

NCC/RC SYSTEM 2012-13 EVALUATION REPORT

NCC Evaluation Workgroup

*First Report of the
2012-2015 Funding
Period*



TABLE OF CONTENTS

Acknowledgements	3
Executive Summary	5
Introduction	8
Background	10
Methods	12
Evaluation Framework	12
Common Evaluation Measures and Protocols	13
Data Collection and Analysis	16
RC Common Measures Findings	17
Collaborations	17
Public Information	20
Education and Training Resources	22
Newborn Screening Capacity Building	25
Access to Care—Medical Home	26
Survey on Access to Care for Individuals with Genetic Conditions	27
Survey on Medical Home and Transition	32
Discussion	40
Challenges and Limitations	41
Future Directions	43
Supplement 1: RC Highlights	44
<u>List of Tables</u>	
1.Common Measures for Year 1 Measurement	14
2.Evaluation Terms, Framework, and Measurement Protocols	15
3. Counts of People and Organizations that the RCs Work With	17
4. Use of Websites and Social Media	21
5. Education and Training Sessions	22
6. State/Territorial Adoption of CCHD and SCID RUSP	25
<u>List of Figures</u>	
Figure 1. Map of 7 RCs and NCC	8
Figure 2. Counts of People and Organizations by Type	18
Figure 3. Education and Training Activities by Participant Type (%)	23
Figure 4. Education and Training Activities by Participant Type (N)	24
Figure 5. Survey Responses by Regional Collaborative	27
Figure 6. Genetic Conditions Identified	28
Figure 7. Age When Genetic Condition Was First Identified	29
Figure 8. Insurance Coverage Meets Needs	29
Figure 9. Personal Doctor or Nurse	30
Figure 10. Communication with the Family	32
Figure 11. Communication with Other Providers	33
Figure 12. Care coordination with Families	34
Figure 13. Care Coordination Support	35
Figure 14. Care coordination with Other Providers	36
Figure 15. Transition Preparation	37



Appendices

- A. NCC Organization Structure
- B. NC/RC Project Director and Program Manager Roster
- C. NCC Evaluation Workgroup Roster
- D. Roster of the Members of the Technical Workgroup
- E. Three-tier ASPIRE Model of Evaluation
- F. NCC Logic Model
- G. RC Activities Mapped to Healthy People 2020 Objectives (November 2012)
- H. RC Activities Mapped to HRSA RC Priorities (November 2012)
- I. Working Together
 - Survey Instrument
 - Data
- J. HRSA Performance Measure Form #41
- K. NGEEN Survey
 - Survey Instrument
 - Data
- L. ACMG Survey
 - Survey Instrument
 - Data



ACKNOWLEDGMENTS

On behalf of the National Coordinating Center (NCC) for the seven Regional Genetic Service Collaboratives (RCs), I am pleased to share the results of the first year evaluation. This work reflects considerable effort and represents the collective work of many dedicated individuals.

The RC Project Directors/Project Managers played a key role in guiding the development a set of common evaluation measures. The Evaluation Workgroup established the framework for this measurement effort and the means for collecting the data. A federal interagency Technical Advisory Workgroup offered additional assistance in this first year's effort. The names and affiliations of these contributors are found in the Appendices.

Several staff should be recognized for their leadership to this effort. These include Alisha Keehn, the NCC Project Manager, Dr. Barry Thompson, the ACMG Medical Director, Sophie Stich, the NCC Project Coordinator, and Deborah Maiese, the NCC Evaluation Consultant. Through the Genetic Alliance, which partners with us on the newly established National Genetics Education and Consumer Network (NGECN), I want to recognize Sharon Terry, James O'Leary, and Sharon Romelczyk for their work to engage consumers.

Through the NCC Workgroups we tapped the expertise of volunteers across the country who played an important role in identifying what is important to measure and existing tools to perform the measurement. In particular, I want to thank all of the people who participated in the Medical Home and the Transition Workgroups.

I also acknowledge with gratitude the guidance that has been provided by the staff at the Health Resources and Services Administration (HRSA) Maternal and Child Health Bureau, Genetic Services Division, Bonnie Strickland, Division Director, and Jill Shuger, the NCC/RC Project Officer.

I believe that this first-year evaluation document fulfills the guidance set forth in the HRSA RFA. These data provide a baseline from which we will measure our progress over the coming years.



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EXECUTIVE SUMMARY

The mission of the seven Regional Genetics Service Collaboratives (RCs) and their National Coordinating Center (NCC) is to strengthen and support the genetic and newborn screening capacity of the states and to improve the availability, accessibility, and quality of genetic services and resources for individuals with, or at risk for, genetic conditions and their families. (Mission Statement adopted January 2013) To fulfill this mission, the NCC/RCs engage in education and training for health and public health professionals and information dissemination for consumers of genetic services, the public, and policymakers. The purpose of this capacity-building cooperative agreement and the seven RC grants is to bring together genetics expertise to support health and public health providers to improve the lives of people at risk for or with genetic conditions.

In the first year of the Cooperative Agreement (#U22MC24100), the NCC engaged the RCs in a consensus process to conduct a national evaluation. In accordance with the Health Resources and Services Administration (HRSA) Guidance (12-139), the NCC is to document its level of success in achieving five Healthy People 2020 (HP 2020) objectives. Five HP 2020 objectives were chosen by the NCC/RCs to encompass their activities. These objectives focus on access to care for people with genetic conditions and are found in the following Maternal, Infant, and Child Health (MICH) and Disability and Health (DH) program focus areas:

- MICH-30. Increase the proportion of children, including those with special health care needs, who have access to a medical home.
- MICH-31. Increase the proportion of children with special health care needs who receive their care in family-centered, comprehensive, coordinated systems.
- MICH-32.2. Increase the proportion of screen-positive children who receive follow-up testing within the recommended time period
- MICH 32.3. Increase the proportion of children with a diagnosed condition identified through newborn screening who have an annual assessment of services needed and received.
- DH-5. Increase the proportion of youth with special health care needs whose health care provider has discussed transition planning from pediatric to adult health care.

This report focuses on the national evaluation process, its framework, measures, and findings. Over a seven-month process (October 2012 – April 2013), the NCC/RC Evaluation Workgroup (WG) used the HP 2020 framework to find commonalities across the RCs' activities. The WG developed a set of common evaluation measures for activities where five or more RCs are pursuing similar activities. The WG found existing measurement protocols and the data collection was administered in the summer of 2013. The sources of the protocols include HRSA's National Survey of Children with Special Health Care Needs Survey (NS-CSHCN), the Medical Home Index developed by the Center for Medical Home Improvement, and the HRSA Performance Measure Form #41, Medical Home. In partnership with the RCs, the NCC also collected data from the RCs' constituencies using a Working Together instrument [*Working Together: A Profile of Collaboration* (Chrislip & Larson, 1994)].



At the national level, the NCC asked the genetic service professionals in the ACMG Clinical Services Directory to examine their medical home and transition activities. The National Genetic Education and Consumer Network (NGECN) used the Genetic Alliance's database of more than 700 disease-specific organizations and multiple social media platforms to reach consumers of genetic services. Individuals and families with a child who has a genetic condition were asked questions from the NS-CSHCN. The RCs reported information to the NCC on their activities to improve access to genetic services; their workforce development through training and education sessions, their public information and outreach through the Internet and social media, and their newborn screening activities.

The findings from the first year (June 1, 2012 – May 31, 2013) show that the NCC/RCs are enhancing the health and public health workforce's understanding of genetic conditions; promoting medical homes; addressing the issues of paying for care and the challenges of transitioning from pediatric to adult care. The RCs' training and education sessions reached more than 2,100 providers and consumers. The audiences for these sessions were from public health, genetic service specialists, and other disciplines. The NCC/RCs are important resources for public information with almost 60,000 unique visits to their websites.

The first-year data show that RCs are assisting state public health NBS programs, promoting the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (SDACHDNC) Recommended Uniform Screening Panel (RUSP) with activities focused on critical congenital heart disease (CCHD) and severe combined immunodeficiency (SCID). RCs are also involved with the health and public health community to ensure that positive screens get appropriate follow-up.

From the NGECN survey, we now have data to begin to paint a portrait of health care experiences from individuals with genetic conditions and their families. The survey explores care coordination and transition from pediatric to adult health care - HRSA priorities for the NCC/RC system. From the NCC survey, we have the perspectives of genetic service providers on communications and care coordination with families and with other providers.

Key findings from individuals and families include:

- 13.7% (n=229) reported they were not covered by any health insurance during the past 12 months;
- Of respondents who reported there was a time during the past 12 months that they or their family members needed genetic counseling, 43.4 % (n=246) felt they did not receive all the genetic counseling that was needed and 4.6% did not know.
- The top barriers that prevented individuals from getting the genetic counseling they needed were:
 - Cost (18.9%);
 - Insurance coverage issues (17.1%);
 - Doctor did not know how to treat or provide care (15.1%);
 - Did not know where to go for treatment (9.3%); and
 - Not available in area/transportation problems (8.6%).
- The majority of survey respondents (62.0%, n=953) reported that they consider more than one person to be their (or their child's) personal doctor or nurse.



- 83.1% (n=113) of respondents reported their doctors or other health care providers treat only children. Yet, only 10% of respondents (n=13) said that their providers have talked with them about transition to providers who treat adults.
- Only 30% (n=462) of respondents were ever referred to a support and/or advocacy group by a health care provider.

Key findings from genetic specialists, using self-reported answers to questions from the four-level scoring system of the Medical Home Index where Level 1 is the lowest level, include:

- Communications with families 53% (n=41) were at Levels 3 and 4; while 47% were at the Levels 1-2, suggesting an almost equal division between high and low levels of communications with families.
- Communication with other providers 45% (n=35) were at Level 2, suggesting that most genetics specialists felt that they were communicating at minimal levels with other providers, which complements the results found in the NGEEN survey (see above).
- Care coordination levels with families: nearly 68% (n=52) of the respondents were at Levels 1-2, indicating that room for improvement in communication with families exists.
- Care coordination support of families was at 79% for Levels 2-3 (n=61) which indicates that providers feel that, while periodicity of engagement is variable, they are partners with the family in care coordination goals.
- Care coordination with other providers 57% (n=44) was at Level 2, reflecting that most genetics providers passively wait for communication, and do not initiate it.
- Transition preparation for individuals with heritable conditions, 84% (n=59) reported the Levels 1-2, which indicates that youth are seen without their family member after age 14 (as condition permits) and that medical education of their condition and personal needs is initiated, but not comprehensive.

Data from these surveys will serve as a baseline for assessing needs and informing future efforts of the NCC/RCs.

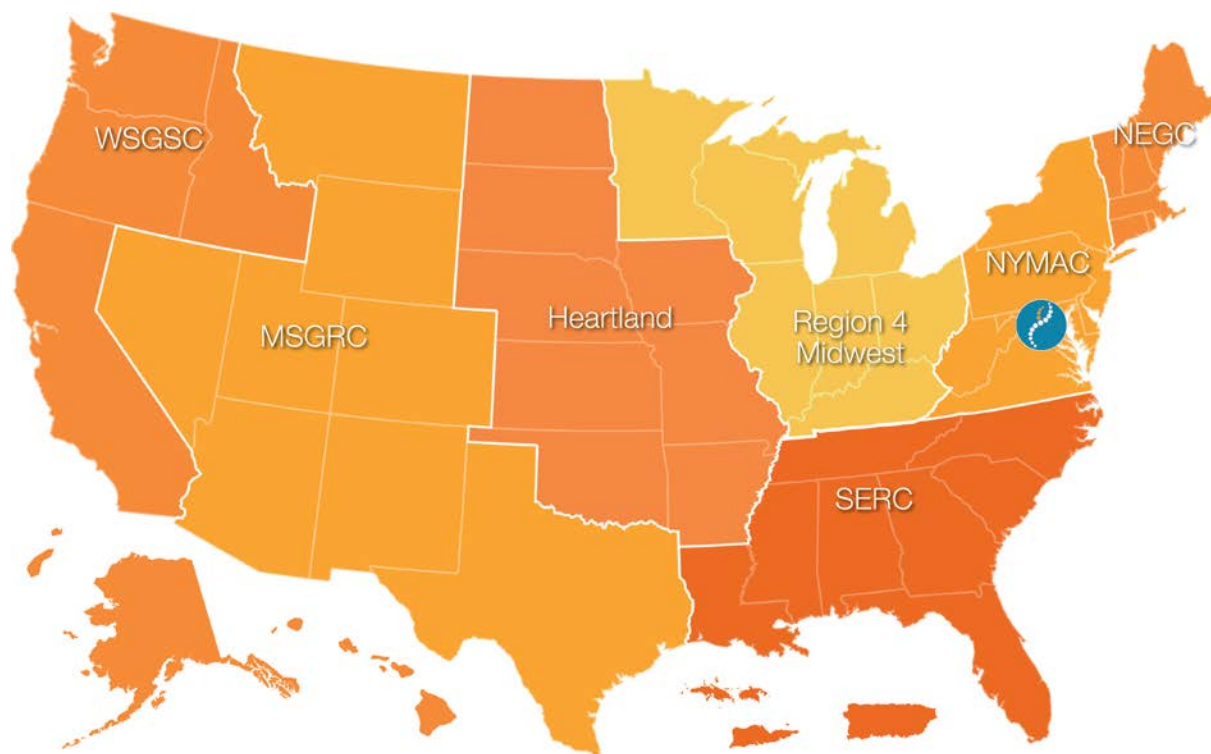
With continued focus on medical home, care coordination and transition from pediatric to adult care, the NCC/RCs are positioned to help HRSA with public outreach and educational activities related to the Patient Protection and Affordable Care Act (ACA). The next year will be focused on opportunities to build the public infrastructure to improve access and to address gaps in services for people who are at-risk of heritable conditions or who live their daily lives with genetic conditions.




INTRODUCTION

In 2012, the Maternal and Child Health Bureau of the Health Resources and Services Administration (MCHB/HRSA), Genetic Services Branch (GSB) awarded grants to seven Regional Genetics Service Collaboratives (RCs) and a cooperative agreement for a National Coordinating Center (NCC). These awards are a part of on-going efforts since 2004 to improve the health of people at risk or living with genetic conditions. While the focus has been on newborns, children and their families, the NCC/RCs are now being asked by HRSA to address populations with genetic conditions over the life course by promoting the translation of genetic medicine into public health and health care services. The NCC is administered by American College of Medical Genetics and Genomics (ACMG) as a partnership with the GSB/MCHB/HRSA and the seven RCs. A roster of the Program Directors/Project Managers (PDs/PMs) is included in Appendix B.

Figure 1. Map of 7 RCs and NCC



 = National Coordinating Center for the seven HRSA Genetics Collaboratives

The NCC structure is comprised of a Principal Investigator, Project Manager, Medical Director, and an Evaluation Consultant. (Appendix A). The NCC includes a National Genetics and Consumer Network (NGECN), administered by the Genetic Alliance. Their mission is consumer engagement. The NCC supports numerous subject matter committees that address ACT Sheets, the Patient Protection and Affordable Care Act (ACA), Family History, Medical Home, Telegenetics and Transition. These committees provide a forum for subject matter experts to share their work and pursue common projects. These committees also contributed to the national evaluation by identifying important topics to measure and the instruments to measure them.



The 2012 HRSA NCC and RC grant guidances (HRSA 12-139 and FOA 12-138) emphasized evaluation. The NCC is to “facilitate integrated data collection and assessment systems using national quality indicators (ex. Healthy People 2020) and data for project evaluation.” The HRSA guidance to the RCs was to “quantitatively and qualitatively evaluate outcomes of projects undertaken by the RCs and the NCC to accomplish these goals. Choose five (5)...measures that are nationally endorsed ...to use for assessment of Regional Collaborative impact...Propose a method for collecting baseline and annual data and means to analyze the changes seen.”

This is the first-year national evaluation progress report for the NCC/RC system for the period June 1, 2012 through May 31, 2013. The report provides a description of the methods used to produce baseline data on a set of common measures with the RCs and on data that has been collected in surveys administered by NGEEN using their network of disease specific advocacy organizations and by the ACMG using their clinical services directory. These data demonstrate that there is a need for NCC/RCs national efforts to improve the health of people at-risk or with genetic conditions.



BACKGROUND

Following the HRSA guidance, evaluation is a prominent component of the current NCC/RC structure. Both the NCC and the RCs engaged evaluators. The NCC contracted for 50% time evaluation consultant; each RC has an evaluator and the RCs' evaluators range in their percent effort. Some have greater roles in their RCs assisting the PD/PM. All contributed to the national evaluation. The NCC convened an Evaluation Workgroup (Appendix A –Structure) with the goal of reaching consensus among the RCs on a set of common evaluation measures to reflect their activities.

The NCC Evaluation Consultant chairs the Evaluation WG with the Region 4 (Midwest) PD serving as the Co-Chair. The WG, comprised of the seven RC evaluators (Appendix C – Roster), first convened in October 2012. The Evaluation WG held monthly teleconference calls and met in-person in November 2012 in conjunction with the PD/PM annual meeting and in March 2013 at the ACMG annual meeting. A majority of the RC PDs/PMs joined in the Evaluation WG calls and the in-person meetings. The HRSA Project Officer participated in these meetings.

A Technical Advisory Workgroup convened in January 2013 (Appendix A - Structure). Comprised of representatives from the Agency for Healthcare Research and Quality (AHRQ), the Centers for Disease Control and Prevention (CDC), HRSA, and the National Institutes of Health (NIH) (Appendix – TA WG Roster), the Technical Advisory Workgroup helped ensure that this evaluation effort was coordinated with other evaluation and measurement activities of the U.S. Department of Health and Human Services (HHS).

The HRSA Genetic Services Branch asked that the NCC/RCs evaluation be guided by three questions:

- How much was done?
- How well was it done?
- Are people with genetic conditions better off because of the NCC/RC system?

This first evaluation report provides answers on how much was accomplished. It provides some insights into how well the NCC/RCs are doing and it lays a foundation for assessing whether people are better off.

The NCC/RCs pursued a three-tier evaluation approach (Appendix E- Three-tier Evaluation Process Pyramid). First is the national effort led by the NCC that includes components that are administered by both the NCC and RCs. Second is the Inter-RC cluster evaluation, where multiple RCs are working on evaluation measures through NCC standing committees, e.g., the Telegenetics Workgroup survey. With 7 RCs participating, the Telegenetics Workgroup reviewed questions that had been used in the Western States RC and Mountain States RC to create a new instrument that was fielded by the NCC to the ACMG clinical services membership database. These cluster projects are considered incubators with measures being tested by four or fewer RCs. When an activity is pursued by 5 or more RCs, the PD/PMs decided that the measure should be adopted as a common national evaluation measure for the NCC/RC system. A third level of evaluation is done by each RC on its HRSA-funded projects. While this report focuses on the top-tier national evaluation process, its framework, measures and findings, we have included an appendix to this report that highlights the individual accomplishments as self-reported by each RC (Supplement 1 – RC Highlights).



The guiding principle for the national evaluation includes putting this work into the context of HP 2020 objectives, MCHB Performance Measures, and the HRSA guidances for the NCC and RCs. Decisions were reached by consensus with the evaluators making proposals to the RC PD/PMs for their approval. The evaluation used existing measurement protocols (to the extent possible) to enable the evaluation findings to be placed in the context of other data and the published literature.

The first-year evaluation activities focused on establishing baseline data. The plans call for these data to be used over the term of the three-year NCC cooperative agreement and the five-year RC grants to document the level of success of the NCC/RCs in achieving the programs' objectives. These data will look at whether the funds expended for the NCC/RCs are having a real and measurable impact and are worth sustaining. Another goal of the national evaluation is to demonstrate the value of a regional approach.



METHODS

Evaluation Framework

ACMG, in their winning proposal to continue as the NCC, set forth a logic model to guide the national evaluation. (Appendix F – Logic Model) Its components include the resources/inputs of the NCC and its workgroups, the RCs, and other HRSA grantees. The activities encompass provider training and education programs; family resources; and collaborations with other HRSA-funded programs and with other national organizations. The initial outputs comprise common definitions of terminology and identification and use of common measurement protocols.

The outcomes envisioned are:

Increased numbers of adult providers who have participated in RC efforts are willing to accept patients with heritable conditions;

Increased numbers of youths have documented care plans/Increased percentages of children with genetic conditions ages 12-17 have doctors who usually/always encourage increasing responsibility for self-care and (when needed) have discussed transitioning to adult health care, changing health care needs, and how to maintain insurance coverage;

Increased numbers of medical homes for people with heritable conditions;

Increased coordination between pediatricians and primary care providers and genetic specialists;

Increased numbers of infants and children with risk assessment counseling have documented referrals; and

Families are satisfied with care and with care transition.

The impact will be:

Improved access to genetic services;

Reduced waiting time for genetic services;

Increases in the proportion of individuals with genetic conditions who have a medical home; and

Improved linkages between specialists and primary care providers to ensure that individuals with genetic conditions have coordinated care

Following this logic model, the NCC's starting point was to measure NCC/RC activities in the context of the national HP 2020 objectives. The initial list showed that RCs had proposed activities in 15 HP 2020 objectives of which five objectives were being pursued by four or more of the RCs. During the course of the first grant year, RCs modified their proposed activities. All



seven RCs began planning for or pursued medical home projects; newborn screening capacity building, and transition from pediatric to adult care activities. (Appendix G – HP2020) Five HP 2020 objectives were chosen by the NCC/RCs to encompass their activities. These objectives focus on access to care for people with genetic conditions and are found in the Maternal, Infant, and Child Health (MICH) and Disability and Health (DH) program focus areas:

- MICH-30. Increase the proportion of children, including those with special health care needs, who have access to a medical home.
- MICH-31. Increase the proportion of children with special health care needs who receive their care in family-centered, comprehensive, coordinated systems.
- MICH-32.2. Increase the proportion of screen-positive children who receive follow-up testing within the recommended time period
- MICH 32.3. Increase the proportion of children with a diagnosed condition identified through newborn screening who have an annual assessment of services needed and received.
- DH-5. Increase the proportion of youth with special health care needs whose health care provider has discussed transition planning from pediatric to adult health care.

The HRSA RC Guidance announcement was used as an additional framework to design the national evaluation. When the RCs' activities are arrayed by these nine HRSA priorities, the diversity of RC planned activities to meet regional needs emerges. (Appendix H--HRSA RC Priorities) The HRSA priorities being worked on by six RCs include collaborations and newborn screening long term follow-up. With the implementation of the ACA, the RCs began to consider working together on health access issues associated with insurance coverage and reimbursement.

Common Evaluation Measures and Protocols

With the evaluation framework established, the Evaluation WG began the process of identifying measures and measurement protocols. Over the course of several meetings and with the input of the PDs/PMs, consensus was reached on the following common measures for the year one evaluation:



Table 1. COMMON MEASURES FOR YEAR 1 MEASUREMENT

HRSA RC Guidance Priorities	Priorities in generic terms	RCs with Year 1 Activity	Measures
1. Treat in the context of a medical home	Access	Heartland MSGRC NEGC NYMAC Region 4 SERC WSGSC	HRSA MCHB Performance Measure #41 – RCs reported activities
4. Build state public health department capacity	Newborn Screening Capacity Building	Heartland MSGRC NEGC NYMAC Region 4 SERC WSGSC	Level of state institutionalization of RUSP, e.g. CCHD and SCID RC activities in support of RUSP
5. Strengthen public-private partnerships	Collaborations	MSGRC NYMAC NEGC Region 4 SERC WSGSC	# of family/disease-specific advocacy groups that RCs reach # of family/disease-specific advocacy groups that RCs work collaboratively with
7. Improve insurance coverage policy and reimbursement	Affordable Care Act Implementation	Region 4, NEGC, NYMAC WSGSC	# of unique hits accessing ACA sections on RC websites # of educational modules e.g., webinars other materials and disciplines of participants
Transition from pediatric to adult care	Access	Heartland, MSGRC, NEGC, NYMAC Region 4, SERC WSGSC	# of unique hits accessing transition sections on RC websites

The Evaluation WG sought to use existing measurement protocols for the evaluation. The HRSA Performance Measure Form #41, Medical Home was used by the RCs to capture data on their medical home activities. In partnership with the RCs, the NCC collected data from the RCs' constituencies with a Working Together instrument [*Working Together: A Profile of Collaboration* (Chrislip & Larson, 1994)] that had previously been used by the Heartland RC. The sources of the protocols selected for the national evaluation include the Children with Special Health Care Needs Survey (NS-CSHCN) developed by HRSA, the Medical Home Index (MHI) developed by the Center for Medical Home Improvement and a transition preparation question from Got Transition?



Because no data currently exist on how individuals with genetic conditions access health care, support, and services, the NGEEN worked with the Evaluation WG to develop a Survey on Access to Care for Individuals with Genetic Conditions. (Appendix K - survey instrument) Sixteen of the 21 questions were from NS-CSHCN and were modified to allow for responses from both parents of individuals with genetic conditions and the individuals with conditions themselves, and to capture responses from respondents of all ages about their own or their child’s care. The survey was reviewed and approved by the Genetic Alliance’s Institutional Review Board.

Similarly because no data exist from the perspective of genetic services providers, the NCC working with the Evaluation WG examined protocols that would be appropriate to use with the ACMG Clinical Services Directory. The MHI was developed for primary care practices serving children with special health care needs and has been shown to be a reliable and valid tool to assess medical “homeness” at the primary practice level. The NCC staff spoke with the Center for Medical Home Improvement about modifying the instrument for use with genetic service respondents. The language in five MHI questions was adapted for this audience. This is the first attempt to use the MHI with genetic specialists. A transition preparation question from Got Transition? was also included.

In both the NGEEN and NCC surveys, questions were asked about the NCC/RCs. This information was useful in assessing the visibility of the NCC/ RCs with individuals or families with genetic conditions and with genetic service providers.

The following table characterizes the terms used in this evaluation effort and in this report, with the corresponding HP 2020 objectives and the HRSA RC priorities. The table also identifies the measurement protocols that were used by the NCC/RCs to capture baseline data.

Table 2. EVALUATION TERMS, FRAMEWORK AND MEASUREMENT PROTOCOLS				
Generic terms	HP 2020 objectives	HRSA RC priorities	RC measurement protocol	NCC measurement protocol
Access	MICH 30	Treat in the context of a medical home & Improve insurance coverage policy and reimbursement	HRSA Performance Measure Form #41, Medical Home	Children with Special Health Care Needs Survey (NS-CSHCN) and the Medical Home Index MHI)
Care Coordination	MICH 31	Expand the pool of the genetic service workforce		NS-CSHCN and MHI
Newborn Screening Follow-up	MICH 32.2 & 32.3	Expand state and regional collaborative systems of cohorts of patients for long-term monitoring and analysis of follow-up and treatment for provider and/or patient access.		
Collaborations	NA- not applicable	Strengthen public-private partnerships	Working Together	NGEEN Survey of the RCs
Transition	DH 5	NA- not applicable		NS-CSHCN Got Transition?



Data Collection and Analysis

As the RCs worked to find a common set of measures, the NGEEN asked the RCs about their consumer engagement activities. Based on questions presented at the November 2012 PD/PM meeting, the RCs were asked in March 2013 about the level and type of consumer participation and the ways that they support consumers in their activities. These data were presented in a June 2013 webinar hosted by the NGEEN.

In May/June 2013, the NCC administered the common evaluation measures. Some of the rules governing the data collection were that RCs should report activities where HRSA grant funds were expended. This includes RC staff time and contributions that partially support other partners' activities (e.g., public health department or other HRSA grantees). RCs should answer with unduplicated counts of activities and participants. For example, an educational session that has a primary purpose of teaching about medical home, but which also touches upon insurance changes under the ACA, should be counted as only a medical home session.

In May 2013, NGEEN distributed the survey to the Genetic Alliance's network of over 700 disease-specific organizations. The survey was also disseminated widely through social media reaching an estimated 10,000-25,000 individuals. The data collection process occurred over a 3-week period. It is important to point out that by using this method of dissemination, responses are not from a random sample of individuals; responses are from individuals who are already connected to or receive information from disease support and advocacy organizations.

In August 2013, the NCC administered questions to the ACMG clinical services membership database. To incentivize members to participate, they could opt into a drawing for a complimentary 2014 ACMG Annual Clinical Genetics and Genomics meeting registration. Over a 3-week period, some 369 people were contacted and 85 responded. The response rate was 23% (n= 85).

The data from the NGEEN survey was collected using the online survey tool, SurveyMonkey. The data was aggregated into a database for data cleaning and imported into SPSS statistical software for quantitative analysis. Descriptive statistics, such as frequencies and percentages, were calculated using SPSS software. The NCC-administered surveys were done through SurveyMonkey and imported into SPSS for quantitative analysis. The on-line platform enabled the RCs to have access to their data and the NCC to analyze the responses.



NCC/RC COMMON MEASURES FINDINGS

Collaborations

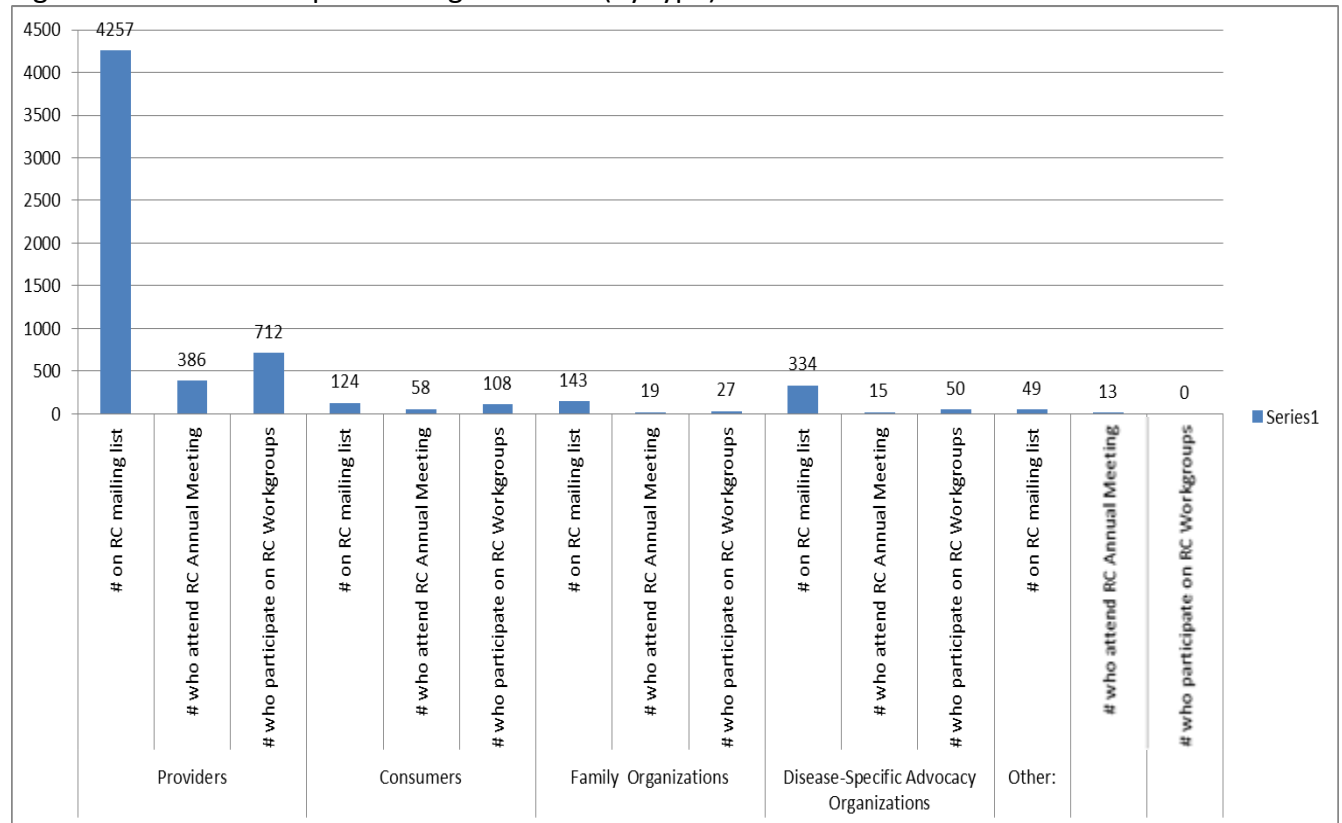
The NCC's partners include the American Academy of Pediatrics (AAP), Association of Public Health Laboratories (APHL), the Association of University Centers on Disabilities (AUCD), Got Transition?, Catalyst Center, Genetics in Primary Care Institute, and other HRSA grantees. Representatives of these groups participate on PD/PM calls and on the NCC Working Groups. In 2012-13, the APHL NewSTEPS program staff and the AUCD Leadership Education in Neurodevelopmental Disabilities (LEND) programs were of particular focus for collaboration.

The RCs are engaging health and public health professionals and consumers in collaborative activities. The mailing lists of the RCs reach more than 4,300 people (See Table X). The RCs collaborate with 108 family and 334 disease-specific or advocacy organizations. Their partners include state chapters of Family Voices, Family to Family Health Information Centers and Parent to Parent organizations, etc. They engage disease specific and advocacy organizations that address Cystic Fibrosis, Down Syndrome, Huntington's Disease, Muscular Dystrophy, PKU and sickle cell disease, etc. They also partner with HRSA grantees such as Baby's First Test, the Catalyst Center and the LEND programs in a few states. These groups also participate in the RCs' annual meetings and working groups. Also found in Table 3 are data on the numbers of people and organizations that participate in RC activities. The RCs are engaging health and public health professionals in their annual meetings (n=386) and in workgroups and committees (n=712).

Table 3				
COUNTS OF PEOPLE AND ORGANIZATIONS THAT THE RCS WORK WITH				
	HEALTH AND PUBLIC HEALTH PROFESSIONALS (MD, PA, RN, Public health, etc.)	CONSUMERS (Affected Individuals & Family members)	FAMILY ORGANIZATIONS	DISEASE-SPECIFIC or ADVOCACY ORGANIZATIONS
# on RC mailing list	Range (per RC): 48-2,756	Range: 13-30	Range: 0-102	Range: 1-285
	Total: 4,257	Total: 124	Total: 108	Total: 334
PARTICIPANTS IN RC ACTIVITIES				
# who attended RC annual meeting	Range: 23-133	Range: 0-16	Range: 0-7	Range: 0-10
	Total: 386	Total: 58	Total: 19	Total: 15
# who participate on RC workgroups and committees	Range: 0-300	Range: 0-35	Range: 0-9	Range: 0-26
	Total: 712	Total: 108	Total: 27	Total: 50



Figure 2. Counts of People and Organizations (by type)



In discussing these ranges, the RC PD/PMs noted that they have different communication philosophies and strategies. One RC has a large e-mail distribution list (2,756) that is used to notify partners of important genetic issues. Another RC uses a blog to notify their stakeholders. One RC PD commented that the size of their network reflects the resources available to pursue outreach and to engage new partners.

Consumer Engagement

While the sizes of the RCs' networks vary, the visibility of the NCC/RCs was low among the respondents to the NGEN survey. Although these people are active in consumer or disease advocacy organizations, less than 11% (n=1580) had interacted in the past 12 months with any one of the NCC/RCs. Among the genetic service respondents, 72% (n=78) knew or had heard of the RCs; 53% (n=79) knew or had heard of the NCC. In the past year, 34% (n=79) had interacted with an RC. Of the 23 of the respondents, who indicated the types of interactions with the RCs, they reported that they sought resources for families; served in an advisory capacity; and participated in training and education sessions. When asked to indicate their level of interest in connecting with the RCs to address common areas of need, 83% (n=77) were definitely, most definitely or very definitely interested. (Appendix K.)

The NGEN collected data from the seven RCs on consumer engagement. Three of the RCs reported that consumer participation in their activities is high; two RCs said it was medium and two low. Compared to the previous grant cycle, five RCs reported that consumer participation is about the same; two said that it was higher. In all seven RCs, consumers participate in the RCs'



annual meetings and serve on RC advisory groups and committees. Among the roles for consumers, six RCs said that consumers encourage/mentor new consumers. In six of the RCs they also offer advice on best practices on RC activities.

The Working Together instrument was designed to evaluate the effectiveness of collaboration efforts across five areas (context, structure of the group, effectiveness of the members, collaboration process, and results of collaboration efforts). The Heartland RC had used this instrument and recommended it be utilized in the national evaluation to assess collaborations. It was administered by the NCC through Survey Monkey to 499 members of the advisory boards and working groups that collaborate with the RCs. The RCs were responsible for identifying and communicating with potential respondents. The response rate was 45% (n=227).

Of the 227 respondents, 27.5% were newborn screening professionals; 18% were consumers either individuals affected by a genetic condition or a family member; nearly 15% were public health professionals (genetics and no-professionals combined) and 10.3% were medical geneticists. The disciplines of the other respondents can be found in Appendix I.

Overall the RCs scored positively on the five components of the Working Together instrument. Based on a four-part scoring system (1 = True; 2 = More True than False; 3 = More False than True; 4 = False) and by calculating the arithmetic means (AM), we learned that most respondents found that the context for the collaboration to be well-timed and responsive to the issues and that the timing is right to address the newborn screening and genetic issues about which we are collaborating (n =211, AM = 1.1707) that the collaborative is responding to the critical issues faced in genetics and newborn screening (n = 211, AM = 1.3555). As for the structure of the RCs, we learned that the respondents felt that while stakeholders have agreed to work together (n = 196, AM = 1.2602) and they had effective means of communications (n =194; AM =1.3454) and access to the expertise for effective meetings (n = 197, AM = 1.3401), there was a need for more clearly defined roles (n = 197, AM = 1.7716).

There is high trust among the RC participants on the following item: Members sufficiently trust each other to honestly and accurately share information, perceptions and feedback (n =190, AM =1.3053). The effectiveness of the collaborative process is advanced by strong leadership (n = 189, AM = 1.2698). However, respondents said that it was more false than true that we frequently discuss how we are working together (n = 189, AM = 1.8836).

As for the results of the collaborative effort, the respondents said that the time and effort of the collaboration is directed at obtaining the goals rather than keeping itself in business (n=188, AM =1.3883). But when asked about whether there is an established method for monitoring performance and providing feedback on goal attainment, the respondents indicated that there was room for improvement (n=185, AM =1.7946).

Appendix I. contains the Working Together survey instrument, the number of responses received on each item and the mean responses.



Public Information

The NCC website is housed on the ACMG website with links to the seven RC websites. The *NCC Collaborator* is the newsletter of the RC/NCC system. Produced by the NCC with submissions from the RCs, it is distributed electronically by the NCC to an email distribution list of just under 100 individual direct e-mails. It is also posted on the NCC website. During the course of 2013, the NCC website had 2,548 visitors with 6,809 page views. Housed on the ACMG website another resource are ACT Sheets. Statistics show that the use of ACT Sheets continues to rise with 31,044 page views of the ACT sheets from June 1, 2012-May 31, 2013.

The RCs are using the Internet and social media to reach the public and healthcare professionals with information on medical homes, newborn screening and transition from pediatric to adult care. The NGEEN survey found that all 7 RCs have websites and all 7 provide links to patient/family organizations. Six of the RCs offer resources for individuals with genetic diseases (or their families) to help them access services. As a part of the national evaluation, the RCs examined the content of their websites based on the HRSA RC priorities. As shown in Table 4, six RCs have website content addressing medical homes, newborn screening, and collaborations. Two RCs have ACA content and five RCs offer information on newborn screening long term follow up and transition from adult to pediatric care. The use of these resources is shown in the Table on the following page.



Table 4 USE OF WEBSITES AND SOCIAL MEDIA

USE OF WEBSITES						
(Specific Website Sections by the following HRSA RC Grant Priorities)						
	Medical Home	Newborn Screening Capacity Building	Collaborations	Affordable Care Act Implementation	Newborn Screening Long Term Follow up	Transition from pediatric to adult care
RC website has pages that address these topics (Y/N)	6	6	6	2	5	5
If yes, # of unique visits	728	2,991	28,369	349	21,319	495
If yes, # of page views	1,338	873	36,324	701	43,780	760
If yes, # of links used	13	3	156	6	4	9
If yes, # of materials downloaded	116	38	0	0	10	32
USE OF SOCIAL MEDIA						
RC uses Social Media (Y/N)	5					
If yes, please indicate type AND Number Of unique followers as of May 31, 2013:	FACEBOOK: __4__ (# hits: 193) TWITTER: __4__ (# hits: 321) PINTEREST: _____ OTHER: __2__ (YouTube – 183 visits)					
If yes, # of Posts related to:	17	27	71	8	9	3
If yes, # of Reposts related to:	1	18	29	3	16	0



Five of the RCs have begun to use social media with four using Facebook and four using Twitter. Two RCs are also using YouTube. Posts and reposts were most frequently done on collaborations.

According to data collected by the NGEEN, four of the RCs have surveyed consumers in their regions and found that they want:

- Social media updates
- Networking opportunities
- Mentoring
- Reliable, accurate information on genetic diagnoses, information about basic genetics and genomics
- Advice on communicating with primary care providers regarding their or their child's genetic condition

Education and Training

Education and training were among the common RC activities. A total of 145 education and training sessions were offered in the first year, 70 of these sessions were offered as in-person trainings; another 50 sessions were delivered through webinars; and 20 were teleconference training sessions. (Table X)

Consumers were nearly 32% (n=675) of the participants in these training and education sessions (n=2,140). Newborn screening professionals (n=282); other healthcare disciplines (n=221) and genetic counselors (n=163). The disciplines of others participating in the training and education sessions are found in Table X below.

Table 5 Total Education and Training Activities by HRSA Priority and by Participant Type

	NBS			Affordable Care Act	NBS		Total
	Medical Home	Capacity Building	Collaboration		LTFU	Transition	
Consumer	155	53	84	94	21	268	675
Genetic Counselor	23	64	37	6	6	27	163
Other Genetic Services Provider	0	10	2	4	2	24	42
Medical Geneticist	11	23	20	4	8	17	83
Non-Geneticist Specialty Physician	16	4	11	9	22	22	84
Primary Care Physician	38	11	16	4	5	6	80
Other Healthcare Provider	94	29	21	3	29	45	221
Social Service Provider	9	2	1	1	3	12	28
Public Health Genetics Professional	16	58	58	9	36	45	222
Public Health Non-Genetics Professional	45	23	15	9	28	16	136
Newborn Screening Professional	21	116	68	14	25	38	282
Health Insurance Representative	0	0	0	0	0	0	0
Legislator or Legislative Staff	0	0	0	0	0	0	0
Other	13	71	3	4	2	31	124

Legend: Counts represent in-person, webinar, & telephone Conference combined



The RCs offered educational content in medical home, newborn screening capacity and long-term follow-up. Through these education and training sessions, people are increasing their knowledge about genetic issues, as well as increasing the number of partners and coalitions that are collaborating on genetic issues (see Figures 3 and 4 below and on next page).

Note: Two of the seven RCs [NEGC and Region 4 (Midwest)] included participants who were supported to attend a training session sponsored by another entity. NEGC also included peer-reviewed posters, publications and presentations, which the other RCs did not.

Figure 3 RC Education and Training Activities by Participant Type (% of total by category)

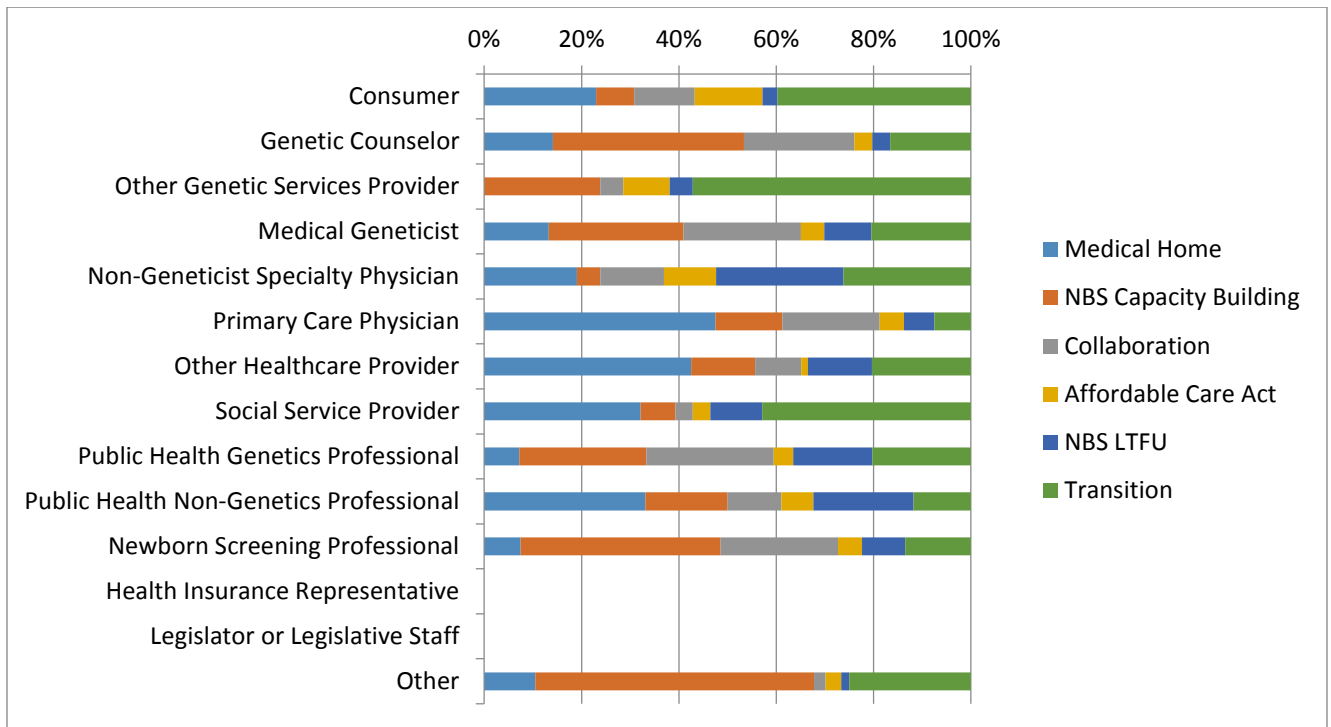
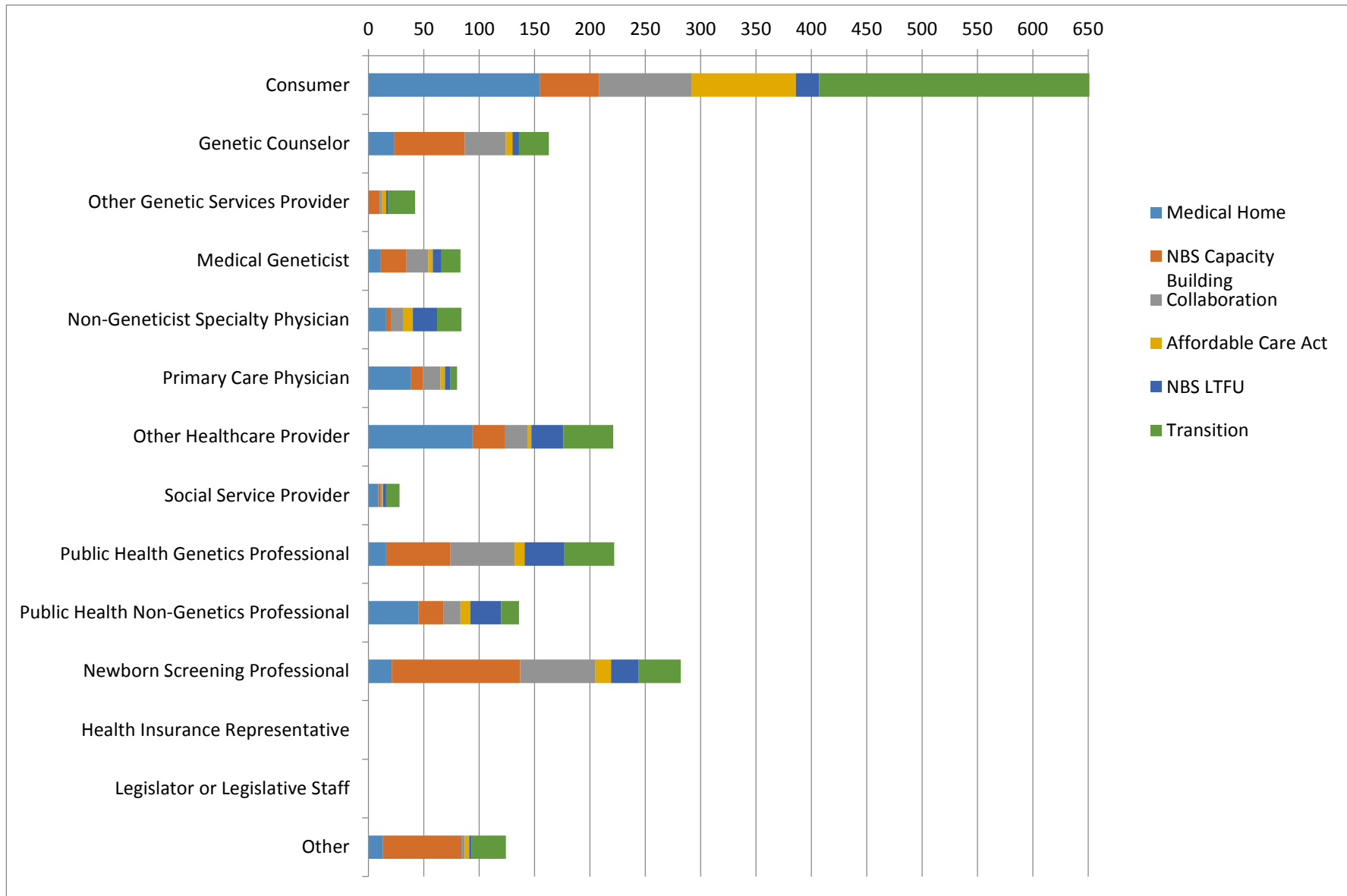


Figure 4 RC Education and Training Activities by Participant Type



Newborn Screening Capacity Building

The first-year data show that RCs are sponsoring activities to build newborn screening capacity. Several RCs hosted working groups and calls and disseminated materials to assist state public health newborn screening programs and to promote the Secretary’s Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (SDACHDNC).

In addition to promoting and financially supporting participation of Region 4 (Midwest) members in SDACHDNC meetings, the Region 4 (Midwest) RC convened five regional SACHDNC Implementation Workgroup meetings; coordinated technical assistance for states applying for CDC SCID funding; disseminated best practice: “CCHD and Public Health” panel discussion; disseminated best practice: “SCID Testing: Perspectives from Wisconsin and Michigan;” disseminated best practice: “Cora’s Story, Using Social Media to Advocate Change” (See <http://corasstory.com/>); identified states’ implementation status and barriers; discussed possible resolutions to the barriers with states; posted state testing algorithms on Region 4 (Midwest) SharePoint site; and initiated a newborn screening payment structure study.

NYMAC RC began and continues to support monthly CCHD calls. Following a June 2012 symposium, New York leads monthly SCID calls and provides extensive consulting to states in implementing SCID. Similarly, the Mountain States RC reported that they support a region-wide workgroup focused on NBS and a subgroup of NBS workgroup focused on point-of-care screening. The NBS workgroup agendas (for calls and in person meetings) frequently include items related to implementation of new disorders, including CCHD and SCID.

A regional project to implement Critical Congenital Heart Disease newborn screening has enrolled nine birthing centers in four New England states. A uniform screening protocol has been developed and a Tier 2 educational brochure for parents/families has been developed, printed and disseminated. Six of the nine centers have begun screening, and >10,000 babies have been screened through June 2013. There have been 3 positive screens, all were false positives. (See Supplement 1—RC Highlights)

Using definitions developed by the Region 4 (Midwest) RC, the RCs reported on state/Territorial adoption of two conditions on the Recommended Uniform Screening Panel (RUSP) - CCHD and SCID. As shown in Table 6, as of June 2013, screening is provided for all newborns in 13 jurisdictions for CCHD and in 11states and territories for SCID.

Table 6			
STATE/TERRITORIAL ADOPTION OF CCHD AND SCID RUSP			
(As of June 2013)			
	CCHD	SCID	
Universal	13	11	
Partial	26	5	
Planned	7	24	
No Activity	6	12	
Total:	52	52	
Universal: Screening is provided for all newborns Partial: Screening is being provided under pilot studies or on a voluntary basis by some hospitals Planned: State is considering No Activity: Not under consideration			



Access to Care – Medical Home

During the first grant year, all RCs began medical home activities. Because these projects are diverse targeting different audiences with different strategies, the Evaluation WG recommended that the HRSA Performance Measure #41 Medical Home Detail Sheet be used to establish a baseline on the RCs' medical home activities. The purpose of this form is to assess "the degree to which grantees have assisted in developing, supporting, and promoting medical homes for MCH populations." A total of 24 elements are arrayed in five categories for grantees to report on how they "contribute to a family/patient-centered, accessible, comprehensive, continuous, and compassionate system of care..." In addition to the OMB-approved four response categories (0 = Not Met; 1 = Partially Met; 2 = Mostly Met; 3 = Completely Met), the RCs also reported when an element was not applicable (NA). NA was coded as a zero for the purpose of this analysis.

Out of a possible total score of 72, the seven RCs had an average score of 18 (range 0-45). In year one, five RCs reported activities in three elements, as follows:

- 15. The grantee has developed Web sites and/or other mechanisms to disseminate medical home information;
- 19. The grantee has coordinated and/or facilitated communication among stakeholders serving MCH populations...; and
- 22. The degree to which the grantee has shared medical home tools with other communities and States.

Three RCs reported year-one activities in the following elements:

- 8. The grantee has developed/implemented quality improvement activities to support medical home implementation;
- 17. The grantee has engaged in public education campaigns about the medical home; and
- 18. The grantee has established a multidisciplinary advisory group, including families and consumers representative of the populations served, to oversee medical home activities.



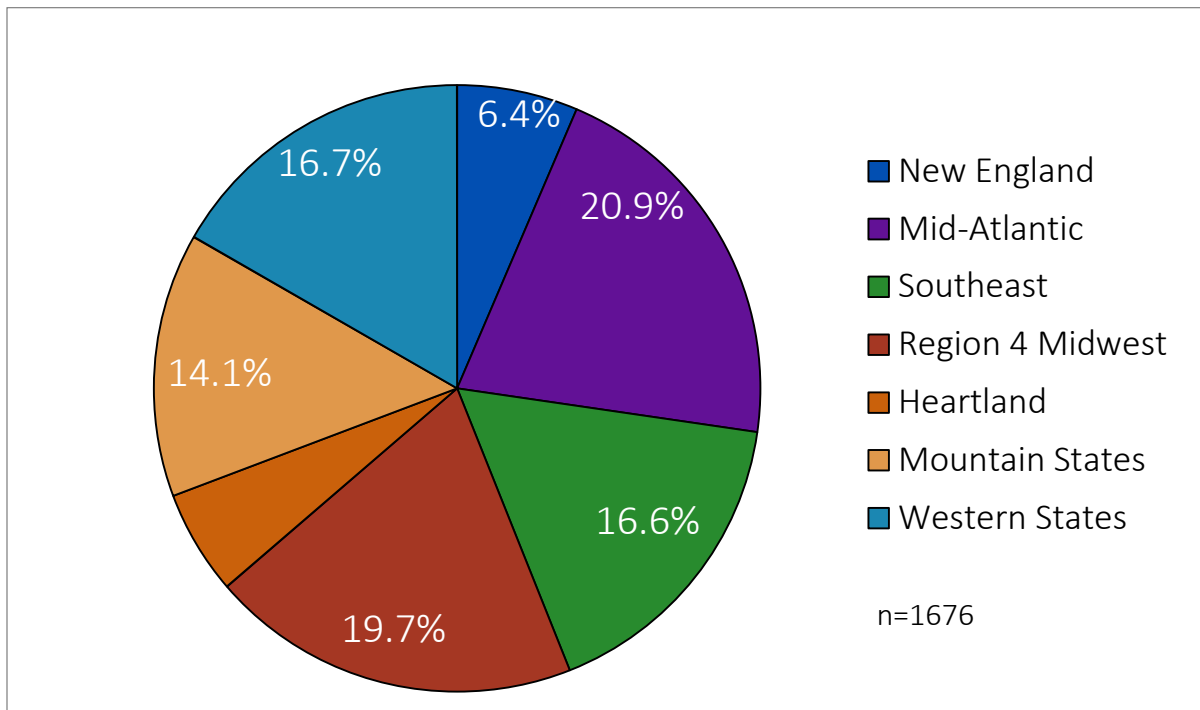
Survey on Access to Care for Individuals with Genetic Conditions

As described in the Methods section of this report, a major NGEEN activity was to conduct a survey of consumers – individuals affected with genetic conditions or their family members. The Survey on Access to Care for Individuals with Genetic Conditions was developed to begin to paint a portrait of health care experiences e.g., care coordination, medical home, and transition from pediatric to adult health care - HRSA priorities for the NCC/RC system.

Demographics

The survey had a total of 1895 respondents who indicated that their doctor or other health care provider had told them that they or their child have a genetic condition. Of those respondents (n=1661), 53.1% indicated that they were individuals with a genetic condition and 46.9% were parents of one or more individuals with a genetic condition. The remaining percentage did not indicate their status but were included in the overall analysis. Respondents who indicated they were parents were asked to answer the survey questions on behalf of their oldest living child. Data include only responses from individuals within the United States. Figure 5 shows the distribution of respondents by RC.

Figure 5. Survey Responses by Regional Collaborative

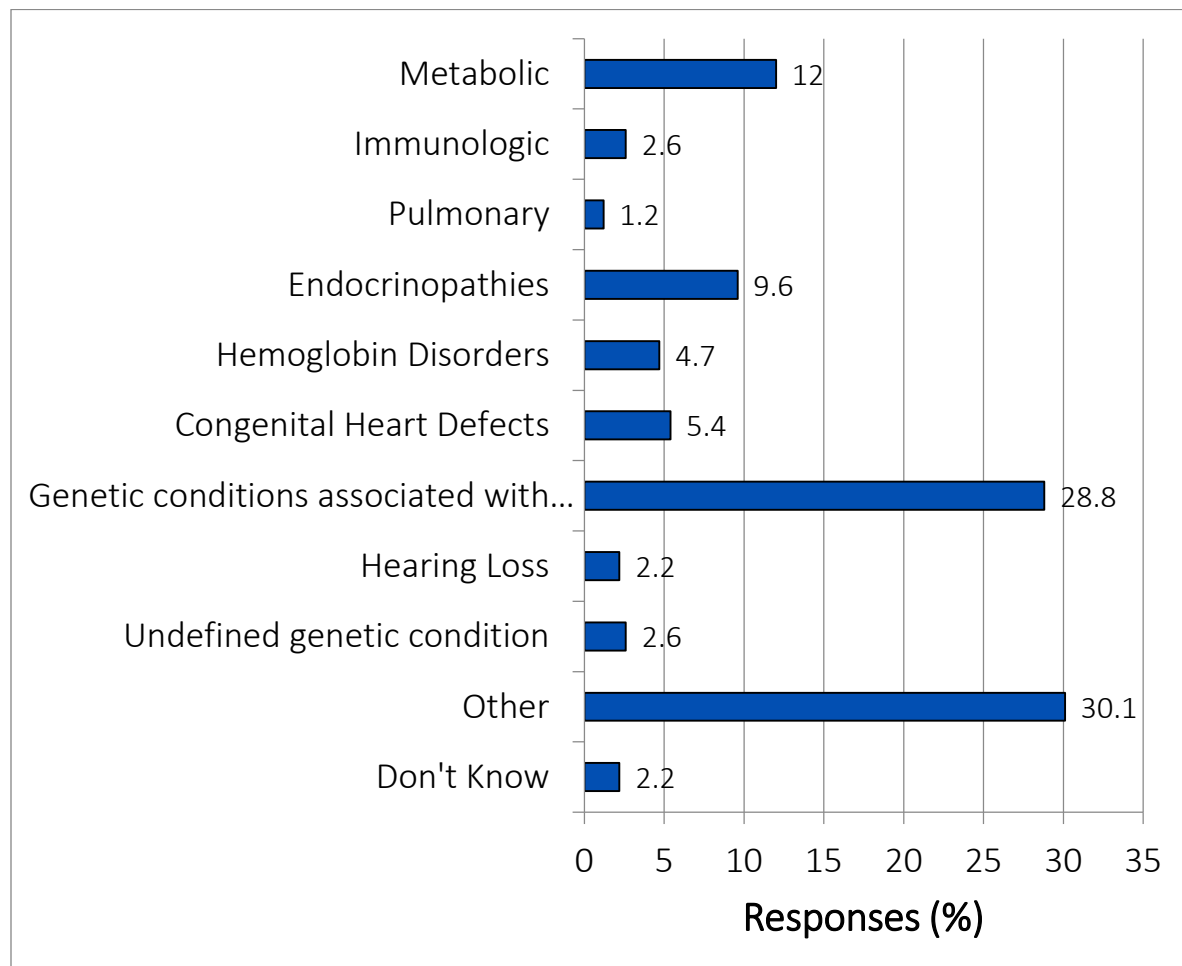


Genetic Conditions

Respondents were asked to select all of the conditions for which they have ever been given a diagnosis from a list of categories of genetic conditions (i.e. Metabolic, Immunologic).

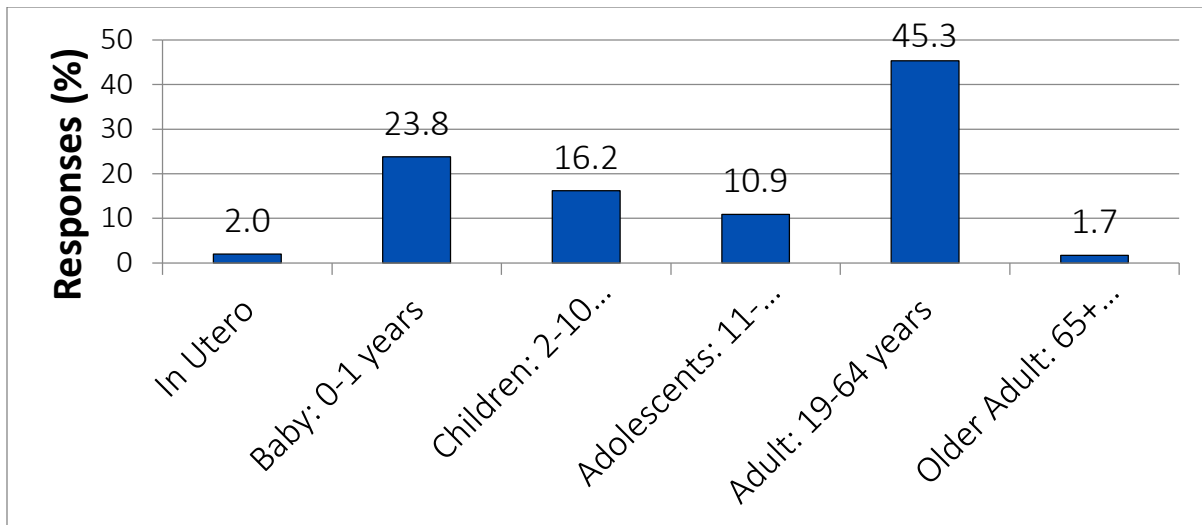
Respondents had the option of specifying other conditions, or that they have an undefined and/or unknown genetic condition. Figure 6 shows that respondents were most likely to report having a condition other than those listed (30.1%). Twenty-nine percent (28.8%) of respondents reported having a genetic condition associated with other conditions, such as having chromosomal abnormality, or both deafness and heart defects. The complete list of conditions can be found in Appendix X., Question 3c. In the "Other Condition" category (n=570), the three most frequently reported conditions were: Hereditary Hemorrhagic Telangiectasia (HHT) (34.6%; n = 197 responses); Pseudoxanthoma Elasticum (PXE) (13.2%; n = 75); and Long QT Syndrome (10.9%; n = 62).

Figure 6. Genetic Conditions Identified



In response to the question, “When were you or your child first identified as having a genetic condition?” (N=1668) most respondents (45.3%) reported the genetic condition was found in adulthood (19-64 years old). Twenty-four percent (23.8%) of respondents reported they were first identified as a baby (0-1 years). Figure 7 shows the distribution of ages when respondents reported they were first told they have a genetic condition. The genetic condition was identified by newborn screening for 10.6% (n=177) of respondents.

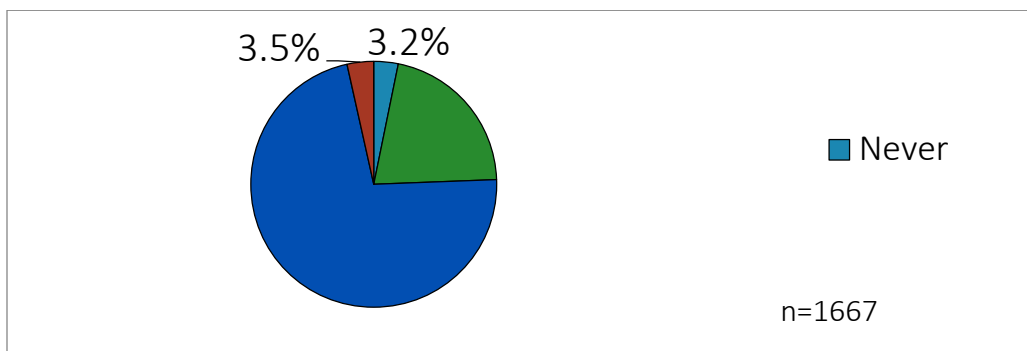
Figure 7. Age When Genetic Condition Was First Identified



Access to Care

Survey questions asked about health insurance coverage to better understand how coverage may affect access to care. The survey asked respondents about whether their health insurance coverage offers benefits or covers services that meet their (or their child’s) needs. The majority of respondents (72.1%) reported that their health insurance usually or always covered their needs (total n=1667) (See Figure 8). Nearly a quarter (24.4%) of respondents felt their insurance sometimes or never meets their needs (Appendix K. Question 4a.) When asked about coverage during the past 12 months (n=1666), only 13.7% (n=229) reported they were not covered by any health insurance.

Figure 8. Insurance Coverage Meets Needs

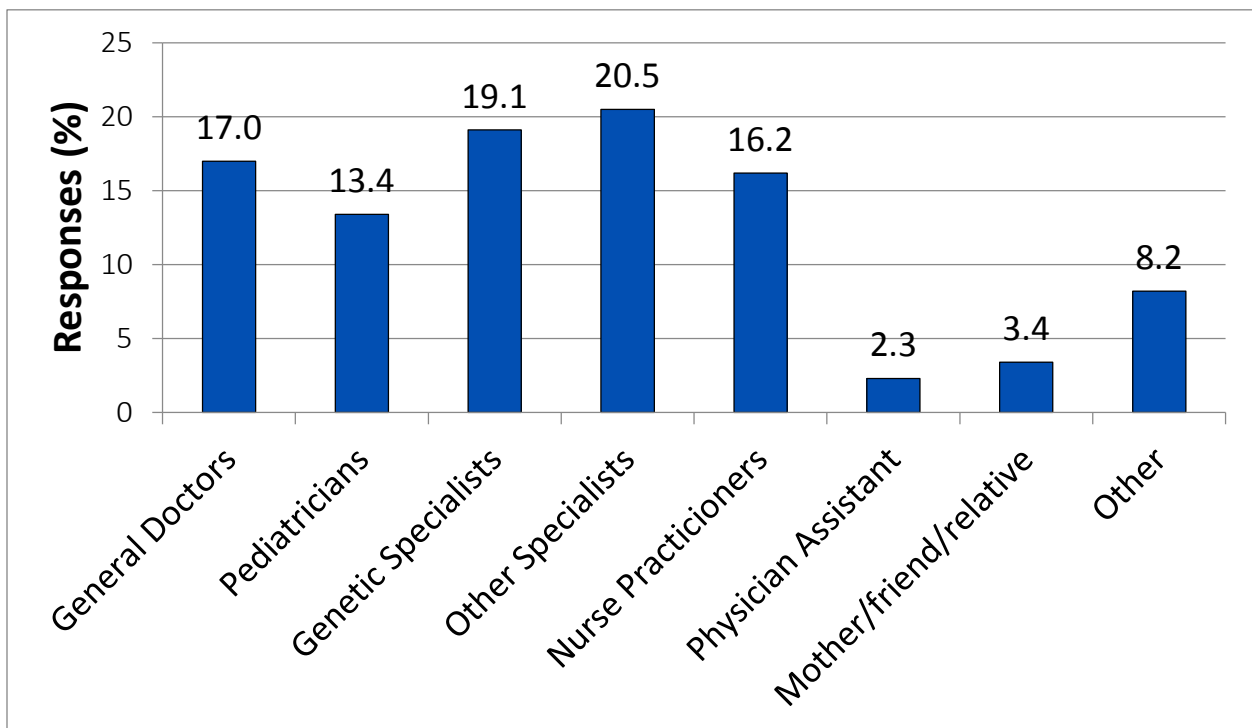


Thirty-five percent (35.1%; n=571) of respondents reported there was a time during the past 12 months that they or their family members needed genetic counseling for advice about inherited conditions related to medical, behavioral, or other health conditions. Of those respondents (n=568), 52.1% reported they received all the genetic counseling that was needed. Forty-three (43.4%) percent of respondents felt they did not receive all the genetic counseling that was needed and 4.6% did not know. The most frequently reported reasons for not getting all of the needed genetic counseling were: “Cost” (18.9%); “Insurance Coverage Issues” (17.1%); and, the “Doctor did not know how to treat or provide care” (15.1%). (Appendix K. Question 5b.)

Medical Home

Health professionals interact and play a unique role in each person’s health and quality of care. The majority of survey respondents (62.0%, n= 953) reported that they consider more than one person to be their (or their child’s) personal doctor or nurse: a health professional who knows their (or their child’s) health history. For 32% of respondents, this role is played by only one person. Respondents were asked to identify the kind of health professional or person they think of as their (or their child’s) personal doctor or nurse. Respondents were able to select multiple categories. The most frequently reported responses were “Other Specialists” (20.5%); “Genetic Specialists” (19.1%); General Doctors (17.0%); and Nurse Practitioners (16.2%). See Figure 9 for the distribution of responses by category.

Figure 9. Personal Doctor or Nurse



Care Coordination and Communication

Respondents were asked whether during the past 12 months they got as much help as they wanted with arranging or coordinating their (or their child's) care (n=1522). The majority of respondents (65.3%, n=994) reported they always or usually got the help they wanted with care coordination. More than 22% of respondents reported that someone helps arrange or coordinates their or their child's care among the different doctors or services that they use. Overall, respondents were satisfied (74.7%) with the communication among their doctors and health care providers. Seventy-five percent (75.3%) of respondents said their doctors always or usually listen carefully to them as patients. Almost 70% (n=1066) of respondents felt that they are always or usually partners in their (or their child's) care.

Transition from Pediatric to Adult Health Care

Three questions were asked about transition for respondents 12-17 years old (adolescents). The first was whether providers limit their practices to either pediatrics or adults (n=136). Eighty-three percent (83.1%) of respondents reported their doctors or other health care providers treat only children. Eleven percent (11%) of respondents said "no," indicating that their providers did not limit their practice to either children or adults (n=136). Seventy-nine percent (78%; n=106) of transition age respondents (n=167) reported their doctors have not talked with them about their health care needs as they become an adult. Only 10% (n=13) of respondents (n=135) said that their providers have talked with them about transition, or seeing doctors or other health care providers who treat adults.

Information and Support

Support is an important part of health and access to care. Forty-two percent (41.8%; n =638) of respondents reported they never or rarely get the social and emotional support they need from a health care provider or support/advocacy group (n=1526). Only 30.1% (n=462) of respondents said they were referred by a health care provider to a support and/or advocacy group. This finding is especially interesting as all of the survey respondents received the survey through being connected with a disease support or advocacy organization. Finally, family health history was discussed with a majority of respondents (79.2%, n=1275).



ACMG Survey on Care Coordination

As described in the Methods section of this report, the NCC administered a survey to the genetic service providers in the ACMG clinical services database. Five questions from the MHI instrument and one question from Got Transition? use a four-level scoring system (Level 1 – 4). Level 1 is the lowest level; subsequent levels include the items in Level 1 and build in additional items, which creates a progressive continuum of items from Level 1 to 4. The respondents selected the level that best describes how their practices currently provide care.

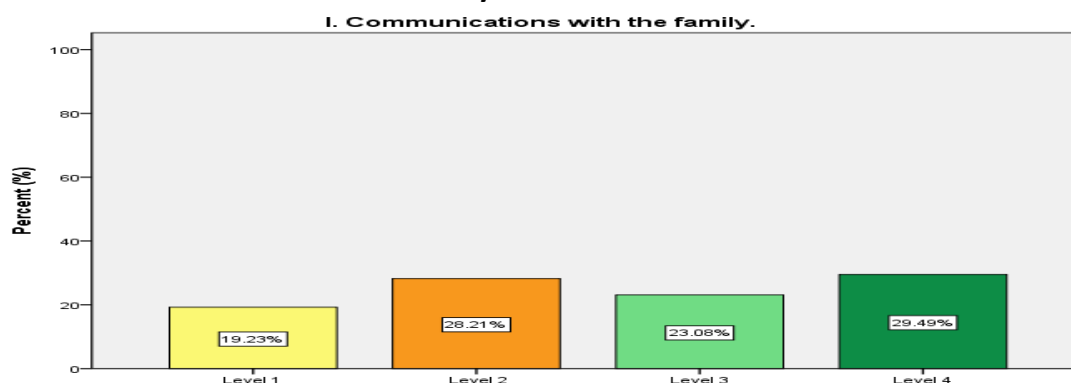
Demographics

The survey had a total of 85 respondents all of whom are genetics professionals (medical geneticists, genetic counselors, laboratory geneticists, etc.). They are all ACMG members. Some 27 respondents indicated they interacted with an RC in the past year; 20 identified which RC.

Communication

Communications between families and providers and among providers are particularly important for care coordination for people with genetic conditions. Two questions were asked of the genetic specialists about how their practices communicate with families and with other providers. The distribution of the responses was about equal between the four levels of communications with families. Almost 30% of the practices (n=23) were at Level 4: encouraging individual requests; with flexible access; and communication preferences documented in the care plan.

Figure 10. Communications with the Family



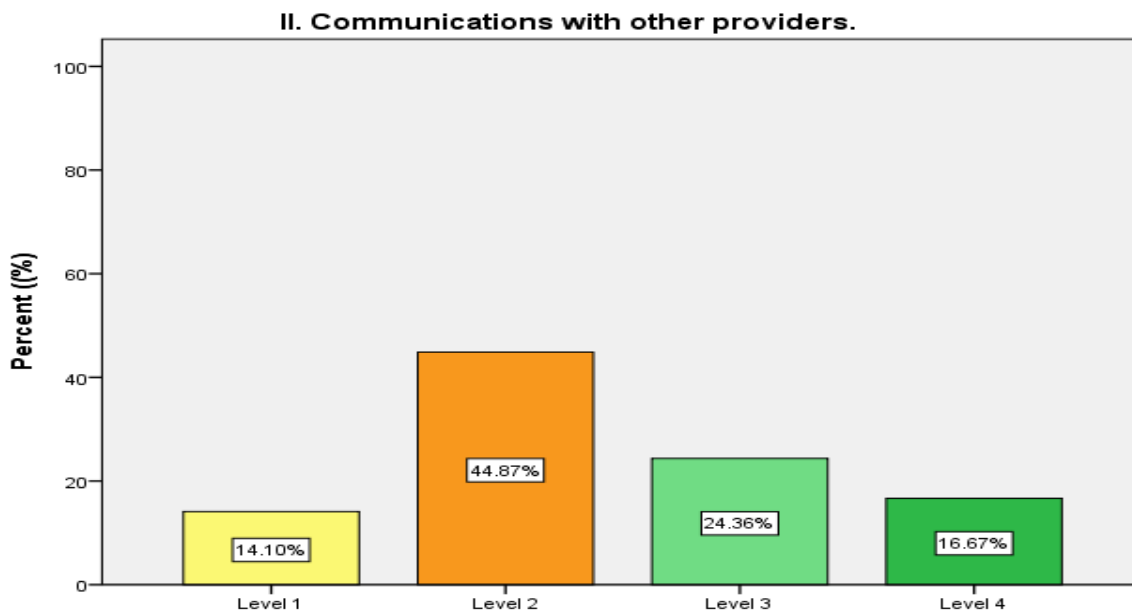
	N	Valid Percent	Cumulative Percent
Level 1: Communication between the family and the provider occurs as a result of family inquiry; provider contacts with the family for test result delivery or planned medical follow-up.	15	19.2	19.2
Level 2: In addition to Level 1, standardized office communication methods are identified to the family by the practice.	22	28.2	47.4
Level 3: In addition to Level 2, practice and family communication at agreed upon intervals and both agree on "best time and way to contact me," individual needs prompt weekend or other special appointments.	18	23.1	70.5
Level 4: In addition to Level 3, office activities encourage individual requests for flexible access; access and communication preferences are documented in the care plan and used by other practice staff.	23	29.5	100.0
Total	78	100.0	

Nearly 45% (n=35) of the respondents said that their communication with other providers was at Level 2: provider makes requests and/or responds to requests from agencies or schools; all communication is documented. Another 24.4% (n=19) of the respondents said that their



communications were at Level 3: systematic practice activities foster communication among the practice, family, and external providers such as schools, other community professionals; these methods are documented and may include information exchange forms or ad hoc meetings with external providers.

Figure 11. Communication with Other Providers



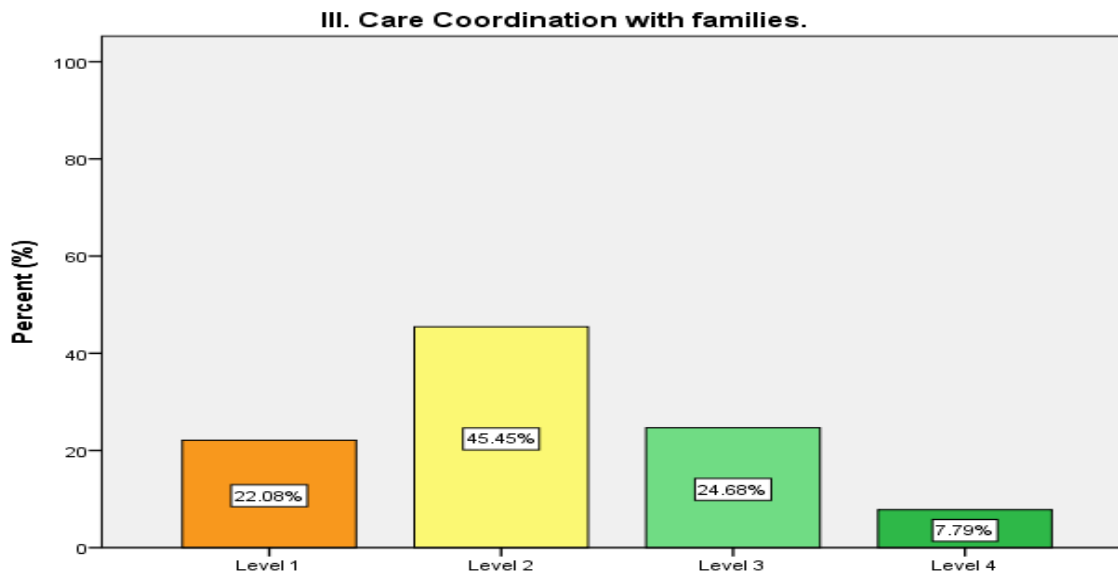
	N	%	Cumulative Percent
Level 1: Communication among the primary care provider, specialists, therapists, and school happens as needs arise.	11	14.1	14.1
Level 2: In addition to Level 1, provider makes requests and/or responds to requests from agencies or schools; all communication is documented.	35	44.9	59.0
Level 3: In addition to Level 2, systematic practice activities foster communication among the practice, family, and external providers such as schools, other community professionals; these methods are documented and may include information exchange forms or ad hoc meetings with external providers.	19	24.4	83.3
Level 4: In addition to Level 3, a method is used to convene the family and key professionals on behalf of children with more complex health concerns, specific issues are brought to this group and they all share and use a written plan of care.	13	16.7	100.0
Total	78	100.0	



Care Coordination

Three questions explored care coordination. Some 45.5% (n=35) said that their practice is at Level 2: ... families (and their older children) are regularly asked what care supports they need; treatment decisions are made jointly with the provider.

Figure 12. Care coordination with Families

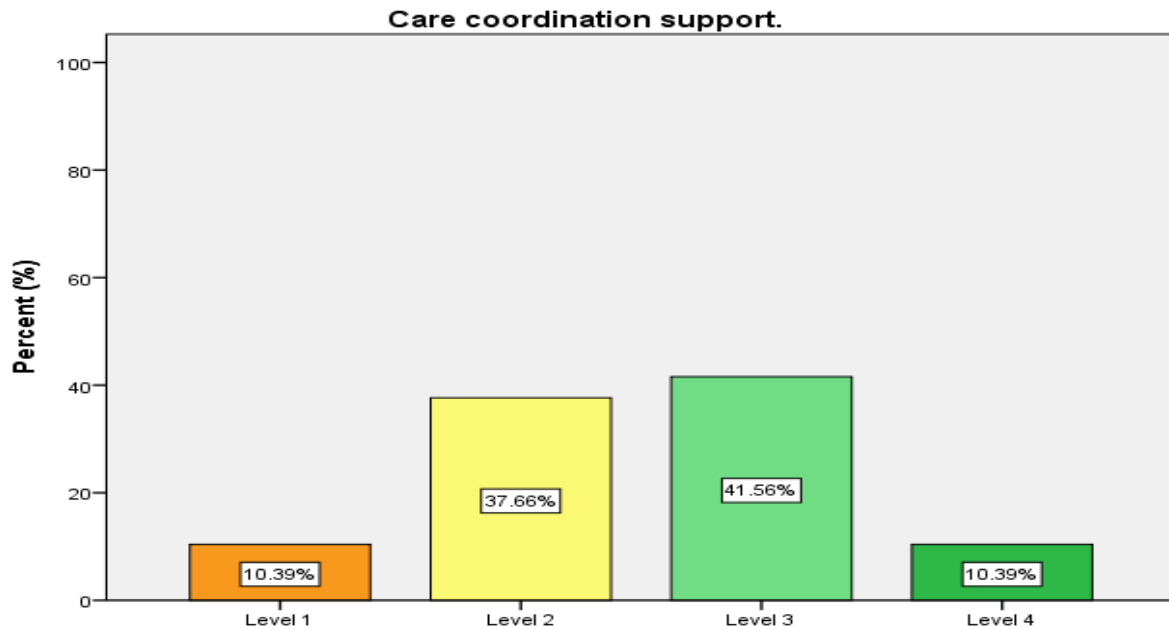


	N	%	Cumulative Percent
Level 1: The provider makes medical recommendations and defines care coordination needs, the family carries these out.	17	22.1	22.1
Level 2: In addition to Level 1, families (and their older children) are regularly asked what care supports they need; treatment decisions are made jointly with the provider.	35	45.5	67.5
Level 3: In addition to Level 2, families (and older children) are given the option of centralizing care coordination at and in partnership with the practice.	19	24.7	92.2
Level 4: In addition to Level 3, children and families contribute to a description of care coordination activities; a care coordinator specifically develops and implements this practice capacity which is evaluated by families and designated supervisors.	6	7.8	100.0
Total	77	100.0	



In terms of care coordination support, the respondents (41.6%, n= 32) said that their current practice was at Level 3: ... care coordination activities are based upon ongoing assessments of child and family needs; the practice partners with the family (and older children) to accomplish care coordination goals.

Figure 13. Care Coordination Support

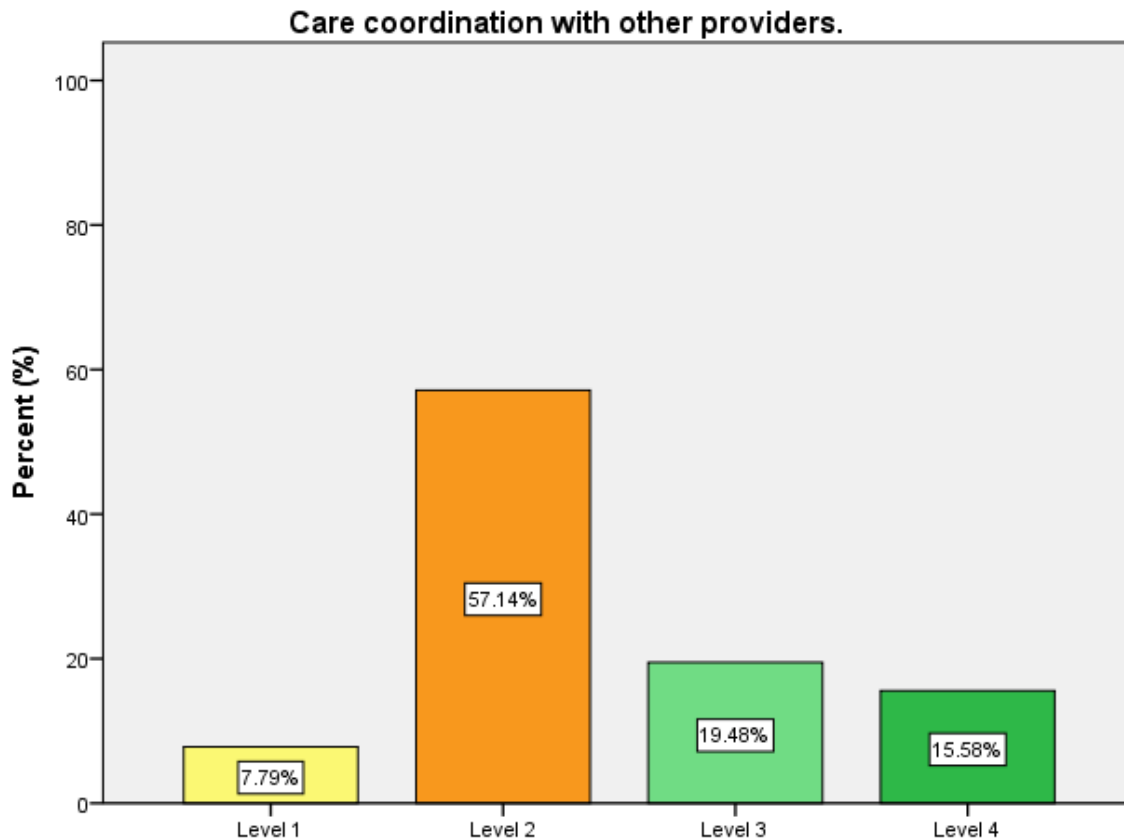


	N	%	Cumulative Percent
Level 1: The family coordinates care without specific support; they integrate office recommendations into their child's care.	8	10.4	10.4
Level 2: In addition to Level 1, the provider or a staff member engages in care support activities as needed; involvement with the family is variable.	29	37.7	48.1
Level 3: In addition to Level 2, care coordination activities are based upon ongoing assessments of child and family needs; the practice partners with the family (and older children) to accomplish care coordination goals.	32	41.6	89.6
Level 4: In addition to Level 3, practice staff offer a set of care coordination activities; their level of involvement fluctuates according to family needs/wishes. A designated care coordinator ensures the availability of these activities including written care plans with ongoing monitoring.	8	10.4	100.0
Total	77	100.0	



Care coordination with other providers (57.1%, n=44) was at Level 2: ... specialty referrals use phone, written and/or electronic communications; the provider waits for or relies upon the other specialists to communicate back their recommendations.

Figure 14. Care Coordination with other Providers



	N	%	Cumulative Percent
Level 1: Referrals occur in response to specific diagnostic and therapeutic needs; families are the main initiators of communication between specialists and their pcp.	6	7.8	7.8
Level 2: In addition to Level 1, specialty referrals use phone, written and/or electronic communications; the provider waits for or relies upon the other specialists to communicate back their recommendations.	44	57	64.9
Level 3: In addition to Level 2, the provider and family set goals for referrals and communicate these to specialists; together they clarify co-management roles among family, pcp, and specialists and determine how feedback to the family is expressed, used, and shared.	15	19	84.4
Level 4: In addition to Level 3, the family has the option of using the practice in a strong coordinating role; parents as partners with the practice manage their child's care using other specialists for consultations and information. The specialist manages the majority of the care related to the genetic condition.	12	16	100.0
Total	77	100	

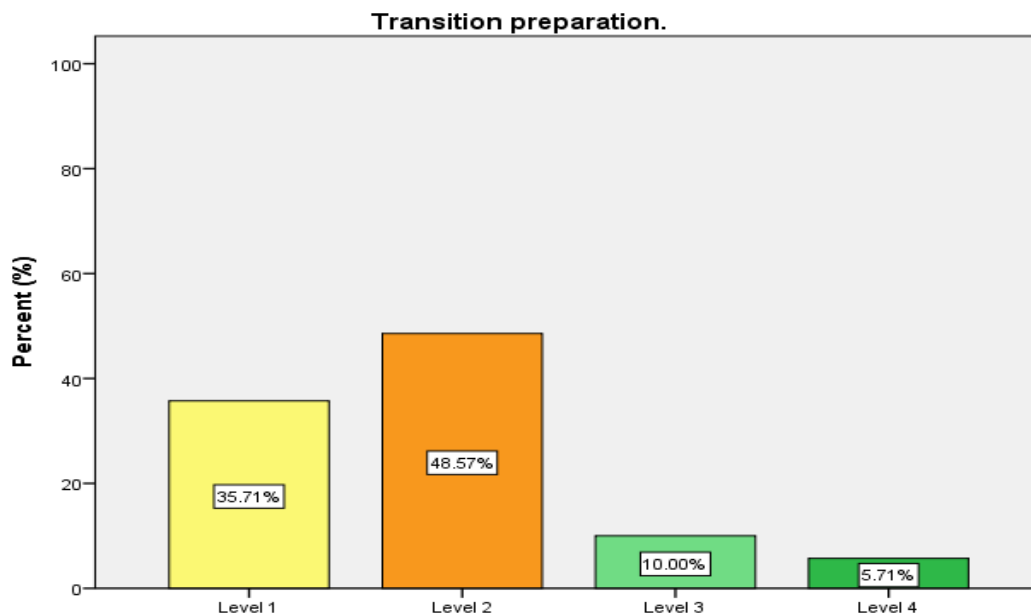


Transition from Pediatric to Adult Health Care

One question asked respondents to identify the level at which their patients begin to participate in their care transition. Some 35.7% (n=25) indicated that their practice was at Level 1 - Youth are seen without their family member for portions of the encounters after age 14, as their medical condition permits.

Nearly half (48.6%, n=34) indicated Level 2:..., by at least age 14, all youth begin a process of knowing their own health and wellness, risk behaviors, allergies, personal and family health history, insurance coverage. Youth with chronic conditions also acquire knowledge about their condition and related medications, specialists, and emergency care needs according to their ability.

Figure 15. Transition Preparation



	N	%	Cumulative Percent
Level 1: Youth are seen without their family member for portions of the encounters after age 14, as their medical condition permits.	25	35.7	35.7
Level 2: In addition to Level 1, by at least age 14, all youth begin a process of knowing their own health and wellness, risk behaviors, allergies, personal and family health history, insurance coverage. Youth with chronic conditions also acquire knowledge about their condition and related medications, specialists, and emergency care needs according to their ability.	34	48.6	84.3
Level 3: In addition to level 2, a practice transition checklist is used to assess a youth's knowledge of their own health needs, the team provides education and support the acquisition of youth's transition skills. Transition preparation targets knowledge gaps and builds youth skills; youth "teach back" their grasp of each checklist item.	7	10.0	94.3
Level 4: In addition to Level 3, families/youth work with staff and/or transition care coordinator to develop transition related skills and elevating levels of self-determination. From age 14, they participate in the development of a portable medical cummary. This summary "travels" with each youth as they age out of a pediatric model of care.	4	5.7	100.0
Total	70	100	

Consumer and Provider Perspectives on Communications, Care Coordination and Transition Preparation



Using the data collected by the NGENC and the NCC, it is possible to compare for the first time the perspectives of consumers who are affected by or at risk of genetic conditions with the views of clinical genetic service providers. While neither of these surveys captured data that can be generalized to the total population of people with genetic conditions or to all healthcare providers, this information offers insights into medical home issues, such as family/provider communications, care coordination and transition to adult care for adolescents. For the most part, the responses of consumers and providers reflect one another about the current state of care.

It is noteworthy, but not surprising for people with genetic conditions, that 62% (n = 953) of consumers reported that they (or their child) see more than one health care professional. This finding reinforces the importance of communications between families and clinicians and among the providers, as well as care coordination among practices and institutions. As for the discipline of the providers, who the consumers consider as their personal doctor, there was an almost equal distribution of the four most frequently reported provider types: “Other Specialists” (20.5%); “Genetic Specialists” (19.1%); General Doctors (17.0%); and Nurse Practitioners (16.2%). See Figure 9 in the Survey on Access to Care for Individuals with Genetic Conditions section for the complete array of responses.

Communications with Families

Overall, 74.7% (n = 450) consumers were satisfied with the communications among their doctors and health care providers. More than 75% (n = 1149) of respondents said their doctors always or usually listen carefully to them as patients. Almost 70% (n=1066) of respondents felt that they are always or usually partners in their (or their child’s) care.

While consumers gave their providers high marks for communications, the genetic specialists’ responses about communications with families was almost equally distributed between the four levels on the MHI scale, where Level 1 is the lowest level and Level 4 is the highest level. About 53% (n=78) of the genetic specialist respondents believe their current practice to be at Levels 3 and 4; while 47% were at the lower Levels 1-2 (See Appendix L, and previous section).

Care Coordination

Consumers were asked whether during the past 12 months they got as much help as they wanted with arranging or coordinating their (or their child’s) care. The majority of respondents (65.3%, n=994) reported they always or usually got the help they wanted with care coordination. However, only 22% (n = 338) of respondents reported that someone helps arrange their (or their child’s) care among the different doctors or services that they use.

The genetic specialists reported that their care coordination levels with families were skewed to the lower Levels 1-2. Nearly 68% (n=77) of the respondents were at Levels 1-2 and only 32% (n = 77) were practicing at the higher levels of care coordination for families. These findings reflect what the genetic specialists reported about their care coordination levels with other providers. Some 65% (n=77) were at Levels 1-2 and only 35% (n=77) considered themselves to be practicing at the higher Levels 3-4. The genetic specialists also acknowledged issues about their communications with other providers. Some 59% (n=78) of the respondents were at Level 1-2 communications with other clinicians.



Transition Preparation

The consumer survey found very low levels of transition preparation. Some 78% (n=106) of transition age (12-17 years) respondents reported their doctors have **not** talked with them about their approaching adulthood and its corresponding health care issues. The percentage having a discussion about transition to adult or other health care providers was only 10% (n=13).

Genetic specialists acknowledged that their transition preparation efforts to be low. Some 84% (n=70) stated their practices are at Levels 1-2. Among the questions in this survey, this was the highest percentage of respondents who reported the lowest levels on the MHI scale.

Because 84% (n=167), of the respondents with 12-17 years old (adolescents), reported that their clinicians only treat children, transition to adult care will be inevitable. This finding reinforces the importance of the work already being done by the NCC and RCCS. It also indicates an opportunity for the NCC/RC system to identify new ways to support both families and providers in transition preparation.

Summary of the Comparison of the NGEEN and NCC Surveys/Consumer and Providers Perspectives

From these surveys, it is clear that there is room for improvement to make a medical home a reality for more people who are living with genetic conditions and for their families. Of the consumers who reported there was a time during the past 12 months when they or a family member needed genetic counseling for advice about inherited conditions related to medical, behavioral, or other health conditions, 15.1% (n=571) reported they did not get the genetic counseling because the “doctor did not know how to treat or provide care.” Finding ways to connect people and clinicians and networking among providers to help people on their diagnostic and care odyssey will be a part of the NCC/RC mission.

Only 30.1% of respondents said they were referred by a health care provider to a support and/or advocacy group (n=462). Considering that the respondents received the survey through their affiliation with a disease support or advocacy organization, this finding is surprising. Ensuring that clinicians know available resources and helping individuals and families get connected with these well-respected resources is an opportunity for the NCC, NGEEN and RCs. This data points to needs and opportunities that can be addressed by the NCC/RC system in the coming years. We plan to publish these data in the near future so to make this information available to others. We will use this data to inform the activities of the NCC workgroups. While this report presents a summary of the aggregate data, the NGEEN and NCC have provided each RC with their region-specific data so that it might be actionable on a regional level.



DISCUSSION

The first-year national evaluation of the NCC/RC system has been an extensive and time intensive effort. It has been a collaborative enterprise that required the input of HRSA, the PDs/PMs, the Evaluation WG, and a Technical Advisory WG. The evaluation has been challenging because no measurement protocols specifically address the population living with genetic conditions nor have any survey instruments targeted genetic service healthcare providers.

By adopting existing protocols (e.g. NS-CSHCN and MHI), the NCC was able to collect data for the first time to inform HRSA and the RCs about the needs of individuals and families living with genetic conditions and to capture the perspective of genetic service providers. These data clearly show unmet needs for social and emotional support for families and affected individuals; transition preparation; and genetic counseling. The provider survey points to an opportunity to work with clinicians to improve communications and care coordination with families and among providers. Data from the NGEEN and ACMG surveys will serve as a baseline for assessing needs and informing future efforts of the NCC/RCs.

This evaluation finds that the RCs have strong collaborative relationships with the partners in their region, but do not have visibility among affected consumers. The public information resources that the NCC/RCs have posted on the Internet are attracting users, but are these resources widely known and meeting the needs of the information seekers? Through social media and increased outreach can the NCC/RCs become the go-to resource for information about genetic conditions and services? Through this evaluation and future data collection, we will strive to answer these questions.

For public health and clinical providers, the RCs are offering education and training resources that address a number of HRSA priorities. The ACMG provider survey found that the respondents were very definitely or most definitely interested in the following RC activities in the coming year:

- 67.5% (n=74) resources for families with genetic conditions;
- 52.6% (n=76) resources/information on uses of new genetic testing;
- 46.6% (n=75) ethical/legal/social implications of genetic testing; and
- 45.3% (n=75) reports on genetics-related activities.

This evaluation effort will continue to monitor the RCs' newborn screening capacity building activities. With the implementation of the ACA, the evaluation will strive to inform HRSA on access to care, particularly medical home and care coordination issues.

There is a commitment among the RCs to repeat the data collection that began this year. But there is also a need to seek some changes in the measures to better capture the types of projects that the NCC/RCs will pursue in coming years. For example, the Performance Measure #41 Medical Home Detail Sheet is based on what State Title V programs report and this is a broad definition. Several RC PDs/PMs expressed concern about the use of this form for



measuring medical home. Therefore a focus of the year-two evaluation effort will be on finding more tightly defined measures that reflect the RCs' medical home activities.

Challenges and Limitations

The diversity of RC activities undertaken to meet local needs made it challenging to find common evaluation measures. There are regional differences in genetic service resources and consumer needs and the states that they collaborate with are in various stages of ACA implementation. The RCs have different organizational affiliations that lead to different collaborations and activities. Three RCs are affiliated with academic institutions; two with state public health departments, and two with public health institutes. As grantees, the RCs proposed to HRSA specific first-year activities based on input from providers and consumers in their regions. Their work plans fall within nine broad HRSA priority areas. Within any priority area many of the RC activities are unique. A point of discussion with HRSA has been whether the RCs are expected to change their work plans to reflect the common evaluation measures.

The PD/PMs also had concerns about the use of the HP 2020 objectives as these population measures are beyond the scope of the RCs' infrastructure-building budgets. They felt that being held accountable for moving the needle on national health outcomes was not possible for several reasons. The small population numbers of persons with genetic disease conditions make it difficult to demonstrate an impact. Most regions lack person-level data and many of the regions are doing infrastructure, not clinical services projects.

Through its membership directories the NCC and NGEEN administered questions from existing national measurement protocols. The NS-CSHCN was chosen for the NGEEN consumer survey and the MHI and Got Transition? instruments were utilized with the ACMG clinical membership directory. The MHI was developed for primary care practices serving children with special health care needs. The NCC staff spoke with Dr. W. Carl Cooley about modifying the instrument for use with genetic services respondents. The language was adapted to fit this audience. The MHI has been shown to be a reliable and valid tool to assess medical "homeness" at the primary practice level. This is the first attempt to use it with genetic specialists.

The Evaluation WG created new instruments to capture data on the RCs' activities in the areas of medical home, care coordination, transition, collaboration, NBS screening and NBS follow-up. Despite discussions, some terminology was interpreted differently by the RCs. This raises questions about the comparability of the results. For example, some RCs have traditional mailing lists and large numbers; another RC uses a blog and does not do blast email distributions. We also know that there was variability in the RC responses. The response categories on the HRSA Performance Measure 41 were not defined in the OMB clearance package and are therefore subject to each RC's interpretation. Continued discussion in the Evaluation WG will lead to a more uniform approach in reporting.

The time period for data collection was the grant year, June 1, 2012 through May 31, 2013. However, it took seven months for the NCC to reach consensus with the RCs on an evaluation plan. The common measures were not decided upon until late spring 2013. This resulted in attempts to capture the data retrospectively. Going forward, we have standardized the



reporting of disciplines of participants in training and education sessions, which will help ensure greater comparability of results in future years.

While this first progress report establishes a baseline for the first year of this grant cycle, this is not necessarily a true baseline. Because the RCs have been working with states and localities and providers and consumers since 2004, this represents a baseline that reflects considerable efforts by the NCC/RCs to improve genetic services.

The indicators that were chosen for the evaluation are measurable approximations of outcomes. Outcomes such as reductions in emergency room visits due to improved care coordination or reduced costs or wait time for genetic counseling services through telemedicine are important indicators to have, but are not possible to acquire with the scope of resources available.

The results are not a representative sample of genetic providers or of consumers and are not generalizable.

In the first year, it is difficult to demonstrate regional or systems level impact.



Future Directions

With the full implementation of the ACA, the NCC/RCs stand ready to contribute their expertise to help ensure that providers are equipped to deliver coordinated quality care to people at risk or who have genetic conditions and to help assure that consumers have access to the services that they need. Through this first year's evaluation, we successfully demonstrated our ability to collect data from consumers through the NGEEN Access to Care for Individuals with Genetic Conditions instrument and from genetic specialists in the ACMG clinical membership directory.

The NCC will continue to support inter-RC cluster evaluation measures, e.g., telemedicine metrics such as genetic service visits provided by distance strategies. To the extent that five RCs begin to work on telegenetics, then these measures will be raised to the level of common NCC/RC evaluation measures.

Over the course of the coming years, new common evaluation measures are likely to emerge. These measures might include assessment of the regional approach, State adoption of case definitions and use of family history tools. We stand ready to be informed by other measurement efforts and to use these measures, e.g., Children's Health Insurance Program Reauthorization Act (CHIPRA) care coordination measures. The NCC/RCs will place additional emphasis on addressing genetics across the life course.

The NCC will continue to work on the inclusion of questions to identify persons with genetic conditions on national surveys. Only through these national efforts will there be definitive information about the segment of the population who are affected by heritable conditions.

The NCC/RCs will continue to refine its measures and replicate these data collection efforts in 2015 and 2017. The impacts we expect to achieve include:

- infrastructure improvements
- increased coordination between primary care and specialty care
- better educated providers
- increased consumer involvement
- translating new knowledge into practice
- better informed public policies and
- improved health and well-being of the people at risk or who are living with genetic conditions

