NC Public Health Genetics and Genomics Plan

Introduction

Genetics and genomics are areas of medicine that involve the study of our genetic information, including gene function and interaction, heredity, and how altered genes may cause disease. Scientific advances in human genetics and genomics, combined with an expansive growth in knowledge and skills, have increased our ability to understand the contributions of genetics and genomics to the health of individuals and the public.

As the application of discoveries in genetics and genomics continues to evolve, it will become important for providers to intermittently reassess their potential use to improve health. To that end, an updated 2020 North Carolina Public Health Genetics and Genomics Plan was developed through a public and private partnership. The plan includes goals and objectives for the next five years (2020-2025) to be accomplished in partnership with families, patients, health care providers, researchers, and other stakeholders. This Plan will also foster ongoing conversation and collaboration across the North Carolina Division of Public Health (NC DPH) and with other state, regional and local community partners.

History

North Carolina has been a leader in understanding the important role of genetics and genomics for the health of our individual residents and communities. North Carolina began screening newborns for phenylketonuria (PKU) in 1966. However, the NC DPH recognized early on that the need for genetic testing and services is more extensive than newborn screening. The Genetic Health Care Program was created within NC DPH in 1970, where a social worker provided genetic education and outreach to North Carolina residents throughout the state. In 1976, a team of Regional Genetic Counselors was created through funding from the National Genetic Disease Act, with a focus to educate providers and patients and to improve access to genetic services through satellite clinics. At that time, North Carolina was one of only a few states to implement such an expanded genetics network for providing patient care.

As public health genetic services in North Carolina increased, there was growing interest in evaluating the impact of each of those services on the health of individuals within the state. In 2001, NC DPH applied for and received a grant from the Maternal and Child Health Bureau to conduct a needs assessment of the NC genetic programs and services. The Task Force on Genomics and Public Health was created in 2003, which involved a collaboration of stakeholders from government, academia, medicine, industry, non-profit organizations, and the public.

After 18 months, the 2004 North Carolina Public Health Genomic Plan was created, with broad goals related to communication and education, genetic testing and services, and epidemiology and surveillance. The 2004 Plan was intended to be a starting point for public health leaders and policymakers. It was hoped that the 2004 Plan would lay the foundation for a comprehensive strategy for integrating genomics into public health practice in North Carolina.

The Planning Process

The 2004 North Carolina Public Health Genomic Plan was a bold and collaborative effort but was difficult to implement due to lack of designated funding. As a result, there was no process in place to execute the Plan's recommendations, despite their importance and relevance in public health. However, the need for providing appropriate genetics and genomics services to NC citizens persisted and grew with increasing genomic and genetic knowledge and technologies. In response, management within the Children and Youth Branch brought together state public health agencies, public and private academia, industry, families, medicine and community genetic and genomic stakeholders for further discussion about updating the North Carolina Public Health Genomic Plan. In May 2018, an initial meeting was held, with over 40 stakeholders present to discuss the state of genetics and genomics services across the lifespan of North Carolinians, and to receive updates from state programs in birth defects, genetics and newborn screening, the sickle cell syndrome program and individual institutions that provide genetics services. There was a consensus reached that the 2004 North Carolina Public Health Genomic Plan needed an update. The participants at the meeting set a goal of providing a draft to Branch management within 18 months. A Leadership Team was formed with the following members:

- A clinical geneticist (current president of NCMGA) - *Co-chair*
- A Branch Family Partner - *Co-chair*
- The State Public Health Genetic Counselor
- The Branch Pediatric Medical Consultant
- The Genetics and Newborn Screening Unit manager

Several state and national partners were asked to participate in key informant interviews and provide feedback to help with the development of goals, objectives and actions for the plan. This included but was not limited to representatives from the American Public Health Association and Office of Public Health Genomics, Center for Surveillance, Epidemiology and Laboratory Services at the Centers for Disease Control and Prevention. There were five in-person meetings held between September 2018 and May 2019. Over 100 stakeholders were invited to participate at the in-person meetings or through electronic communication over the course of the planning process.

Public Health Genetics and Genomics Advisory Committee

One key recommendation was to create a Genetics and Genomics Advisory Committee of subject matter experts to provide advice and help with problem solving on key topics of interest in genetics and genomics in North Carolina. The committee will be staffed by the Children and Youth Branch on behalf of the Division of Public Health. The committee will consist of 15-18 people, with individuals and their families who are impacted by genetic conditions representing about 25% of the committee. The committee will also include experts recruited from a diverse group of stakeholders in medical practice, education, health systems, advocacy, research, and state level agencies.

There will be the ability to have smaller working groups to engage others on specific topics and issues. The Advisory Committee will meet quarterly, with the first meeting tentatively scheduled for May 2020.***The 2020 North Carolina Public Health Genetics and Genomics Plan***

The 2020 North Carolina Public Health Genetics and Genomics Plan outlines a bold vision for the future of genetic and genomic health. The success for implementation of this plan will require partnerships forged and maintained by the Advisory Committee, as well as securing additional support and funding needed to achieve the recommended goals and objectives outlined below. The ability to make these recommendations possible will not only allow for improved care of all North Carolina residents, but will also place North Carolina at the forefront of genetic and genomic medicine.

The 2020 North Carolina Public Health Genetics and Genomics Plan: Three Priority Areas

There are three priority areas for recommendations in the 2020 North Carolina Public Health Genetics and Genomic Plan: Genetic Services and Testing, Education and Communication, and Epidemiology and Surveillance.

The Genetics and Genomics Advisory Committee will help to monitor progress on the objectives and actions from the 2020 NC Public Health Genetic and Genomic Plan.

The overall goals of the strategic plan are to:

1) Ensure quality, comprehensive and timely access to genetic and genomic services including evaluation, testing, follow up, and social support
2) Enhance the capacity for public health to collect, analyze, link and store genetic and genomic data to monitor health outcomes of individuals and populations in North Carolina.
3) Increase genetic and genomic awareness, knowledge, skills and abilities among health care providers, patients and the general public.

There are tables for each of the three priority areas for recommendations that follow with objectives and actions for each.

Genetic Services and Testing

<table>
<tr><td colspan="2">GOAL: Ensure quality, comprehensive, and timely access to genetic and genomic services, including evaluation, testing, follow up, and social support to citizens throughout North Carolina</td></tr>
<tr><td colspan="2">Objectives Overview
1. Improve access and support for genetic services to patients and families in North Carolina affected with genetic conditions.
2. Increase the ability for all patients to receive necessary and appropriate genetic and genomic testing.</td></tr>
<tr><td>Objective 1: Improve access and support for genetic services to patients and families in North Carolina affected with genetic conditions.</td><td>Action Items: Overview
1. Expand the workforce of genetics providers.
2. Increase telemedicine capabilities.
3. Provide appropriate state-funded genetic counseling, social support, and follow up services.

1. Expand the workforce of genetics providers.

BY DECEMBER 2020
• Assess the shortage of genetics providers (e.g., geneticist, genetic counselor, advanced practitioners, etc) across North Carolina by determining the number of current practicing providers and open positions at all of the centers/institutions/departments providing clinical and public health genetics services
• Identify the demand for the types of genetic evaluations (prenatal counseling, pediatric/adult evaluations, cancer counseling, etc) across all of the centers/institutions/departments
• Support advocacy efforts by additional groups (NCMGA, NCPS, NCAFP, NCMS, etc) for genetic counselor licensure legislation, expanded clinical training sites for genetic counseling students, and retaining graduates of North Carolina training programs to continue their clinical practice in North Carolina.

BY DECEMBER 2021
• Determine the most appropriate provider(s) to fill the areas of highest demand and target recruitment efforts
• Provide information via various platforms on careers in genetics to high schools, colleges/universities, medical schools, and advanced practitioner training programs
• Initiate partnerships with primary care providers to address appropriate referrals</td></tr>
</table>

	BY DECEMBER 2023 • Increase the number of practicing genetics providers in North Carolina increased by at least 20% • Survey centers/institutions/departments providing clinical and public health genetics services regarding number of practicing providers and open positions • Conduct regular monitoring of access to genetic services, and reduce wait times for an appointment by at least 20% **2. Increase telemedicine capabilities.** BY DECEMBER 2020 • Work with the NCMGA and other stakeholders to determine areas/locations of need for improving access to genetic services (screening to counseling), genetics practitioners currently providing telemedicine services, future interest in providing telemedicine services across the various centers/institutions/departments, and resources currently available to assist with developing telegenetic programs. BY DECEMBER 2021 • Identify a person or develop a position for coordinating telegenetics services across the state BY DECEMBER 2022 • Expand telemedicine services across North Carolina by at least 20%, with education to local providers that telemedicine services are available. BY DECEMBER 2024 • Evaluate access (e.g., decreased distances traveled, decreased wait time for appointments) and outcomes (e.g., diagnoses and appropriate referrals made sooner) in regions with increased telemedicine services vs those done in-person. • Determine if local education for telemedicine services increases referrals to genetic services providers.

	3. Provide appropriate state-funded genetic counseling, center/institution/department grants, social support to families/patients affected with a genetic condition and follow up services. BY DECEMBER 2020 • Identify at least 3 families who have a relative affected with a genetic condition and would be willing to share their story to highlight the need for genetic counseling, social support, and follow up services. • Examine other state-funded social support programs (e.g., sickle cell syndrome) and their funding sources • Identify sustainable funding sources and create target amount necessary for state-funded genetic counselor, center/institution/department grants, social support, and follow up services • Determine need and scope of work of state-funded genetic counselor positions BY DECEMBER 2022 • Initiate program funding streams and allocate resources (amounts to be determined based on volume and complexity of patients and services provided) • Hire appropriate number of genetic counselors based on needs identified previously • Implement programs for social support and follow up services BY DECEMBER 2024 • Evaluate effectiveness of implemented programs by measuring family/patient satisfaction with support services, reduced hospitalizations, number of families impacted, etc. • Evaluate utilization of state-funded genetic counselors by local health departments, scope of patient contact, reduction in wait times or time to diagnosis
Objective 2: Increase the ability for all patients to receive necessary and appropriate genetic and genomic testing.	**Action Items Overview:** 1. Advocate for improved coverage and reimbursement by public and private insurers for genetic and genomic testing. 2. Partner with state-wide societies, advisory/industry groups, and families to determine shared goals around needs for genetic and genomic testing.

	1. Advocate for improved coverage and reimbursement by public and private insurers for genetic and genomic testing. BY DECEMBER 2020 • Assess for evidence-based genetic testing recommendations that are currently not being covered by insurers • Evaluate needs for genetic testing by surveying genetics providers regarding barriers to accessing testing for their patients • Obtain at least 3 family stories about how genetic and genomic testing (or lack thereof) has impacted their lives • Support the need for expanded genetic testing coverage by Medicaid, Medicare, and private insurers (NCMGA to submit RFP to Medicaid for exome sequencing with goal for this to be covered by 12/2020) BY DECEMBER 2022 • Determine additional/new genetic tests that are not being covered, and consider adding requests for coverage of evidence-based genetic testing **2. Partner with state-wide societies, advisory/industry groups, and families to determine shared goals around needs for genetic and genomic testing.** BY DECEMBER 2020 • Assess genetic testing needs of appropriate stakeholders, such as North Carolina Pediatric Society, North Carolina Academy of Family Physicians, North Carolina Medical Society, North Carolina Advisory Council on Rare Diseases, North Carolina Biotech, North Carolina Child, AHECs, County Public Health Departments, etc BY DECEMBER 2022 • Develop various educational materials to distribute to appropriate stakeholders regarding needs identified

Epidemiology and Surveillance

GOAL: Enhance the capacity to collect, analyze, link and store genetic and genomic data that allows for tracking health outcomes of individuals and populations in North Carolina.

Objectives Overview

1. **Improve collection, analysis and storage of genetic and genomic-related data for public health programs**
2. **Increase the ability of genetic and genomic-related data to assess impacts on health and social factors of individuals, families and communities.**
3. **Increase surveillance around awareness and use of genetic counseling, testing and other services**

Objective 1: Improve collection, analysis and storage of genetic and genomic-related data for public health programs

Action Items Overview:

1. Expand use of genetic and genomic data.
2. Improve the efficiency and appropriateness of data abstraction for public health purposes.
3. Improve storage of public health genetics and genomic data

1. Expand use of genetic and genomic data.

BY DECEMBER 2020

- Assess the need and capacity for the staff and data systems in public health programs (i.e., newborn screening, North Carolina Birth Defects Monitoring Program, Cancer Registry, and Communicable Disease) to use genetic and genomic-related data

BY DECEMBER 2021

- Evaluate ability for improved access to data from hospital and practice EMR and state HIE (Health Information Exchange) by public health programs
- Determine the demand from academic and community partners for public health data for genetic and genomic research
- Determine the capacity of public health systems to meet the demand for data for research
- Explore capacity for formal partnership between state public health and academic centers for student and other workforce experiences in applied epidemiology (or other public health areas) in other public health branches beyond the Communicable Disease Branch

	BY DECEMBER 2022 • Explore the need for appropriate legislation, rule, policy and/or state-funded support for enhancing genetic and genomic related data and staff for increased public health surveillance • Explore ability to standardize data use agreement processes and templates • Develop additional agreements or make changes in current agreements that facilitate appropriate access to data and sharing between public health and other agencies • Explore hospital EMR system tools or coding to improve maternal and family histories available from inpatient hospital records. If feasible, pilot a formal partnership between a state public health program (i.e., State Lab or North Carolina Birth Defects Monitoring Program) and academic centers for experiences in applied epidemiology related to genetics BY DECEMBER 2024 • Evaluate/survey use of genetic and genomic data by public health programs • Evaluate implementation of rules, policies or processes to support enhanced surveillance • Standardize a place in the EMR to be able to access complete historical information • Evaluate applied epidemiology experience in genetics, if pilot done • Evaluate effectiveness of legislation, rule, policy and funding to increase access to and use of genetic and genomic data
	2. Improve the efficiency and appropriateness of data abstraction for public health purposes. BY DECEMBER 2020 • Assess the current workforce of professionals who perform data abstraction • Assess rules and legislation in place related to data abstraction for public health programs • Survey current data abstractors and programs about challenges and needs in the workforce BY DECEMBER 2022 • Pilot the broader use of an existing or new standardized location in hospital EMRs to access complete historical information on patients (i.e, family health history) • Assess public health data use agreements and processes in place

	BY DECEMBER 2024 • Determine need to partner with DHHS, academic centers, and other partners to advocate for changes in rule, legislation or policy that support data abstraction • Work with North Carolina Birth Defects Monitoring Program, North Carolina Health Care Association and hospital systems to increase data abstraction for public health purposes
	3. Improve storage of public health genetic and genomic data BY DECEMBER 2020 • Work with the State Lab and newborn screening advisory committee to explore options in rule or policy for a permanent home for newborn screening results (in child's medical record vs. birth certificate add on BY DECEMBER 2022 • Work with the newborn screening advisory committee to explore options in rules or policy for a permanent home for tandem mass spectrometry data outside of STAR LIMS BY DECEMBER 2024 • Implement policy or rule about use of permanent location for newborn screening results • Implement policy or rule about use of permanent location for tandem mass spectrometry data

Objective 2: Increase the ability of genetic and genomic-related data to assess impacts on health and social factors of individuals, families and communities.	**Action Items Overview:** 1. Partner with public and private agencies to support increasing access to follow up data for genetic conditions and molecular genomics in public health. 2. Increase data sources about family and social impact used in public health **1. Partner with public and private agencies to support increasing access to follow up data for genetic conditions and molecular genomics in public health.** BY DECEMBER 2021 • Evaluate gaps in current access to data sources (HIE, EMR) for public health programs (i.e., newborn screening, NC BDMP, cancer) that could be used for follow-up of certain genetic conditions • Explore the ability for data access and mining through several academic centers using Electronic Medical Research Search Engine (EMERSE) with Carolina Translational Science Association (CTSA) with North Carolina Health Care Association • Evaluate data, funding and staff strategies used by other states that support short, middle and long term follow up in newborn screening and BDMP's for specific genetic conditions or birth defects. BY DECEMBER 2022 • Determine which conditions or defects and systems would be appropriate to use to pilot for data collection for follow up and apply for funding or resources if needed • Determine the processes needed to expand agreements or policies to expand access to needed data systems (i.e., HIE, EMR) for follow up • Determine appropriate policies and processes for sharing of data from specimens from state lab with partners for academic (research) and public health purposes related to reportable diseases in partnership with DPH legal and other stakeholders BY DECEMBER 2024 • Pilot use of one of the data systems to track health and social outcomes of a specific birth defect or genetic condition.

	• Establish appropriate policies and processes for data sharing of lab specimens used in follow up. **2. Increase data sources about family and social impact used in public health** BY DECEMBER 2021 • Assess the family impact work being done as part of registries such as Cancer, Rhett's Syndrome, Duchenne's Muscular Dystrophy, Fragile X, etc. and with Early Check and All of Us. • Explore use of the SCHS Social Determinants of Health Regional Needs Data (food, housing, transportation, etc.) • Assess/research current privacy and legal, social and ethical issues related to accessing and applying genetic and genomic data • Work with Branch Family Partners and academic centers to determine families who would be interested in sharing family stories. BY DECEMBER 2022 • Determine roles for public health with efforts that collect qualitative and quantitative data from patients such as All of Us and Early Check • Determine which conditions or defects and systems would be appropriate to use to pilot and that would include qualitative family (and social) data collection as part of follow-up. BY DECEMBER 2024 • Pilot use of one of the data systems to track health and social outcomes of a specific birth defect or genetic condition. • Review qualitative data and determine if can be used for reports, advocacy or other purposes.

Objective 3: Increase surveillance around awareness and use of genetic counseling, testing and other services	**Action Items Overview:** 1. Use existing efforts to ask about patient and public awareness related to use of genetic counseling, testing, and other services. **1. Use existing efforts to ask about patient and public awareness related to use of genetic counseling, testing, and other services**. BY DECEMBER 2020 • WCH Section staff will use the MCH Block Grant Needs Assessment to assess awareness and use of genetic risk assessment, counseling and genetic testing/services. • Explore one question being used or added to surveys (i.e., Child Health Assessment Monitoring Program Survey (CHAMPS), Pregnancy Risk Assessment Monitoring Survey (PRAMS) or Behavioral Risk Factor Surveillance Survey (BRFSS) about family history, screening test done or offered, cancer type, use or coverage of recommended genetic testing and counseling. BY DECEMBER 2021 • Secure funding to pay for addition of a question to one state survey about genetics or genomics • Evaluate qualitative results from MCH Needs Assessment and determine how they can be incorporated as a strategy in the MCH Block Grant BY DECEMBER 2024 • Assess baseline data on survey questions and if there is any change after two or three years

Education and Communication

<table>
<tr><td colspan="2">Goal: increase genetic and genomic awareness and competency among providers and members of the general public through education to make informed decisions in response to ongoing and increasing knowledge in genetics and genomics for the improvement of public health.</td></tr>
<tr><td colspan="2">Objectives Overview
1. Increase genetic and genomic education in training programs of health care providers.
2. Increase professional genetic and genomic continuing education for health care providers.
3. Increase awareness of and utilization of the integral role of genetics and genomics in the health of patients and the general public.</td></tr>
<tr><td>Objective 1: Increase genetic and genomic education in training programs of health care providers.</td><td>Action Items Overview:
1. Increase genetic and genomic education in training programs of health care providers.
2. Develop suggestions to assist with meeting any identified needs related to genetic and genomic education within current programs.

1. Increase genetic and genomic education in training programs of health care providers.

BY JUNE 2020
• Identify current training programs for healthcare providers in North Carolina including but not limited to MDs, DOs, PAs, NPs and RNs.

BY DECEMBER 2021
• Develop surveys to send out to identified health care training programs regarding inclusion of curriculum with respect to genetics and genomics education. This will help determine educational strengths as well as weaknesses that benefit at the very least from bringing training/curriculum and workforce standards up to date with the core genetics and genomics competencies as put forth by the National Coalition for Health Professional Education in Genetics (NCHPEG) and Centers for Disease Control and Prevention (CDC).]

BY DECEMBER 2022
• Send out surveys to respective training programs and analyze returning results to identify the status of the educational needs.</td></tr>
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	2. Develop suggestions to assist with meeting any identified needs related to genetic and genomic education within current programs. BY DECEMBER 2023 • Determine training program needs based on survey results and develop individual training program suggestions based on the core genetics and genomics competencies as put forth by NCHPEG and CDC. BY DECEMBER 2024 • Engage training programs regarding available educational resources to supplement genetic and genomics educational practice based on the suggestions set forth from 2023.
Objective 2: Increase professional genetic and genomic continuing education for health care providers.	**Action Items Overview:** 1. Assess current practices in continuing genetics and genomics professional education for health care practitioners. 2. Promote professional genetic and genomic continuing education for health care practitioners with health care state organizations. **1. Assess current practices in continuing genetics and genomics professional education for health care practitioners.** BY DECEMBER 2020 • Identify existing continuing genetic and genomic education requirements for practitioners. BY DECEMBER 2021 • Identify practitioner specialties to survey for continuing education. This list will include perhaps 4 or 5 specialties selected by engaging the North Carolina Medical Society for suggestions on specialties to survey. BY DECEMBER 2022 • Analyze returned surveys to look for strengths and weaknesses as they pertain to the specialty in questions with regards to the core competencies as set forth by NCHPEG and CDC.

	BY DECEMBER 2023 • Provide resources (evidence based and obtained from CDC, NIH and other government genetic and genomic resources) to include case scenarios based on survey responses to increase genomic and genetic utilization within, and further personalize and improve patient care. BY DECEMBER 202 • Resend and analyze surveys to initial specialty groups to help determine any increased uptake of genetic and genomic integration into patient care
Objective 3: Increase awareness of and utilization of the integral role of genetics and genomics in the health of patients and the general public.	**Action Items: Overview:** 1. Promote the use of evidence based pertinent genetic and genomic information and knowledge by the general public. **2. Promote the use of evidence based pertinent genetic and genomic information and knowledge by the general public.** BY DECEMBER 2020 • Create a DHHS genetic and genomic website presence designed for patients and families geared toward genetics and genomics. Include benefit of Family Health History tools and provide website tools from CDC and NIH to encourage this utilization. BY DECEMBER 2021 • Add patient stories to DHHS genetic and genomic website. Enlist families from Family Partners. BY DECEMBER 2022 • Include information and any latest guidelines on current use of direct to consumer genetic and genomic testing resources BY DECEMBER 2023 • Enlist and have in place genetic counselors at community health fairs and other events.