a larger scale regionally before similar uptake and utilization trends are seen.

As illustrated in Table 3, not all telephone and email inquiries were provided with referrals in the form of contact information for a genetic service location(s) or other support resource(s). For the inquiries which came in via email, the most common reason for not making a referral was lack of email response containing information needed to identify relevant genetic services from the original person submitting the email inquiry. For the inquiries made via phone, referral success was limited by what information the caller was hoping to receive from the phone line. Interestingly, three callers contacted the phone line hoping for more information related to ancestry and/or DTC testing. While there are genetic services and resources within the region that are available to these individuals, all three declined a referral. NYMAC had not predicted this area of service need during phone line development and implementation, and the growing interest in DTC testing in today's society,^{41,46} as well as these phone line calls, preliminarily suggest that resource development focused on information about DTC and ancestry testing may be helpful to individuals and healthcare providers in the region.

Phone calls appeared to cluster on days of the week where phone line staff were most available; however, this may be due to phone line hours being listed on website. For example, if the phone line was "open," then someone may have been more likely to call on that day/time. Based on the current call patterns, it appears the addition of weekend and early morning hours would not provide any additional benefit. The addition of more evening hours may increase phone line availability for individuals who prefer to call during available hours and not leave a voice message.

Like the NYMAC website analytics, the reach of the marketing campaigns can be interpreted as somewhat successful, as the digital advertisements led to more NYMAC website and phone line webpage clicks. However, the ratio of website clicks to impressions delivered was approximately 0.5% or less for social media and web banners, and approximately 2.8% for internet search engine advertisements, suggesting that digital marketing provided some success, but may not be the best approach to reach the target population. Email marketing was also largely ineffective, with only 6.7% of healthcare providers opening the email and generating only one (potentially two) call to the phone line. With email increasing as a common method of communication⁷⁹, it may not be the best way to market this type of resource, as there stands to be a risk of it getting deleted without being opened. While incomplete at the time of this project's completion, the most recent marketing campaign targeted towards individuals living in MUA/Ps

in New York State showed similar trends to the prior marketing campaign. At the time of project completion, NYMAC phone line webpage activity continued to increase, and multiple call and email inquiries had been received.

3.4.2 Provider Perspectives

Interviews with PCPs in the NYMAC region provided valuable insight into perceived barriers to access to genetic services in the region, resource needs for PCPs, and provider opinions about the phone line and implementation process. All providers interviewed reported practicing in medically underserved areas or reported that they would consider at least some of their patient population as underserved, making them uniquely situated to weigh in on the state of barriers and access to genetic services for MUA/Ps in the NYMAC region. These interviews identified common themes and PCP perceptions, including those falling under the categories of patient- and provider-related barriers to genetic services, NYMAC phone line, and additional resource suggestions.

Prior studies amongst healthcare providers and patients in rural or underserved areas have found appointment wait times, cost, location/distance to services, and lack of knowledge and awareness as some perceived barriers to patients accessing genetic services.^{1,2,14–18} This project had similar findings, with PCPs having concerns about the time it takes for individuals to get an appointment with a genetic service provider, with one provider attributing this to the workforce shortage of healthcare providers trained in genetics, including genetic counselors.^{9,10} As with other studies,¹³ the PCPs interviewed for this project also felt that the patient populations they serve may have more pressing health concerns, may not have a good understanding of the utility or relevance of a genetics appointment, or may have some uncertainty about what warrants a genetics

appointment. These patient-related barriers may have impacted the utilization of the phone line, should potential patients not appreciate the relevance or necessity of a genetics referral, then they may be unlikely to utilize a resource that can help facilitate such a referral.

Interviews were also helpful in identifying provider-related barriers to facilitating patient access to genetic services. The provider-related barriers are congruent with findings from other studies, and included provider knowledge/understanding of genetics and importance of genetics education early in medical training.^{32,35–37,43} When providers are uncomfortable or unable to identify patients who may benefit from a genetics referral, then they too may be less likely to utilize the phone line as a resource. This suggests the importance of focusing efforts on providing primary care and other healthcare providers with education and resources that can help them properly support and identify individuals who may benefit from a genetics referral.

Regarding the NYMAC phone line and implementation process, all PCPs interviewed reported viewing the phone line as a good resource but had varying ideas of how they would approach phone line utilization. Two of the PCPs interviewed felt like their role as a PCP was to facilitate a genetics referral, and they would not be comfortable directing patients to the NYMAC phone line, but rather, would utilize it themselves to obtain information to provide to patients. Conversely, the other two PCPs felt that the phone line could be a useful resource for their patients and would be comfortable providing patients with information about the phone line if they were interested in pursuing genetic services. While the four PCPs interviewed reported seeing the benefit of utilizing a resource like the phone line for themselves, none had used the NYMAC phone line, and did not feel that they would. The common theme associated with this lack of utilization was existing resources. Similar reasoning was provided by healthcare providers in a study evaluating an underutilized HIV information hotline in Africa⁵⁰, and in a prior qualitative study by

Carroll et al., which found that healthcare providers felt that having a "go-to" person or resource would be helpful to them, and increase their comfort level with personalized medicine.⁴¹ The PCPs interviewed as part of this project interestingly had similar sentiments, citing existing relationships and access to alternate resources and information would provide them with the same benefit as potential phone line utilization. The preference for local and/or existing resources suggests the importance of establishing resources for MUA/Ps, which likely have few existing genetics resources for individuals and healthcare providers living and practicing in these areas.

As with prior studies^{1,22,23}, resources that the PCPs interviewed felt they would utilize to decrease barriers to genetic services included telemedicine, as well as easily accessible resources integrated into EHRs. Two providers also suggested a somewhat expanded phone line scope; rather than providing location-based referral services, they felt it would be helpful if there was a staffed phone line readily available for patients to have a telephone consultation with a genetics provider, or for providers to consult with genetics providers about a case in real time. These suggestions can be valuable in the future, as NYMAC and the larger genetics community continue to explore alternate models to increase access to genetic services¹⁹, and NYMAC continues working towards the goal of increasing access to genetic services in underserved areas/populations in the new grant cycle.

3.4.3 Project Limitations

This project had several limitations. Analysis was limited by the small number of calls and emails received by the NYMAC Genetic Services Referral Phone Line, as well as the availability of complete data from phone and email inquiries received. Because targeted marketing campaigns were launched in only three states within the NYMAC region, the findings of this project may not be generalizable to the entire region, as the genetic services and resources, as well as the proportion of MUA/Ps varies from state to state. In addition, the phone line marketing campaigns relied on location-targeted social media and search engine advertising. This marketing strategy assumes that the target population(s) are active on social media, that genetics is a relevant concern for them, and that they are interested in locating genetic services. Analysis of the most recent marketing campaign in New York State was also limited, as it was not yet completed at the time of data analysis.

The sample size for provider interviews was also small, and a larger sample size may be beneficial for more in-depth thematic analysis. The providers who agreed to participate in qualitative interviews may have been biased, as those invited were either already familiar with NYMAC and the phone line, or practiced in Pittsburgh, Pennsylvania, a city with genetic services available across multiple health systems. In addition, only one PCP interviewed practiced in a state where a phone line marketing campaign had been implemented. Additionally, the perceptions of those providers who did agree to participate in interviews may be inherently different than those of the providers who did not agree to participate in interviews. Also, these perceptions may not be generalizable to PCPs practicing in other states within the NYMAC region.

3.4.4 Future Directions

Based on the overall low utilization of the NYMAC Genetic Services Referral Phone Line, a telephone-based service may not meet the current needs of MUA/Ps in the NYMAC region. At the end of the current funding cycle (May 31, 2020), the NYMAC Genetic Services Referral Phone line will no longer be active. Analysis of phone line utilization and reach of the marketing campaigns following its complete implementation would be helpful in determining the overall impact of the phone line and number individuals assisted in the NYMAC region. After official launch of the completed web-based ACMG genetic services directory, analysis of this service's website activity may also provide additional information for NYMAC, as well as the other RGNs, regarding utilization of an online-based genetic services location resource.

Based on the increased NYMAC website activity during active marketing campaigns, it may be useful for NYMAC to focus some effort to developing online resources for patients and healthcare providers. The current evaluation suggests that digital marketing may be necessary to raise awareness about the website, and it may be beneficial to consider future marketing strategies for NYMAC. Interviews completed with PCPs as part of this project suggest that resources that are easily accessible are preferred, and that online educational resources for both healthcare providers and individuals/consumers could also work towards addressing barriers to genetic services. As the interviews completed as part of this project were a pilot, additional interviews, surveys, or focus groups involving more PCPs, as well as other healthcare providers, individual consumers, or other potential stakeholders could provide valuable information about perceptions of genetic services in the region and resource needs across the region. Continuing efforts to expand telemedicine/telegenetics services in the NYMAC region will also likely be beneficial in helping to increase access to genetic services for populations in need.

3.5 Conclusion

This project was successful in describing the implementation process for the NYMAC Genetic Services Referral Phone Line and assessing phone line utilization from August 2018 through February 2020. Evaluation of phone line use and targeted marketing campaign reach showed an increase in NYMAC website activity and phone line usage during times of active marketing campaigns, with a notable decrease in activity when the phone line was not being marketed, suggesting that advertising to raise awareness of the service was an integral part of phone line implementation. Despite the increased phone line activity surrounding marketing campaigns, phone line utilization remained low, with NYMAC staff handling less than twenty phone and email inquiries over an 18-month time period. The discordance between increased phone line website activity and consistently low call volume suggests that the individuals for whom the phone line was designed to serve may prefer getting their information via other means. Thematic analysis of qualitative PCP interviews also suggested additional healthcare provider-related barriers that may have impacted utilization of the phone line, most notably provider education and understanding of genetics and genetic services.

While this quality improvement assessment project was small in size, and limited to only parts of the NYMAC region, results can be helpful in guiding future NYMAC efforts to address barriers and increase access to genetic services. Although low, utilization of the phone line to date suggests that the Genetic Services Referral Phone Line has the potential to be a helpful resource; however, strategies to reduce other barriers which impact access to genetic services may need to be addressed before such a phone line could function at its full potential. Future studies involving individuals, healthcare providers, and genetics professionals may be warranted to determine appropriate next steps in NYMAC resource development to continue to work towards increased access to genetic services, especially for individuals in the NYMAC region.

4.0 Research Significance to Genetic Counseling and Public Health

This project evaluated the implementation process and utilization of the NYMAC Genetic Services Referral Phone Line, a resource developed with an overarching goal of connecting individuals in medically underserved areas with genetic services. The development and implementation of the phone line, as well as much of the efforts coordinated by NYMAC, the NCC, and the RGNs, are of significance to the fields of genetic counseling and public health.

Maintained and published by the Accreditation Council for Genetic Counseling, the Practice-Based Competencies for Genetic Counselors outlines twenty-two competencies that all genetic counselors must demonstrate.⁸⁰ In addition to having adequate knowledge of genetics, psychosocial counseling skills, and effective educational strategies, all genetic counselors must "[a]dvocate for individuals, families, communities and the genetic counseling profession," with this advocacy role notably including a need to "employ strategies that to increase/promote access to genetic counseling services."⁸⁰

Improving access to genetic services can allow more individuals to benefit from a discussion with a trained genetics professional, which has been shown to lead to increased positive outcomes. Outcomes-based studies, primarily in the cancer genetics setting, focused on the impact of genetic counseling have suggested that individuals who receive genetic counseling often have increased knowledge about genetics and/or genetic testing when compared to individuals who did not receive genetic counseling.⁸¹ With unique training to provide both education and psychosocial support, genetic counseling is associated with more accurate risk perception, increased patient satisfaction, and positive changes in health behaviors, including management recommendation

adherence.⁸¹ The availability of genetic counseling has also been associated with increased patient engagement which results in better health outcomes.⁸²

The findings of this evaluation project may help genetic counselors and other genetics professionals better understand barriers to genetic services, as well as potential interventions to address these barriers. In continuing efforts to develop interventions and resources to engage patients and providers and increase access to genetic services, this project can provide insight as to interventions, such as the phone line, which may not be appropriate for targeting underserved populations, as well as efforts to focus on, such as telegenetics and provider education.

Increasing access to genetic services across the nation is the NCC's overarching goal, and thus, the goal of NYMAC and the other RGNs. With the implementation of the NYMAC Genetic Services Referral Phone Line as a prospective way to connect medically underserved individuals with relevant genetic services, this goal directly ties into the three core functions of public health and essential public health services.⁸³ NYMAC's projects, including the phone line, fulfill all three core functions of public health: assessment, policy development, and assurance.⁸³ NYMAC is continuously working to identify and explore genetic health concerns within the region, as well as working towards solutions for those concerns within the region, encompassing the assessment function of public health. This project, as it relates to understanding barriers related to accessing genetics services, ties into the investigation portion of the essential public health services. In addition, using partnerships with community resources and regional healthcare providers, NYMAC has developed and implemented a variety of resources and interventions, including the phone line, to address barriers and other health-related concerns for individuals living in the region.

NYMAC has also been involved in execution of policies and interventions designed to protect public health, correlating with the policy development function of public health. Most notably, the implementation of the Genetic Services Referral Phone line directly relates to the assurance function of public health. The phone line was designed to link individuals within the region, especially in underserved areas, with genetic services, an essential public health service. This project specifically relates to the public health service of evaluation by assessing the effectiveness of the phone line and other related resources currently available. Through this project, NYMAC has gained insight from utilization evaluation and healthcare provider perceptions that can influence future efforts to address regional barriers to genetic services.

Addressing barriers and increasing access to genetic services for individuals in MUA/Ps can likely not be achieved by one group or addressed with one single intervention. However, through collaborative efforts between groups like NYMAC, the other RGNs and the NCC, as well as genetic counselors, patients, other healthcare providers and stakeholders, resource and intervention development can continue, and these efforts can continue to work towards making genetic services accessible to all patients.

Appendix A University of Pittsburgh IRB Approval



University of Pittsburgh Institutional Review Board

3500 Fifth Avenue Suite 106 Pittsburgh, PA 15213 (412) 383-1480 (412) 383-1508 (fax)

MEMORANDUM

TO: Andrea L. Durst

FROM: Human Research Protection Office (HRPO)

DATE: July 12, 2018

SUBJECT: IRB# 1807003: Establish Infrastructure to Connect Underserved Populations to Genetic Services through a Toll-free Phone Line in New York-Mid Atlantic Consortium (NYMAC) Region

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(d)

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.

Appendix B NYMAC Social Media Marketing Campaign Image Examples



Figure 6. Social Media Advertisement Examples - Link to Website

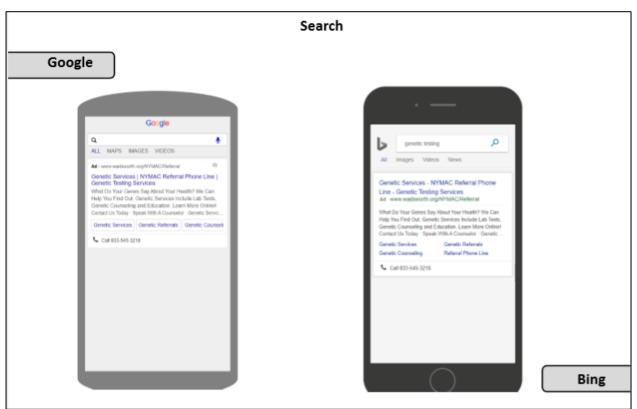


Figure 7. Search Engine Advertisement Examples

Appendix C MUA/Ps Targeted in Initial Market Campaign

State	Counties	Marketing Campaign Dates
Maryland	Anne Arundel Baltimore Baltimore City Caroline Dorchester Somerset Talbot Wicomico Worcester	January 7, 2019 – May 31, 2019
Delaware	Kent Sussex	March 1, 2019 – May 31, 2019

Appendix D NYMAC Provider Email Campaign Message

Subject: [MARKETING] Be your patient's source for genetic information



The NYMAC Regional Genetics Network's goal is to improve access to genetic services in the region, including Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia and West Virginia.

Individuals from our region with genetic conditions and their families, advocates, healthcare professionals and public health professionals collaborate to ensure that individuals with heritable conditions and their families have access to quality care and appropriate genetic expertise.

Genetic Services Referral Phone Line

NYMAC has implemented a Genetic Services Referral Phone Line, which can help connect your patients to the right genetic services, including information about the nearest genetic services provider(s) in the area, clinics offering telegenetics, etc.

The Genetic Services Referral Phone Line is 833-545-3218. Phone line hours and more information are on the Genetic Services Referral Phone Line website.

Benefits of Phone Line for Genetic Services

Speaking with a genetics provider can help if your patients have questions or need information about:

- Inherited diseases
- Prenatal counseling
- · Testing for inherited conditions
- Steps to take, such as screenings, if your patients are at risk for hereditary cancers

Please do not hesitate to contact us at nymac@health.ny.gov for more information.

> 120 New Scotland Avenue Albany, New York 12208

Appendix E Provider Interview Email Invitation

Dear Dr. [name],

I hope this email finds you well.

My name is Alyson Evans, and I am a current Master's in Genetic Counseling student at the University of Pittsburgh. I am also a graduate student worker for the NYMAC Regional Genetics Network, which includes the state of [state]. I am writing today with an invitation to participate in a telephone interview regarding the NYMAC Genetic Services Referral Phone Line.

In January 2019, NYMAC launched a Genetic Services Referral Phone Line following a regional needs assessment of potential ways to increase access to genetics services to medically underserved populations. The phone line is open to individuals and healthcare providers. Using a regional clinical genetics service directory, NYMAC Genetic Services Referral Phone Line staff are trained to assist callers with locating relevant genetic services in their area. The phone line does not provide medical advice, but staff are also able to refer callers with medical questions or additional support needs to appropriate resources.

Despite targeted marketing to both individuals and healthcare providers over the past year, the phone line has been greatly under-utilized. As part of my role as a NYMAC graduate student worker and in partial fulfillment of my Master's in Genetic Counseling thesis, I am hoping to complete qualitative interviews about healthcare providers' perspectives about the phone line and implementation process.

If you would be willing to participate in a brief interview (approximately 30 minutes), your opinion would be greatly valued and appreciated. Per the Institutional Review Board at the University of Pittsburgh, these interviews are designated as quality improvement and IRB review was not required. Interviews will be conducted via Skype for Business audio and recorded for later analysis. Email addresses will not be saved, and interview transcripts will be de-identified prior to analysis.

If you are interested in participating in an interview, please email me at <u>aee31@pitt.edu</u>. Please feel free to contact me with any questions or concerns.

Best,

Alyson Evans MS Genetic Counseling Condidate, University of Pittsburgh, Class of 2020 Graduate Student Worker, NYMAC Regional Genetics Network

Project Advisors: Andrea Durst, MS, DrPH, LCGC Steering Committee Chair, NYMAC Regional Genetics Network Associate Director, Genetic Counseling Program, University of Pittsburgh

Beth Vagel, MS, CGC Project Manager, NYIMAC Regional Genetics Network

Appendix F Semi-Structured Interview Questions

- Can you tell me what type of healthcare provider you are and what practice area or specialty you practice in?
- How would you describe the area/patients that you serve?
 - Would you say that you practice in a medically underserved area?
- Have you referred patients to genetic services?
 - Would you say this is something you do routinely?
 - What sort of patients do you refer to genetics?
 - Can you tell me about any difficulties or barriers you've experienced in making a genetics referral?
 - What type of resources do you think would be helpful to address these barriers?
 - Can you identify any additional barriers to your patients getting genetic services?
- Were you familiar with the NYMAC Genetic Services Referral Phone Line prior to being contacted for this interview?
 - If so, how did you come to hear about it?
- Have you, or would you utilize the phone line as a resource for yourself? For example, you call us to find a genetic service for a patient.
 - Why or why not?
 - If no, what do you think would be a better resource for you?
- Have you, or would you utilize the phone line as a resource for your patients? For example, you provide information about the phone line to a patient to call us to find genetic services themselves.
 - Why or why not?
 - If no, what do you think would be a better resource for your patients?
- What are your thoughts about the phone line model as it stands currently?
- Have you had a chance to visit the NYMAC/Phone Line website?
 - If so, do you feel like it provides helpful information?
 - If no, what would you like to see included on this website?
- Do you have any additional thoughts on potential ways to address barriers to genetic services?

Appendix G NYMAC Website Analytics

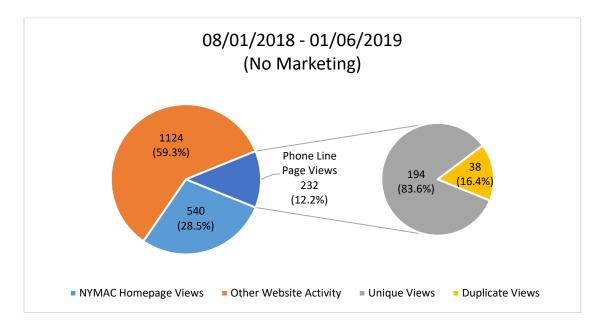


Figure 8. NYMAC Website Analytics 08/01/2018 - 01/06/2019

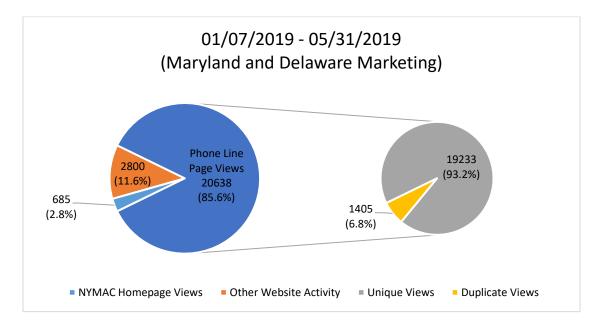


Figure 9. NYMAC Website Analytics 01/07/2019 - 05/31/2019

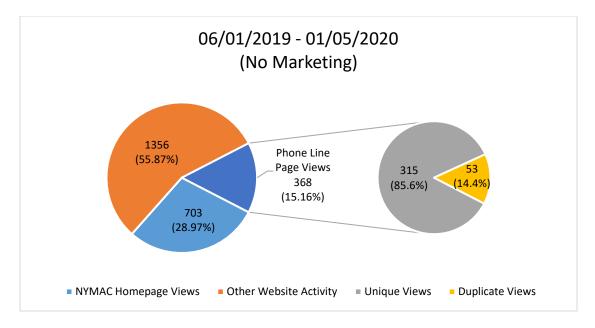


Figure 10. NYMAC Website Analytics 06/01/2019 - 01/05/2020

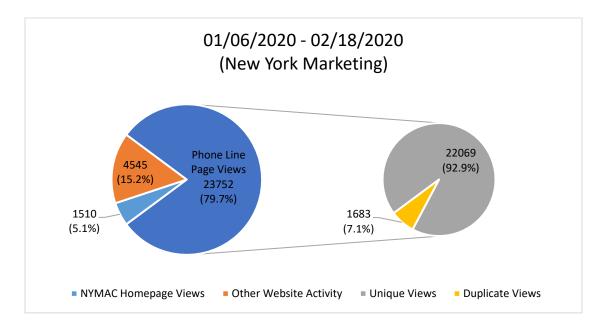


Figure 11. NYMAC Website Analytics 01/06/2020 - 02/18/2020

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