HRSA Evaluation Brief

Year 5 Evaluation Summary Data

From the National Coordinating Center for the Seven HRSA Regional Genetic Service Collaboratives

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Executive Summary

This is the final report to the Health Resources and Services Administration (HRSA) on the five-year national evaluation led by the National Coordinating Center (NCC) for the seven Regional Genetic Service Collaboratives (RCs) for years 2012-2017. The American College of Medical Genetics and Genomics (ACMG) administers the NCC under a HRSA cooperative agreement. The National Genetics Education and Consumer Network (NGECN) is a NCC component run by the Genetic Alliance. Each of the RCs operates under an individually awarded HRSA grant.

The overarching goal of this work 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.” (Source: NCC/RC Mission Statement Approved January 2013).

Beginning in 2012, a NCC Evaluation Workgroup convened evaluators from each RC. Through monthly conference calls, they identified and continue to monitor common data elements related to HRSA’s priorities (e.g., collaboration; public information; education and training; and telegenetics) and to Healthy People 2020 objectives that focus on care coordination; newborn screening (NBS) capacity building; and long-term follow-up (LTFU). These common evaluation measures are used as the framework for this report. In addition, the supplements to this report answer the three questions that have guided this evaluation from its inception: How much did you do? How well did you do it? Was anyone better off?

Because the fifth year of this grant cycle ends on the May 31, 2017, the data in this report reflect 9 months of program activity (June 1, 2016 to February 28, 2017). In order to provide this report by the conclusion of the funding period, HRSA asked that a shortened reporting period be utilized for this report. Therefore, caution should be used in comparing the fifth year data to the prior four years that reported 12 month data intervals.

This evaluation brief highlights NCC/RC activities and provides summary data on these programs. This information is designed to demonstrate the value of regional approaches and tracks the impact of NCC/RC programming from local activities at the RC level through to national activities at the NCC/NGECN level.

SELECT 2016-17 ACCOMPLISHMENTS include:

More states and territories adopted the recommended uniform screening panel (RUSP) for critical congenital heart disease (CCHD) and severe combined immunodeficiency (SCID) conditions. This means that babies are being screened for **CCHD in 50 states and territories** and for **SCID in 46 states** and territories in 2017.

Underserved populations have access to genetic services through RC-sponsored genetic consultations in the New England region and in Alaska and Hawaii.

**Public health capacity has been assessed in 32 states in five regions** that have used the Genetics Services Assessment tools.

Providers and families accessed actionable information from NCC/RC resources on the web and received education and training in sessions that were hosted by the NCC/RCs.

Providers were trained to deliver genetic services through telehealth videoconferencing.

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1 Cooperative Agreement No. U22MC24100 (National Coordinating Center for the Regional Genetic Services Collaboratives, total award amount: $1,599,999) from the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, to the American College of Medical Genetics and Genomics.
KEY NCC/RC FINDINGS 2016-2017

- RCs worked with more than 4,070 providers; 416 consumers; and had 400 family and disease-specific organizations on their mailing lists.
- All RCs offered education and training sessions; the 382 sessions in the first nine months of year 5 reached nearly 7,500 participants.
- All RCs had websites that addressed the HRSA priorities of medical home, newborn screening, and long-term follow up. In the first nine months of 2017, these websites had 41,041 unique visits and 101,671 home page visits.
- All RCs and the NCC use social media; they reported having 1,960 followers as of February 28, 2017. Three RCs (NEGC, SERC and Region 4 Midwest) and the NCC use Twitter with 631 followers and four RCs (NYMAC, Region 4 Midwest, Heartland and WSGSC) and the NCC use YouTube with 8,532 video views in the first nine months of the fifth grant year.
- All RCs addressed medical home. Using the HRSA Performance Measure (PM) #41 Medical Home, the RCs recorded annual increases in their average scores from 18 in year 1 to 32 out of a possible 72 in year 5 – a 77% increase. The PM 41 scores ranged from 9 to 49 in 2017.

IMPACT OF THE NCC/RC ACTIVITIES

- The NEGC partnership with the Genetic Metabolic Center for Education (GMCE) entitles clinics in the New England region to free unlimited access to GMCE’s Metabolic Telehealth Service through May 31, 2017. GMCE has provided 97 consults in the New England area, with 63 consults focused on management, 18 on diagnosis, 13 on diagnosis and management, and 3 on counseling.
- Through the WSGSC Genetic Visit Outreach Outcomes, hundreds of families in Alaska and Hawaii have received genetic services for diagnosis, ongoing treatment and management. They would not otherwise have had access to genetic services.
- The NEGC-supported Genetics Materials for School Success (GEMSS) Resource Website for Schools, Families, and their Children (http://www.gemssforschools.org/) provides information on how to best support children with a specific genetic condition in a school setting. Dissemination efforts resulted in another year’s increase in reach of the GEMSS website (15,000 in 2014, 20,000 reached in 2015, 19,000 in the first 9 months of this grant year.
- The NYMAC Deafness and Genetics project created and distributed a patient-targeted handout and a provider-targeted handout. The provider material details the considerations for a genetics referral/consultation for deafness or hard of hearing children, such as when to refer, what is needed, and how to refer (including CPT codes for connexin testing).
- The Region 4 Midwest’s family forum produced videos and other educational resources including the Partnering with your Doctor: The Medical Home Approach Guide and the Journey through Diagnosis Guide (https://www.region4genetics.org/education/families/).
- Newborn Screening Parent Fact Sheets are developed, updated and maintained by the WSGSC. This website has 20,000 unique visitors per month.
- With American College of Medical Genetics and Genomics (ACMG)-approved ACT Sheets and algorithms, providers have the just-in-time information that they need to help their patients.
- 60,904 individuals accessed information on genetics and health, testing, and other genetic services through GenesInLife.org, with 4,175 individuals accessing links to support groups via DiseaseInfoSearch.org.
- NEGCN distributed 6,000 family health history booklets.
- A heritable condition response category was added to the National Survey of Child Health (NSCH). The question asks whether a doctor has told you that your child has a heritable condition. The survey was fielded in 2016 with data set to become available later in 2017. This NSCH variable will provide, for the first time, a population estimate of children affected by genetic conditions.

Also see Supplements 2, 3, and 4 for additional descriptions of NCC/RC activities.
Evaluation Measure: Value of a Regional Approach

The Regional Collaboratives (RCs) were created to support improved access to high quality genetic services at the local level. As HRSA-grantees, the RCs approach this goal by concentrating on the HRSA-defined priorities (FOA HRSA 12-138) The RCs work on these priorities within their regions (intra-regional activities), with other RCs (inter-regional activities) and at the national level through their work with the NCC and other national partners. Intra-regional activities allow the RCs to build coalitions that recognize and address the unique strengths and issues among the states in their region. Inter-regional activities build on the experiences of one region that can be exported to another region or regions, allowing innovation and expertise to spread quickly from one region to another. National activities take advantage of other national partners and emphasize themes that are common to all regions. With this layered approach, the NCC/RC system can be responsive to local issues and needs, take advantage of innovation and experience between regions, while addressing national priorities.

Table 1. Intra-/Inter-Regional Activities and National Activities (2016-2017)

<table>
<thead>
<tr>
<th>HRSA 12-138 Priorities for the RCs</th>
<th>Intra-regional*</th>
<th>Inter-regional**</th>
<th>National***</th>
</tr>
</thead>
<tbody>
<tr>
<td>Medical Home</td>
<td>7</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>NBS Capacity Building</td>
<td>16</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>Collaboration</td>
<td>15</td>
<td>9</td>
<td>16</td>
</tr>
<tr>
<td>Affordable Care Act</td>
<td>9</td>
<td>5</td>
<td>17</td>
</tr>
<tr>
<td>Long-term Follow-up</td>
<td>3</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>Transition from Pediatric to Adult Care</td>
<td>8</td>
<td>1</td>
<td>0****</td>
</tr>
<tr>
<td>Telegenetics</td>
<td>9</td>
<td>5</td>
<td>3</td>
</tr>
<tr>
<td>Other</td>
<td>4</td>
<td>4</td>
<td></td>
</tr>
</tbody>
</table>

* Intra-regional – RC works on these HRSA priorities with States and other organizations in their region.
** Inter-regional – RC works with other RCs on these HRSA priorities.
*** NCC and NGECN activities
**** NCC merged its Medical Home and Transition Workgroups 6/1/16
Source: Supplement 3 RC Intra-/Inter-Regional Activities

SELECT INTRA-REGIONAL ACTIVITY HIGHLIGHTS

- The NEGQ Quality Improvement (QI) Workgroup seeks to engage metabolic centers in the continuous quality improvement of patients with inborn errors of metabolism. One of the primary mechanisms supporting this initiative is a shared registry for individuals with DD/ID, PKU, or MCAD. Five clinic sites are addressing developmental delay and intellectual disability and 10 clinic sites for PKU or MCAD. Through November 2016, 2,116 individuals with DD/ID have been entered into the registry.

- As part of NYMAC’s NBS timeliness project, a survey was distributed to hospitals in the region to gather information about current NBS practices. Information gathered included the staff responsible for collecting and managing blood spots, packaging and sending of specimens, and barriers to the hospital sending blood spots within 24 hours of collection. One hundred and forty-nine hospitals have completed the survey from five states; approximately 50% of the hospitals in the region.

- The goal of the Heartland’s Hispanic Access Advisory Council (HAAC) project is to identify the barriers to accessing genetic services for persons of Hispanic origin and to identify strategies to support genetic service providers in providing culturally competent quality care. Interviews were conducted with 26 families across three states in the region. Eight recommendations were identified and the top three strategies were prioritized. The HAAC has begun to identify implementation activities to address these three priority areas with the goal of improving access to genetic services.
**SELECT INTER-REGIONAL ACTIVITY HIGHLIGHTS**

*Genetics Needs Assessment*

- As a part of the 2016 national needs assessment, the **Genetic Services Assessment (GSA)** tool developed by the Heartland RC was promoted to measure state-level public health programs in five domains: state capacity for services; access; clinical process and quality improvement; performance reporting/ improvement; and workforce. Through RCs’ efforts, **32 states have implemented the GSA tool** (past and current versions). This is an example of how a tool developed by one RC is adopted by others and used to improve public health genetics capacity (See Table 1). Heartland collected and compared GSA data over two time periods to frame a continuous improvement process and monitor progress.

- The **SERC Genetics Needs Assessment** was adapted from the Heartland Collaborative’s needs assessment survey. This survey was approved through the Emory IRB and was administered via SurveyMonkey. The SERC Genetics Needs Assessment targets state public health officials with the hope of impacting the funding and resources given to both short and LTFU clinics for NBS and other genetic conditions.

- **WSGSC** contacted each state Title V program to conduct assessment meetings or key informant interviews to obtain information regarding their perceived needs related to genetic services.

*Health Insurance and Access*

- **NEGC** fielded a survey to capture information on families’ experiences with health insurance, cost, affordability and barriers to care. This data was incorporated into a policy brief that was distributed to state legislators in the region. Four other RCs (NYMAC, Region 4 Midwest, MSGRC, WSGSC) adopted and adapted the survey to address access issues in their regions. The information collected in these surveys is being used to educate community leaders and shape policies/services in New England.

- Another NCC project, in partnership with Catalyst Center, developed the **State Statutes & Regulations on Dietary Treatment of Disorders Identified Through Newborn Screening**, which ascertains information about medical foods legislation in every state.

- The NEGC Health Care Access and Financing work group developed a series of resources aimed at providing families with the useful tools on navigating insurance appeals processes. Based on literature and website reviews, interviews with State insurance commissioners, and ongoing conversations with stakeholders, the group is developing a tip sheet for families, providing examples of effective appeals letters, handy info graphics, and links to a range of resources to improve family ability to access the coverage they need and deserve.

- The NCC Healthcare Access and Financing Workgroup continues to work in three subgroups: The Identifying Genetics Services subgroup conducting two webinars and is working on a white paper; the Coverage of Genetic Services subgroup collected Information about state Medicaid policies on genetic services; and the Consumer Role in Genetic Services subgroup created a Consumer Advocacy Resource Database.

*Mapping*

- **MSGRC** developed a mapping project to **assess access to a genetic care services**. Using the ACMG database, providers in the regions were identified and this information was overlaid with expected number of birth defects by county to estimate caseloads.

- In Region 4 Midwest, the **Michigan Newborn Screening mapping project was completed**. Results indicate that not just rural patients, but any genetics patient living at a great distance from their specialist provider may be consider underserved. The findings were presented at the 2016 Regional Meeting in October.

- MSGRC continued work on an evolving mapping project by conducting interviews with genetics providers to ask about provider capacity, delivery of outreach and telemedicine services, and...
geographic region served. 12 clinics in the region completed surveys. Mapping will be completed by the end of Year 5.

**Care Coordination**

- **Region 4 Midwest’s Care Coordination: Empowering Families** Training was translated into Spanish by Spanish-speaking trainers in the Heartland and Midwest RCs. Training curriculum and participant materials were reviewed for cultural competency and applicability for Spanish speaking population.

- **Inborn Errors of Metabolism Project Information System (IBEM-IS).** The focus of year 5 is data entry in the IBEM-IS - an ongoing collaboration with the Inborn Errors of Metabolism Center (IBEMC) and NIH-funded metabolic centers in national effort to collect data on metabolic disorders. Region 4 Midwest supports the Medical College of Wisconsin and the University of Louisville, Weisskopf Child Evaluation Center in data entry. Heartland and MSGRC are also participating.

**Longitudinal Pediatric Data Resource (LPDR)**

- Three states in the SERC region have met with individuals from NewSteps, APHL, and ACMG to discuss how to expand their current short-term follow-up (STFU) database to include data elements from the LPDR to capture the long-term follow-up (LTFU) public health data.

- In Region 4 Midwest, the states of Michigan and Minnesota moved forward with participation the LPDR Long-term Follow-up Pilot project through the NCC.

*Table 2. Inter-Regional Project Adoption (2012-2017)*
The NCC/RCs work with consumers, health and public health professionals and many organizations. In 2017 the RCs’ mailing lists had 4,070 providers; 416 consumers; and had 400 family and disease-specific organizations. Of these stakeholders, 379 providers and consumers participated in the RCs’ annual meeting as did 96 of the organizations. All of the RCs hold annual regional meetings that include public health genetics and NBS leaders, family advocates, genetic specialists, primary care representatives, LEND and Title V leaders convene. These meetings provide a forum for sharing information and developing regional priorities. Because NEGC and MSGRC held their annual meetings in April 2017, which was outside the reporting period for this fifth-year grant cycle, the data from these two RCs are not included.

As for trends over the past five years, more consumers have become involved in NCC/RC workgroups and advisory committees. More than 542 people and 126 organizations now serve on RCs’ workgroups and committees.

The NCC/RC staff participate on the Advisory Committee on Heritable Disorders in Newborn Children (ACHDNC).

Table 3. Inter-Regional Project Adoption (2012-2017)

*Because of the May 31, 2017 end to this grant cycle and with input from the HRSA Project Officer, the NCC/RC data in this report reflects nine months of program activity (June 1, 2016 to February 28, 2017)
SELECT INTRA- AND INTER-REGIONAL ACTIVITIES

• NCC, NEG, and RC staff continue to collaborate with the AUCD-LEND working group on genetics education for LEND programs. This year the national group is finalizing core curriculum in genetics for LEND programs.

• NYMAC engaged with NCHAM and EHDI in their hearing loss survey and in the development of consumer and provider materials. A survey of pediatricians was developed, distributed, and analyzed. The survey featured questions surrounding knowledge of the EHDI 1-3-6 guidelines, family referrals for hearing loss, and confidence speaking with families about genetic causes of hearing loss among others. Over 250 physicians completed the survey. Data were presented at the NYMAC Summit and EHDI meeting.

• The NYMAC Regional Genetics Education Network (RGEN) is an organization of volunteer patients, advocates, providers, genetic counselors willing to provide in-person or webinar-based education on genetics and newborn screening. There are currently 57 individuals registered for the RGEN who are available to speak about genetics and NBS topics. Three educational events have occurred.

• The SERC Consumer Alliance is a leader in nationwide advocacy efforts for recognizing medical foods as the primary nutritional treatment for inherited metabolic disorders. Their Consumer Alliance, in collaboration with the National PKU Alliance, American Academy of Pediatrics, and North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition, have been active in Authorization Act for Fiscal Year 2017, Sec. 714, reads: “Provides Tricare program coverage for medically necessary food and vitamins for digestive disorders and inherited metabolic disorders.”

• Using the NBS Connect registry infrastructure, SERC has identified groups to invite people to an inter-regional meeting entitled, “Tyrosinemia Type I: Making Connections Count Meeting” that it is hosting in Atlanta, GA. The purpose of this meeting is to connect families in the US to understand the clinical needs for medical nutritional management and give families and individuals with Tyrosinemia Type I the opportunity to interact with one another and with health professionals in the field of inherited metabolic disorders.

• Members of Region 4 Midwest’s Family Forum completed the development of a guide for families that have recently been diagnosed with a genetic condition. Entitled Journey through Diagnosis guide for families, the booklet provides helpful tips, information and insight for families of a recently diagnosed child(ren). Topics covered include managing emotions, finding support, family relationships and challenges, identifying resources and cultivating positive parent/professional relationships within a medical home. To date, 1,415 guides have been distributed in print and 438 have been downloaded from the Region 4 Midwest website.

• The Heartland Youth Advocate Mentoring has a goal of developing the advocacy skills in youth who have a genetic/chromosomal condition. Six mentors trained; mentees identified in OK, NE, and IA. Through their attendance at the annual Heartland regional meeting, these mentors and mentees met one another. More matching and mentoring is in process.
**Evaluation Measure: Public Information**

The NCC/RCs use a range of avenues to reach providers, consumers, and state public health agencies with information about genetic conditions and the resources they offer. In Year 5, all RCs have websites that addressed the HRSA priorities of medical home, NBS, and the ACA. In the first nine months of 2017, these websites had 41,041 unique visits and 101,671 home page visits. All RCs and the NCC use social media; they reported having 1,960 Facebook followers as of February 28, 2017. Three RCs (NEGC, SERC and Region 4 Midwest) and the NCC use Twitter with 631 followers and four RCs (NYMAC, Region 4 Midwest, Heartland and WSGSC) and the NCC use You Tube with 8,532 video views in the first nine months of the fifth grant year.

**Table 4. Unique Website Visits (excluding GEMSS and NGECN)**

<table>
<thead>
<tr>
<th>Year</th>
<th>Visits</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year 1</td>
<td>52,541</td>
</tr>
<tr>
<td>Year 2</td>
<td>83,776</td>
</tr>
<tr>
<td>Year 3</td>
<td>89,528</td>
</tr>
<tr>
<td>Year 4</td>
<td>81,460</td>
</tr>
<tr>
<td>Year 5*</td>
<td>41,041</td>
</tr>
</tbody>
</table>

*The Year 5 NCC/RC data in this report reflects nine months of program activity (June 1, 2016 to February 28, 2017)*

NYMAC, working with NCHAM, HRSA, HRSA Title V Region III, and EDHI, created and disseminated a plan for the providers’ handout that included: an upload on the NYMAC website, distribution to stakeholders through emails, NYMAC news and Facebook, advertisement in pediatrics journals, distribution at the EHDI conference, publication in the EHDI AAP newsletter, and distribution through NCHAM and EHDI coordinators networks. The plan for the patients’ handout included dissemination through Facebook, and NCHAM and EHDI coordinators.

The Medical Foods and Formula Toolkit was developed to be a resource for state health departments, advocates, patients, and their family members across the United States working towards better coverage of medical foods and formulas for individuals diagnosed with inborn errors of metabolism or other conditions. This online toolkit was developed by the Region 4 Midwest LTFU Workgroup.

Heartland provides recorded webinars on a variety of genetic-related topics for a broad spectrum of stakeholders. In 2016-7, Heartland added two additional videos (NORD 48 views and GARD 23 views) for a total of 12 webinars. The link is: [https://www.youtube.com/user/HeartlandGenetics](https://www.youtube.com/user/HeartlandGenetics).

The WSGSC has a newborn screening website ([www.newbornscreening.info](http://www.newbornscreening.info)) with parent fact sheets. That resource was utilized by 258,117 unique visitors and had 385,289 total visits. The WSGSC has Affordable Care Act through the Life Course Webpages – annually, hundreds of page views of the ACA pages in the Western states and throughout the nation reflect the interest in users learning about the impact and specifics of the ACA. Anecdotally, we have heard from many providers and family advocates who use and share the website with their patients or clients.
Evaluation Measure: Workforce Development, Consumer Education, and Training Resources

A major focus of the NCC/RCs is on consumer education and workforce development through the sponsorship of educational and training sessions. In nine months of the 2016-17 grant period, the NCC/RCs offered 382 education and training sessions, which reached nearly 7,500 participants. Compared with first-year data with nine months of year-five data, the number of session and number of participants in RC-sponsored consumer education and training has risen (Sessions n = 145 in Year 1 and 382 in Year 5; Participants n= 2,110 in Year 1 and n= 7,463 in Year 5). More than 81 percent of the sessions were delivered via webinars and teleconferences; the other 19 percent were done in-person. As for the content of these sessions, the RCs reported the following distribution of sessions by HRSA priorities for the NCC/RC system:

Table 5. Distribution of Education and Training Sessions by HRSA Priorities for the NCC/RC System

- 83 (23%) Treat in the context of medical home
- 32 (9%) Cultural competency and diversity in outreach projects
- 34 (10%) Expand the pool of the genetic service workforce
- 73 (21%) Build state public health department capacity
- 57 (16%) Strengthen public private partnerships
- 32 (9%) Collaborate and partner with HRSA MCHB-funded programs that promote the scaling up of effective practices
- 9 (2%) Improve insurance coverage policy and reimbursement-ACA Implementation
- 21 (6%) Other
Table 6. Distribution of Participants in NCC/RC Education and Training Sessions by HRSA Priorities

<table>
<thead>
<tr>
<th>Category</th>
<th>Participants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Treat in the context of medical home</td>
<td>672, 10%</td>
</tr>
<tr>
<td>Cultural competency and diversity in outreach projects</td>
<td>460, 7%</td>
</tr>
<tr>
<td>Expand the pool of the genetic service workforce</td>
<td>731, 10%</td>
</tr>
<tr>
<td>Build state public health department capacity</td>
<td>874, 13%</td>
</tr>
<tr>
<td>Strengthen public private partnerships</td>
<td>3006, 44%</td>
</tr>
<tr>
<td>Collaborate and partner with HRSA MCHB-funded programs that promote the scaling up of effective practices</td>
<td></td>
</tr>
</tbody>
</table>

- **NEGIC continued to support improvements in scope and depth of the GEMSS website by adding 1 new condition (Mitochondrial Disorders), and preparing 5 “Meet a Child” stories (Achondroplasia, Kabuki, Klinefelter, MECP2, Rubinstein-Taybi, and Smith-Magenis).**
- **Three Leadership Education in Neurodevelopmental Disorders (LEND) trainees** are integrated into three different NEGIC projects.
- The **NYMAC Genetics/Genomics Public Health (GPH) Fellowship** provided educational and leadership opportunities to 13 professionals and trainees from genetic counseling programs and LEND programs in the region.
- **SERC’s electronic Genetic Nutrition Academy (eGNA)** is a three-phase online case module series comprised of clinical case presentations via live webinars, research journal club, and web-based discussion forum facilitated by national experts. eGNA Case Conference was implemented on February 1, 2017 with more than 50 individuals attending the live webinar presentation.
- SERC held a lunch and learn event for healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students. In October 2016, the topic was “Medical Foods Insurance Coverage.” A total of 24 individuals participated in this in–person event, while 10 individuals participated via webex throughout the region. The pre/post-test showed a 9% increase in knowledge about medical foods insurance coverage.
- The **Newborn Screening: What Prenatal Care Providers Need to Know online course** was made available on Region 4’s website this reporting period. The on-line course teaches ways to heighten expectant parents’ awareness of newborn screening. In 2016-7, 50 prenatal care providers, child birth educators, and other health care professionals from across the U.S. took the online course.
- **The Diet for Life Metabolic Food and Formula Video Project** is a partnership of the Region 4 Midwest RC with the Michigan Department of Health and Human Services and the Michigan Family to Family Health Information Center. The 4- minute video that was produced addresses about the importance for individuals with metabolic conditions to stay on diet. The video is housed on the Region 4 Midwest YouTube page.
**Evaluation Measure: Newborn Screening (NBS) Capacity Building/Emergency Preparedness**

The NCC/RCs engage in a number of activities to build newborn screening capacity and to promote the recommendations of the Advisory Committee on Heritable Disorders in Newborn Children (ACHDNC). Through technical assistance, webinars, RC websites, and other means, State public health departments’ capacity to address new conditions of critical congenital heart disease (CCHD) and Severe Combined Immunodeficiency (SCID) was expanded and parents and clinicians were given up-to-date information.

*Table 7. State and Territorial Adoption of the Recommended Uniform Screening Panel for Critical Congenital Heart Disease (CCHD) and Severe Combined Immunodeficiency (SCID) February 2017*

**SELECT INTRA- AND INTER-REGIONAL ACTIVITIES**

**NEGC**
- Following a successful screening project across 8 birthing centers in Maine, New Hampshire, Vermont, Rhode Island and Connecticut, NEGC continues to support an electronic resource with psychosocial and other supports for families affected by CCHD. The website ([www.necongenitalheartresources.org](http://www.necongenitalheartresources.org)) went live in May 2016. During the current project year, 81 unique users visited the project page.

**SERC**
- Through a collaboration with GMDI members, parent organizations, and AHRQ, guidelines for MSUD and PKU have been released on the project’s web based portal. Propionic Acidemia guidelines are currently being reviewed and will be published in 2017. PKU guidelines were accepted and released through AHRQ.

**Region 4 Midwest**
- The Hemoglobinopathies Workgroup has drafted a manuscript outlining the follow-up recommendations and the process of development for Short –Term Follow-Up Recommendations for Sickle Cell Trait follow-up.
• The Newborn Screening Laboratory Technical Assistance and Resource Sharing forum facilitated technical assistance among the Region 4 states newborn screening laboratories regarding the recall of CF testing kits and implementation of new testing methods, Pompe and MPS1 screening implementation, using HL7 messaging for newborn screening results, and implementation NBS into state health information exchanges.

• The MI Newborn Screening mapping project was completed. Results indicate that not just rural patients, but any genetics patient living at a great distance from their specialist provider may be consider underserved. The findings were presented at the 2016 Regional Meeting in October. Planning has begun for seven-state baseline maps of underserved populations to be completed by May 31, 2017.

Heartland
• The Iowa NBS program received an award from NewSTEPs for the planning and implementation of a Collaborative Improvement and Innovation Network (CoIIN) for timeliness in newborn screening two years ago. Heartland supported additional states in the region to convene and extend the CoIIN project across the region (Kansas, Iowa, Arkansas, Missouri, North Dakota, and Oklahoma). The rationale was to enable programs to engage in quality improvement activities through shared learning of evidence-based strategies for improving timeliness within their program’s newborn screening system. States convened a CoIIN team of newborn screening stakeholders and met in Kansas City May 5-6, 2016. NewSTEPs facilitator presented on the quality improvement process, states reported on the specific section(s) of Quality Indicator 5 that pertains to their state project, and states shared barriers and strategies. All participants ranked the training as valuable or very valuable. After the meeting, Iowa, Kansas, Nebraska, North Dakota, and Oklahoma received awards under NewSTEPs 360 grant to improve timeliness in their NBS programs.

MSGRC
• Dr. Kathryn Hassell leads the MSGRC Hemoglobinopathies Interest Group (HIG). Dr. Hassell continues to serve on the Sickle Cell Guidelines Expert Panel Membership at the National Heart, Lung, and Blood Institute. While the final guidelines were released in 2014, MSGRC has continued to disseminate guidelines to its members in 2016-2017. The HIG is planning a meeting for April 2017 as part of the MSGRC Annual Meeting. Approximately twenty MSGRC members attended the April 2016 in-person HIG meeting. Feedback on dissemination and usefulness of the new guidelines was solicited from attendees.

• Dr. Janet Thomas directs the Metabolic Consortium (MC) which meets in-person every year to discuss patient care guidelines, case studies, and other regional developments. The MC continues to be a forum for distribution and feedback on the previously developed shared datasets. Members have the opportunity to bring case studies for discussion. Quality of newborn screening within the region is discussed including false negative tests or unique situations, new pilot disorders for population screening, and second tier laboratory testing.

WSGSC
• A Newborn Screening Survey was done in HI and WA to obtain parents’ opinions about notification preferences. The data analysis and a report of findings in HI and WA was completed.
Evaluation Measure: Long-Term Follow-Up (LTFU)
The NCC in partnership with the Newborn Screening Translational Research Network (NBSTRN), developed a set of questions and answers and a tool to support longitudinal data collection. This tool, the Longitudinal Pediatric Data Resource (LPDR), is being used by a number of research studies. Currently, NCC is working with public health teams in 14 states to use the LPDR.

SELECT INTRA- AND INTER-REGIONAL ACTIVITIES

LPDR:
- Three states in the SERC region have met with individuals from NewSteps, APHL, and ACMG to discuss how to expand their current short-term follow-up (STFU) database to include data elements from the LPDR to capture the long-term public health data. In Region 4 Midwest, the states of Michigan and Minnesota moved forward with participation in the LPDR.

NYMAC
- A six-part NBS timeliness webinar series was attended by over 500 participants. Topics included analytic timeliness, families affected by NBS, and NBS recommendations, among others.
- A video on specimen collection and timeliness in NBS was created, piloted in hospitals, shared with other RCs through the NYMAC website, and publicized through Facebook and NYMAC News. A “test your knowledge survey” was taken by 87 people before watching, and 61 people after watching.
- A NBS meeting on “Causes and Solutions of Delayed Diagnosis” was held with 50 participants representing state NBS programs’ laboratory and follow-up, specialty care providers, primary care providers, consumers, and national partners.
- The NewSTEPs activities included a two-phase survey regarding hospital practices and electronic capabilities. One hundred and forty-nine hospitals have completed a survey about current NBS practices. Next steps include distributing a survey to hospitals in the region about electronic capabilities including readiness to adopt health information technology strategies.

Region 4 Midwest
- A manuscript entitled, “Congenital Hypothyroidism 3-Year Follow-Up Project: Results from the Region 4 Midwest Genetics Collaborative,” is under review with Pediatrics. This work by Kupper, Wintergerst, and partners in all Region 4 Midwest states identifies the 3-year follow-up management and education patterns of primary care clinicians and pediatric endocrinologists for children diagnosed with congenital hypothyroidism by newborn screening programs.
- The Inborn Errors of Metabolism Information System (IBEM-IS) project’s purpose is to develop a LTFU database and to track the treatments, health, and developmental outcomes of the patients with inborn errors of metabolism. The focus of year 5, data entry in the IBEM-IS, an ongoing collaboration with the Inborn Errors of Metabolism Center (IBEMC) and NIH-funded metabolic centers in national effort to collect data on metabolic disorders. Region 4 Midwest supports the Medical College of Wisconsin and the University of Louisville, Weisskopf Child Evaluation Center in data entry. Heartland’s and Region 4 Midwest’s participation in these pilots helps facilitate the states’ adoption of case definitions for the recommended uniform screening panel. MSGRC funds Dr. Janet Thoma’s research protocol to consent children and families diagnosed with conditions via newborn screening into long term follow-up in association with the IBEM-IS.

WSGSC
- CPT1 Arctic Variant Community Project is a unique opportunity for families in the region living with CPT1 Arctic variant to understand their risks as well as treatment and management of this regional genetic condition. Public health geneticists and researchers may use this project as a model for improving understanding and treatment for rare genetic disorders. Dave Koeller, MD from the Oregon Health and Science University, the Alaskan Native Health organizations, Alaskan Native tribal groups, and the Alaska Department of Health are involved. Dr. Koeller was able to take his work with the WSGSC to apply for and be awarded a NIH grant to do the long-term follow-up project.
**Evaluation Measure: Access to Care—Medical Home**

The NCC/RC system is focused on increasing access to genetic services through medical homes and improved care coordination. All RCs were engaged in medical home activities in Year 5. The RCs use the HRSA Performance Measure (PM) #41 in this annual progress report to HRSA. Nine months of year-five data show there has been a steady rise in the RCs’ report of medical home activities on the PM #41 from an average score of 18 in year one to 32 out of a possible 72 in year five – a **78% increase**. The PM 41 scores ranged from 9 to 49 in 2017.

**Table 8. RC Self-Report on Medical Home Activities using HRSA PM #41**

*The Year 5 NCC/RC data in this report reflects nine months of program activity (June 1, 2016 to February 28, 2017)*

**SELECT INTRA- AND INTER- REGIONAL ACTIVITIES**

- The NEGC Medical Home Work Group sponsored the creation of a new webinar focused on bringing families and professionals closer together. The webinar is entitled, *"Family Voices: Helping Families & Professionals Maneuver the Maze of Complex Conditions"*.
- Two quality improvement work groups, established by the NEGC, continue to work on documenting and sharing clinical care processes and outcomes for children with developmental delay or intellectual disability as well as for children living with PKU or MCAD deficiency. Five clinic sites participate on the DD/ID workgroup (entering data) and 10 participate on the PKU / MCAD workgroup. Through November 2016, **2,116** individuals have been entered into the registry.
- The NEGC Health Care Access and Financing work group has worked on developing a series of resources aimed at providing families with the useful tools on navigating insurance appeals processes. Based on literature and website reviews, interviews with State insurance commissioners, and ongoing conversations with stakeholders, the group is developing a tip sheet for families, providing examples of effective appeals letters, handy info graphics, and links to a range of resources to improve family ability to access the coverage they need and deserve.
- In collaboration with genetic dietitians throughout the mid-Atlantic region, NYMAC developed a **web-based survey for parents/caregivers of persons with PKU, and/or persons with PKU**. The overarching theme of the survey was history of medical formula and low protein food usage, and the availability and challenges to accessing medical formula and low protein foods, particularly surrounding costs, insurance coverage, and sources of financial assistance. The results of the survey were analyzed and shared at the NYMAC Summit and with the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children. The results will be published, and results will be used to create advocacy materials and create resources to help individuals and families obtain coverage.
• Using the Care Coordination: Empowering Families Training, the Region 4 Midwest staff and facilitations trained an additional 84 caregivers in seven trainings offered in collaboration with the Michigan (MI) Department of Health and Human Services, MI Family to Family, and the Pacer Center in Minnesota (MN). The Care Coordination: Empowering Families Training Insurance Module includes information about the Affordable Care Act as it relates to CYSHCN. It includes the Catalyst Center’s “The Affordable Care Act: A side-by-side comparison of major provisions and the implications for children and youth with special health care needs.”

• Heartland teamed with the University of South Dakota (USD) Center for Disabilities to design a project to improve the individual healthcare plans IHP process. This team trained ten participants, (six school nurses, two program nurses, one nurse supervisor and one state Department of Education representative) on effective methods for creating and implementing IHPs. Throughout the school year, those ten participants trained nearly 400 additional school nurses in the IHP process. The participants have also helped develop over 4,800 IHPs since the initial training.

• The MSGRC Parent Partner Project trains parents of children with special healthcare needs to assist other families with special needs to navigate the non-medical components of the medical home. The Parent Partner Project is currently in eight pediatric practices and is expanding to an Indian reservation site and Air Force base site, both in Wyoming. The project in Montana is now completely funded out of the state’s CYSHCN budget. A second evaluation was undertaken in 2015-2016 to measure the impact the program is having on family’s abilities to identify and access resources.

• The NCC Healthcare Access and Financing Workgroup, working in partnership with regional collaborative state-based leads and respective Medicaid programs, gathered state level Medicaid policy information for the reimbursement and coverage of genetic testing and genetic services.

• Patient navigation is an intervention that integrates case management and peer support to address obstacles to care. On August 2, 2016, the NGECN conducted a webinar on Patient Navigation: Care Coordination Meets Peer Support. NGECN produced a monograph entitled Next Steps toward Patient Navigation Perspectives from Partners in Care that will help promote this critical component of care coordination.
Evaluation Measure: Transition from Pediatric to Adult Care

In 2016-7, five RCs engaged in activities related to transition from pediatric to adult care:

**NEG C**
- The “Health Survey in Adults” continues to collect data, with 145 respondents to date, 93 of whom have or may be impacted by a metabolic disorder, and 52 who do not have a metabolic disorder.
- The NEGC continues to support the Face Forward’s efforts to educate and train youth in becoming more effective self and group advocates. **Twenty-five youth participated in the 2016 camp** including participants sponsored by the Heartland RC.
- Dr. Susan Waisbren continues to support local initiatives for improved community and advocacy among those with genetic conditions. As a part of this effort, Dr. Waisbren continues to support a young adult's efforts to coordinate a homocystinuria/metabolic support group. 4 participants joined in the last meeting on 6/18/16.

**NYMAC**
- At the 2016 NYMAC Summit meeting in Baltimore, MD, a session was dedicated to transition. There were two learning objectives, 1: “Explain models of transition from pediatric to adult medical care for individuals with genetic disease using sickle cell disease as an example.” And 2. “Discuss the interplay between a primary care provider and a specialist in the transition care process.” NYMAC resources regarding transition were distributed to attendees.

**SERC**
- The SERC Medical Home and Transitioning Work Group with the SERC Consumer Alliance developed a **Delphi study on the healthcare transitioning process**. The study has been designed to achieve a consumer perspective on transitioning. As of March 1, 2017, 20 participants completed the verbal informed consent process. The short-term goal is to have a sample size of 50 participants for the first Delphi survey, and conduct up to three rounds of the survey. The study team plans to publish and disseminate the survey results among healthcare professionals to guide the development of stronger, patient-centered transition programs for emerging adults with special health care needs.

**Region 4 Midwest**
- The transition module of the **Care Coordination: Empowering Families** training emphasizes the need to plan for transition for children with genetic conditions. Participants with older children regularly cite this module as informative and important.
- The region worked in partnership with the Michigan children’s special health care services program and Michigan family to family health information center, and the result was the Children’s Special Health Care Services **Online Transition Course with six modules** of information and resources geared towards parents and young adults with special health care needs including genetic conditions.

**Heartland**
- Heartland’s **mentoring project develops advocacy skills in youth** who have a genetic/chromosomal condition. A core activity of the project developed and implemented a mentoring curriculum in collaboration with the Midlands Mentoring Partnership, a member of the national MENTOR network. Mentoring was initiated in January 2017 with mentoring agreements being signed and goals for each youth being Healthcare Transition for individuals with genetic conditions established.
- The purpose of the Healthcare Transition for individuals with genetic conditions project was to determine the **role of the genetics team in healthcare transition**. In the current project year, the transition clinic and curriculum have continued to be implemented and refined by all of the work has been conducted in South Dakota. A total of 13 trainees (4 residents, 1 genetic counselor, and 8 genetic counseling students) completed the curriculum. Nine patients served in the clinic.
Cluster Measure: Telegenetics/Distance Strategy
To improve access to genetic services, all seven RCs began pursuing telegenetics and distance strategy projects in 2015 - 2016 and continued with additional activities during the fifth year of the grant cycle. As a part of the RC needs assessment activities, information was gathered about existing telegenetics resources. The following data about known sites should be regarded as an initial assessment and used with caution in that it was collected through various methodologies and at different points in time. The NCC offered the following definitions in the instructions for reporting telegenetic sites. Telegenetic sites are defined as follows:
• the originating site is where the patient is seen from;
• the distant site is where the specialist is based; and
• patient encounters via telephone should not be considered telegenetic services.

The NCC/RC evaluators consider this information as a start point for additional discussion about creating a set of common measures about telegenetic services.

<table>
<thead>
<tr>
<th>Table 9. Sites Doing Telegenetics Services</th>
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</thead>
<tbody>
<tr>
<td><strong>Regional Collaborative</strong></td>
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<tr>
<td>-------------------------------</td>
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<tr>
<td>NEGC</td>
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<tr>
<td>NYMAC*</td>
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<tr>
<td>SERC</td>
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<tr>
<td><strong>Region 4 Midwest</strong></td>
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<tr>
<td>Region</td>
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<td>Heartland</td>
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<table>
<thead>
<tr>
<th>Region</th>
<th>Activity and/or Site Information</th>
<th>Date</th>
<th>Activities and Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heartland</td>
<td>Distant - Where the provider is: 16</td>
<td>October 2016-February 2017</td>
<td>Structured telephone survey of clinic staff Email outreach to providers</td>
</tr>
</tbody>
</table>

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<thead>
<tr>
<th>Region</th>
<th>Activity and/or Site Information</th>
<th>Date</th>
<th>Activities and Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>MSCRC</td>
<td>Colorado – 1 distant site to 6 sites</td>
<td>October 2016-February 2017</td>
<td>Email outreach to providers</td>
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<tr>
<td></td>
<td>Montana -3 distant sites to 20 sites</td>
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<tr>
<td></td>
<td>Texas – 1 distant site to 5 sites</td>
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<tr>
<th>Region</th>
<th>Activity and/or Site Information</th>
<th>Date</th>
<th>Activities and Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>WSGSC</td>
<td>Alaska – No telegenetics</td>
<td>March 2017</td>
<td>Contacted the Northwest Regional Telehealth; the California Telehealth Resource Centers; the California Department of Public Health; and the Southwest Telehealth Resource Center and searched the ACMG online database of genetic centers and surveyed contacts at those centers regarding telemedicine use.</td>
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<tr>
<td></td>
<td>California 1 distant to 1 originating site</td>
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<td></td>
<td>Guam – none known</td>
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<tr>
<td></td>
<td>Hawaii – 1 distant site on Oahu and 5 remote sites on neighbor islands</td>
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<td></td>
<td>Idaho – No telegenetics</td>
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<tr>
<td></td>
<td>Oregon - 1 distant to 1 originating site</td>
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<tr>
<td></td>
<td>Washington – 4 distant to 9 originating sites &amp; patients’ homes</td>
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*The NYMAC needs assessment did not use the terms “originating site” and “distant” site as defined by the NCC. The NYMAC Survey also found that residents in the region were served by genetic providers from other states.*
SELECT INTRA- AND INTER-REGIONAL ACTIVITIES

- Via the NEGc partnership with the Genetic Metabolic Center for Education (GMCE) clinics in the New England region have free unlimited access to GMCE’s Metabolic Telehealth Service until May 31, 2017. To date, the GMCE has provided 97 consults in the New England area, with 63 consults focused on management, 18 on diagnosis, 13 on diagnosis and management, and 3 on counseling.
- The Telegenetics Landscape Survey garnered 54 responses from unique genetics programs that serve patients in the NYMAC region. Respondents answered questions surrounding their staffing, technology use, funding, and barriers and benefits to implementation among other topics.
- Over 100 providers have joined the NYMAC Telegenetics Community of Practice (TCOP). The TCOP online community gathers resources, webinars, and other information for those who are currently practicing or are interested in practicing telegenetics.
- At the annual telegenetics meeting in the Southeast region, presentations ranged from nuts-and-bolts information on setting up a new telegenetics clinic to patient testimonials about using social media in healthcare, and a presentation on training clinicians and genetics counselors for telegenetics. There was a demonstration of the doxy.me software developed by a SERC member and is freely available for use on any basic digital media (e.g., laptop, smartphone, tablet).
- Region 4 Midwest provided support for three regional representatives to attend and participate in the Telegenetics Training at the South Central Telehealth Forum 2016. Done in partnership with the Western States and Heartland Genetics Collaboratives, South Central Telehealth Resource Center, this opportunity allowed the Midwest to train providers in telegenetics and to observe the curriculum for future support and implementation in the region.
  - Heartland telegenetics project educated genetics residents, genetic counseling students and those in practice. Using a curriculum that was co-developed with the Western States, the training involved 14 participants, which included physicians, genetic counselors, nurse practitioners, and a genetic counseling trainee with participants from SERC, Region 4 Midwest, and Mountain States. MSGRC conducted a case study of a successful telegenetics program to identify elements and resources needed for developing telegenetics. This work was accepted as an oral presentation at the ACMG annual meeting.
  - MSGRC continued work on an evolving mapping project by conducting interviews with genetics providers to ask about provider capacity, delivery of outreach and telemedicine services, and geographic region served. Surveys were completed in 12 clinics in the region. Mapping will be completed by the end of Year 5.
  - MSGRC completed an in-depth case study of the telegenetics service line at Cook Children’s Hospital in Fort Worth, TX. Data collection was completed via telephone interviews and email transmission of quantitative data. The case study was accepted for an oral presentation at the 2017 ACMG Annual Meeting (Dr. Janet Thomas presenting).
  - The WSGSC developed ten new on-line modules for the telegenetics training. These were reviewed by the Heartland Regional Genetics Collaborative and Southwest Telehealth Resource Center.
  - Two groups of genetic counselors attended the Telegenetics Training in August 2016, one in Tucson, AZ and one in Kent, WA. Another telegenetics training for a group of WA program administrators was also held in Kent, WA. The next round of training is planned for April 2017.
  - Five clinicians from MSGRC attended the telegenetics training in Nashville, TN sponsored by Heartland Regional Genetics Collaborative. Attendees include 1 geneticist, 1 medical genetics resident, and 3 genetic counselors.
  - The WSGSC evaluator completed a literature review and wrote a framework for an approach to measure health outcomes of genetic services, including information specific to telegenetics. This work has laid the foundation for a process of expert development of consensus outcome measures under the auspices of the NCC Telegenetics Workgroup.
## Supplement 1: Summary of RC Priorities - March 31, 2017

<table>
<thead>
<tr>
<th>Priorities</th>
<th>Region 1-NEGC</th>
<th>Region 2-NYMAC</th>
<th>Region 3-SERC</th>
<th>Region 4-Midwest</th>
<th>Region 5-Heartland</th>
<th>Region 6-MSGRC</th>
<th>Region 7-WSGSC</th>
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<tbody>
<tr>
<td>Treat in the context of a medical home</td>
<td>X</td>
<td>X</td>
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<tr>
<td>Cultural competency and diversity in outreach projects</td>
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<td>X</td>
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<td>Expand the pool of the genetic service workforce</td>
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<td>Build state public health department capacity</td>
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<td>Strengthen public-private partnerships</td>
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<tr>
<td>Collaborate and partner with HRSA MCHB-funded programs that promote the scaling up of effective practices</td>
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<tr>
<td>Improve insurance coverage policy and reimbursement</td>
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<tr>
<td>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.</td>
<td>X</td>
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<tr>
<td>Address emergency preparedness</td>
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<td>Others not in RFA</td>
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How much did you do?

- **NEGC staff continued to support efforts aimed at improving the knowledge base in the field. During 2016/2017, staff were involved in 1 peer reviewed publication and 8 invited oral presentations.**

- **GEMSS: Continued to support improvements in scope and depth of the GEMSS website by adding 1 new condition (Mitochondrial Disorders), and preparing 5 “Meet a Child” stories (Achondroplasia, Kabuki, Klinefelter, MECP2, Rubinstein-Taybi, and Smith-Magenis). The resource was disseminated to a range of audiences locally and nationally via traditional mechanisms, social media, and continued development of collaborative relationships with external partners (such as Pediatric Societies in New England, PT/OT/Speech faculty in New England; and Iowa State Nurses Association).**

- **QI: Two quality improvement work groups, established by the NEGC, continue to work on documenting and sharing clinical care processes and outcomes for children with developmental delay or intellectual disability as well as for children living with PKU or MCAD deficiency. Five clinic sites participate on the DD/ID workgroup (entering data) and 10 participate on the PKU / MCAD workgroup.**

- **Medical Home: Primary care providers, and associated health care professionals who attended the NEGC Medical Home Work Group sponsored the creation of a new webinar focused on bringing families and professionals closer together. Entitled, "Family Voices: Helping Families & Professionals Maneuver the Maze of Complex Conditions", this unique webinar will be provided on March 30, 2017.**

- **NEGC / LEND: Three Leadership Education in Neurodevelopmental Disorders (LEND) trainees are integrated into three different NEGC projects - one is focused on assisting with the evaluation of the NEGC, the second is supporting the Genetics Education Materials for School Success (GEMSS) website, and the third is collaborating with the HAF workgroup to develop resource materials on appeals processes for families seeking coverage for their children with special health care needs. NEGC continues to collaborate with the AUCD-LEND working group on genetics education for LEND programs. This year the national group is finalizing core curriculum in genetics for LEND programs.**

- **The Health Care Access and Financing work group has worked on developing a series of resources aimed at providing families with the useful tools on navigating appeals processes. Based on literature and website reviews, interviews with State insurance commissioners, and ongoing conversations with stakeholders, the group is developing a tip sheet for families, providing examples of effective appeals letters, handy info graphics, and links to a range of resources to improve family ability to access the coverage they need and deserve.**

- **Transition:**
  - The “Health Survey in Adults” continues to collect data, with 145 respondents to date, 93 of whom have or may be impacted by a metabolic disorder, and 52 who do not have a metabolic disorder.
  - The NEGC continues to support the Face Forward’s efforts to educate and train youth in becoming more effective self and group advocates. Twenty-five youth participated in the 2016 camp.
  - Dr. Susan Waisbren continues to support local initiatives for improved community and advocacy among those with genetic conditions. As a part of this effort, Dr. Waisbren continues to support a young adult's efforts to coordinate a homocystinuria/metabolic support group. 4 participants joined in the last meeting on 6/18/16.

- **CCHD: An electronic resource with psychosocial and other supports for families affected by CCHD was developed (www.necongenitalheartresources.org) and went live in May 2016. During the current project year, 81 unique users visited the project page.**
Genetic Metabolic Center for Education (GMCE): The NEGC continued its collaboration with the GMCE this year that seeks to directly support the diagnosis and treatment of inherited metabolic disorders throughout New England.

How well did you do it?

• GEMSS: Conditions added to the GEMSS website reflect the input of professionals in the field, major advocacy organizations, as well as input received via the project website.
  o Broadened effective distribution of GEMSS via use of Parent Ambassadors, dedicated Facebook pages; publications about GEMSS in various newsletters; presentations at major events, and collaboration with partners involved in similar missions.
  o Dissemination efforts resulted in another year’s increase in reach of the GEMSS website (15,000 in 2014, 20,000 reached in 2015, 19,000 in the first 9 months of Year 5)

• QI: The quality improvement learning work groups have recruited participation by 5 clinic sites for addressing developmental delay and intellectual disability and 10 clinic sites for PKU or MCAD. Through November 2016, 2,116 individuals have been entered into the registry.

• NEG/LEND: Three Leadership Education in Neurodevelopmental Disorders (LEND) trainees are successfully implementing three distinct projects. Presentations on these efforts will be made in the last quarter of the 2017 project year.

• HAF: The resources being developed to improve families' ability to successfully navigate insurance appeals processes has been fully supported by the diverse members of the HAF group and has been identified as a needed resource in the region.

• Transition:
  o To date, the Adult Health Survey has been administered to 93 who have or may be impacted by a metabolic disorder, and 52 who do not have a metabolic disorder.
  o The FaceForward Summer Camp Experience: 25 youth participated in a unique weekend experience combining technology, the arts, group and individual development.

• GMCE: The NEGC has continued to publicize its partnership to stakeholders in multiple forums (email blasts, meetings and presentation, website) that described the availability of the GMCE resource and the benefits for joining as a member of the NEGC. As a result, 3 sites are participating in metabolic consultations in 2017.

Was anyone better off?

• GEMSS: During the past nine months, nearly 19,000 people visited the GEMSS website (http://www.gemssforschools.org/) to learn about the multiple ways for supporting children with genetic health conditions to succeed in school. Since its launch in February of 2012, GEMSS has served over 81,000 users from across the world - 53,000 from the US and 28,000 from 188 countries.
  o Past survey work (from our 2013/2014 program year) (N=74) indicated that 75% or more find the site easy to use, 63% looking for specific information found most or all the information they were looking for, 54% found the information to be very useful and 39% somewhat useful, and 80% agreed that they would use the website in the future to find further information about educating children with genetic conditions.

• QI: The aim of the QI work group is to improve care. Five centers are currently involved in entering data and over 2,000 individual patient records are included in the QI registry. The QI group does not have patient-specific outcomes given the level of funds to support this activity. The assumption is that improving care processes improves outcomes for all such patients.

• NEG/LEND: Trainees involved in the NEG/LEND projects produced a range of useful materials, including a summary of appeals recommendations for families, an analysis of stakeholder perspectives on the NEG, and substantive new information for the GEMSS project that will lead to multiple improvements in this critical resource.

• GMCE: To date, GMCE has provided as provided 97 consults in the New England area, with 63 consults focused on management, 18 on diagnosis, 13 on diagnosis & management, and 3 on counseling.
New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) Highlights 2016-17

Supporting the Continued Improvement of Newborn Screening (NBS) and Emergency Preparedness

1. A six-part NBS timeliness webinar series was attended by over 500 participants. Topics included analytic timeliness, families affected by NBS, and NBS recommendations, among others.

2. A video on specimen collection and timeliness in NBS was created, piloted in hospitals, shared with other RCs through the NYMAC website, and publicized through Facebook and NYMAC News. A “test your knowledge survey” was taken by 87 people before watching, and 61 people after watching.

3. A NBS meeting on “Causes and Solutions of Delayed Diagnosis” has been scheduled for 50 participants representing state NBS programs’ laboratory and follow-up, specialty care providers, primary care providers, consumers and national partners.

4. The NewSTEPs activities included a two-phase survey regarding hospital practices and electronic capabilities. One hundred and forty-nine hospitals have completed a survey which gathered information about current NBS practices. In the next phase, a survey will be distributed to hospitals in the region regarding electronic capabilities including readiness to adopt health information technology strategies.

Addressing Primary Care Linkages and Strategies

1. The Telegenetics Landscape Survey garnered 54 responses from unique genetics programs that serve patients in the NYMAC region. Respondents answered questions surrounding their staffing, technology use, funding, and barriers and benefits to implementation among other topics.

2. Over 100 providers have joined the Telegenetics Community of Practice (TCOP). The TCOP online community gathers resources, webinars, and other information for those who are currently practicing or are interested in practicing telegenetics.

3. TCOP Telegenetics webinars to date have included 1) an introduction and summary of the telegenetics landscape survey findings, 2) conducting a needs assessment for telegenetics, 3) Care coordination by telegenetics, and 4) Business planning and financial models for telegenetics.

4. Funding was made available for 7 applicants and 6 speakers from the NYMAC region to attend the Mid-Atlantic Telehealth Resource Center (MATRC) Telehealth Summit in Leesburg, VA. Seventy individuals to date have registered for a pre-summit session on telegenetics.

Addressing the Capacity of the Public Health Programs

1. The NYMAC Genetics/Genomics Public Health (GPH Fellowship provided educational and leadership opportunities to 13 professionals and trainees in the region. Three webinars were offered to GPH Fellows, 1) access to care and care coordination, 2) the role of genetic counselors
in public health, and 3) patient-centered communication, advocacy and cultural humility, and discussion forums were created for fellows.

2. A Hearing Loss-Genetics Needs assessment collected responses from over 250 physicians; the survey featured questions surrounding family referrals for hearing loss, confidence speaking with families about genetic causes of hearing loss, and the use of connexin testing and tracking of failed newborn screening. A poster presenting the data won “best poster” at the EHDI 2017 conference in the EHDI workforce division.

Addressing System Issues Including the Organization of Care, Coverage, Reimbursement and Payment

1. A PKU and Medical Foods Survey received 320 unique responses from adults with PKU, or parents/guardians of a child with PKU. Survey development was guided by a dietitian workgroup; members of the workgroup are now guiding the interpretation of the data.

Strengthening NYMAC’s Capacity

1. The Regional Genetics Education Network remains active with 57 genetic specialists available to speak on genetics and NBS topics
2. Over 1,000 autism related groups were identified for education and outreach and were sent an e-mail offering educational programs on Fragile X. A follow-up e-mail was sent to 325 groups specifically in NJ, where there is greater incidence of diagnosis at an older age. Over 115 health providers (notably, speech, occupational, and physical therapists) have participated in these programs to date. Of the educational programs, two in-person presentations focused on the characteristics of fragile X syndrome, as well as diagnoses, testing, and benefits of early intervention.
The electronic Genetic Nutrition Academy (eGNA) is a three-phase online case module series comprised of clinical case presentations via live webinars, research journal club, and web-based discussion forum facilitated by national experts. The goal of the eGNA is to increase global clinical knowledge about nutrition management and inherited metabolic disorders (IMDs) among genetic metabolic dietitians, nutritionists, and other clinicians and translates this knowledge to inform patient care. For the first year of the pilot project for the eGNA Case Conference, the focus will be on Medical Nutrition Therapy (MNT) education for Urea Cycle Disorders (UCDs). The pilot phase of Phase I for the eGNA Case Conference was implemented on February 1, 2017. More than 50 individuals attended the live webinar presentation. A total of 42 participants completed the pre-test, post-test and course evaluations and earned 1 CEU. There was a 49% increase in score from pre-test to post-test. The presentations are recorded and will be archived in the eGNA Online Library for participants to access later. Two more eGNA Case Conferences are scheduled for this year.

SERC lunch and learn event was organized to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students. The event held in October 2016 focused on the topic of “Medical Foods Insurance Coverage.” A total of 24 individuals participated in this event in-person, while 10 individuals participated via webex throughout the region. The Pre/Post-test administered at this event showed a 9% increase in knowledge about Medical Foods Insurance Coverage among the participants. A second lunch and learn event is planned for April 5th, 2017 and will focus on The Longitudinal Pediatric Data Resource: Accelerating New Knowledge Discovery and Public Health Surveillance in Genetic Disorders.

The SERC SERGG Annual Meeting 2016 was organized to build on and enhance the work of SERC. Annual meetings are held for face-to-face communication between the chairs and participants of the core activities as well as collaborative network building with local, statewide regional and national partners. The 2016 annual meeting was held from July 14–16th, 2016 in Ponte Vedra Beach, Florida. The meeting was attended by 122 healthcare professionals with an interest in genetics and public health. Twelve consumers also attended. Thirty platform and twenty one poster presentations were made covering current issues in all areas of genetics: molecular genetics, cytogenetics, biochemical genetics and clinical genetics. Highlights of the meeting included newborn screening for GAMT deficiency, a live telemedicine demonstration, a report on results from the genetic needs assessment survey and another on the Registry and Surveillance System for Hemoglobinopathies (RuSH) project. Other topics covered were genomics and newborn screening, metabolomics, national nutrition management guidelines, regional collaborative models, newborn screening for X-linked Adrenoleukodystrophy and consensus recommendations for Tyrosinemia Type 1. The Consumer Alliance group included a strategy session on navigating challenges with insurance coverage for medical foods, and discussions about public private partnerships and
consumer advocacy updates from each state. Plans are currently underway to hold the next annual meeting from July 13-15 at the Renaissance Hotel in Asheville, North Carolina.

The **Longitudinal Pediatric Data Resource (LPDR)**, is a data collection tool developed by the National Coordinating Center (NCC), in partnership with the Newborn Screening Translational Research Network (NBSTRN) to support longitudinal data collection.

SERC was awarded a grant to participate in Phase I of this collaboration, to demonstrate the feasibility of answering 14 public health questions to understand public health long-term follow-up by entering data into and utilizing the LPDR. Currently, three states in the SERC region have met with individuals from NewSteps, APHL, and ACMG to discuss how to expand their current short-term follow-up (STFU) database to include data elements from the LPDR to capture the public health data. This will potentially be a national model.

The **SERC Genetics Needs Assessment** was adapted from the Heartland Collaborative's needs assessment survey. This survey was approved through the Emory IRB and was administered via SurveyMonkey. The SERC Genetics Needs assessment targets state public health officials with the hope of impacting the funding and resources given to both short and long term follow up clinics for NBS and other genetic conditions. The data for the regional needs assessment was shared at the 2016 annual SERC/SERGG meeting. In addition to the regional needs assessment, an assessment of telegenetics sites in the region was conducted. Currently, there are 5 states that use telegenetics with at least one new clinic being added in each state over the next 3 years.

The **SERC Consumer Alliance** has been a leader in nationwide advocacy efforts for recognizing medical foods as the primary nutritional treatment for inherited metabolic disorders. In the past year, the Consumer Alliance in collaboration with the National PKU Alliance, American Academy of Pediatrics, and North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition, have been active in the passing of medical foods legislation on the national level for military families. The National Defense Authorization Act for Fiscal Year 2017, Sec. 714, reads: “Provides Tricare program coverage for medically necessary food and vitamins for digestive disorders and inherited metabolic disorders.” The Medical Nutrition Equity Act is additionally moving its way through the legal system, having just gained bi-partisan sponsorship. This Act proposes to mandate the coverage of medically necessary foods through Medicare, Medicaid, and CHIP and has ongoing support and involvement from the Southeast Region. In Georgia, the newborn screening fee was raised by $13 in order to contribute funds towards the Medical Nutrition Therapy for Prevention Program at the Emory University Department of Human Genetics, as well as expansion of the newborn screening panel. Additionally, Senator Johnny Isakson, R-GA, declared December 3rd National PKU Awareness Day. As a group, the Consumer Alliance has published three newsletters in the past fiscal year, with information on events, advocacy and legislation, conferences, camps, and state updates. The group meets on a monthly basis via conference call with approximately 10 members present/call. Currently, the SERC Consumer Alliance has representation from the following states: Alabama – 3; Florida – 4; Georgia – 5; Louisiana – 3; Mississippi – 1; North Carolina – 3; South Carolina – 1; Tennessee – 1. Alabama and Louisiana’s state PKU organizations are currently working to obtain 501(c)3 status.
How much did you do?

Major Region 4 Midwest projects in year 5.

**Sickle Cell Trait Follow-up Initiatives.** Region 4 Midwest continues to be engaged with the Sickle Cell Disease community, especially related to the Hemoglobinopathies (HGB) work group’s efforts around Sickle Cell Trait follow-up and education. Region 4 Midwest supported representatives to present at the Sickle Cell Disease Association of America (SCDAA) and the Sickle Cell Disease Association of Illinois (SCDAI) annual conferences. The HGB work group is preparing a manuscript of their sickle cell trait follow-up recommendations for publication and is currently working towards designing a quality improvement project to work towards full implementation of some of the recommendations.

**Long Term Follow Up.** The Region 4 LTFU Workgroup has developed short, condition-specific question lists for states to use in implementing and/or enhancing their long-term follow-up programs. Conditions include Cystic Fibrosis, Congenital Hypothyroidism, and Sickle Cell Disease. The group has also developed an online toolkit of resources for the coverage of medical foods and formulas for both state programs and patients. **Needs Assessment Activities.** Region 4 Midwest staff culminated several needs assessment activities this project year to understand the state of genetics care and who and where the most underserved patients are in the region. Methods included mapping projects, implementation of the Genetics System Assessment tool, and a provider survey of telegenetics activities. Major findings are highlighted in the section below.

**Care Coordination: Empowering Families Training.** Continued to partner with HRSA-funded projects, family advocacy groups, and the MI Department of Health and Human Services to provide trainings, manage and analyze data, and support new facilitators for the one day training that introduces concepts around Medical Home, Transition, Insurance, Partnering with Providers, Organization, Peer Support, and Self Care for caregivers of children with genetic conditions. In Year 5, Region 4 Midwest trained 84 caregivers in seven trainings in collaboration with the MI Department of Health and Human Services, MI Family 2 Family, and the Pacer Center in MN. Over 400 caregivers have been trained to date.

**Journey through Diagnosis Guide.** Completed and distributed the *Journey through Diagnosis* Guide. The purpose of this guide is to provide information and support to families whose child has been identified with a genetic condition. The guide was conceptualized, written, and edited by members of Region 4’s Family Forum. It was presented at the 2016 Regional Meeting where there were requests among stakeholders for 1,415 printed copies. An additional 438 electronic copies have been downloaded from the Region 4 Midwest’s website to date.

**Regional Meeting.** Hosted the Annual Regional Meeting on October 17&18, 2016 in Lansing, MI for 62 representatives from all seven Midwest states. Stakeholders included genetic service providers, families, and public health professionals. On day one, presentations and discussions around emerging priority topics (Telegenetics, Genetics Education, Quality Improvement and Healthcare Access, and Health Equity) were led by regional experts and national partners. On day two, Region 4 Midwest staff and stakeholders participated in facilitated strategic planning sessions. Conversations resulted in input from diverse stakeholders regarding issues relevant to HRSA including, but not limited to, how Region 4 might improve access to genetic services through expansion of telemedicine, health equity and quality improvement practices.

How well did you do it?

**Highlights of successful implementation of several of Region 4 Midwest’s projects.**

**Needs Assessment Findings.** This year saw several needs assessment efforts come to fruition. Dr. Chou delivered and presented on results of the GSA. Overall, Region 4 Midwest states scored highly on the high-impact elements of the assessment, indicating strong foundations for the genetics care system. The group facilitation and GSA results both indicated potential
disconnects between state services provided and consumer knowledge of and access to those services. Results from the MI NBS mapping project demonstrated three main findings: 1) different conditions have specific population distributions; 2) many metabolic patients live a great distance from specialty care (33%, 245 patients, live greater than 120 miles from the metabolic clinic in Detroit); 3) while there was no association between distance from care and active status for metabolic patients, these distances present an opportunity to incorporate telemedicine to reduce burden on these families. Finally, a survey of genetics care providers in the Midwest (n=32) showed only two (6%) reported they were currently using telemedicine to see genetics patients. Of the remaining 30 providers, 28 (93%) indicated they were interested in using telemedicine in the future.

**Dissemination of Findings.** Region 4 Midwest staff and partners have worked to disseminate findings through publications and presentations as the five year grant cycle comes to an end. The following three publications represent major project work in Congenital Hypothyroidism three-year follow-up, the Care Coordination: Empowering Families training evaluation, and the Sickle Cell Trait short term follow-up recommendations.


**Regional Meeting and Collaboration.** For this final year of the project, Regional Meeting was focused on transition, both celebrating successes and having conversations about future directions for the collaborative. The formal meeting evaluation and informal feedback showed that the majority of stakeholders were positive about a new emphasis on health equity and addressing underserved patients within the region.

- *I appreciated the strong focus on health equity during this meeting, a positive change from previous meetings.* –Public Health Non-genetics Professional
- *This was an excellent meeting, the best I have attended so far. The presentations were great, and the meeting provided an excellent structure for the exchange of ideas, concerns, challenges, and opportunities. The dialogue around many of the talks was excellent.* –Newborn Screening Professional

**Was anyone better off?**

*Highlights of positive impact from several of Region 4 Midwest’s projects.*

**Sickle Cell Trait Follow-up Initiatives.** Region 4 partner, the Minnesota Department of Health, was nominated for the Governor’s 2016 Better Government Award for the work they did to provide sickle cell trait short-term follow-up in their newborn screening program. This work was a culmination of the efforts and partnerships of the Region 4 Hemoglobinopathies work group’s sickle cell trait follow-up initiatives. The Newborn Screening Program at the Minnesota Department of Health reached out to the most at-risk communities to find out their preferred methods of SCT notification. Changes to the program improved notification to families from about 38% to 96%.

**NBS Laboratory Forum.** Following the voluntary recall by Hologic of their molecular test for Cystic Fibrosis on newborn dried blood spot specimens, the Region 4 Midwest was able to facilitate technical assistance and back up laboratories for testing through the Newborn Screening Laboratory Forum. Laboratory staff from Midwest states used Region 4’s Forum to come together and troubleshoot changes in CF testing and prepare for back-up testing. As a result babies born in states using the recalled Hologic kit were able to continue to be screened.

**Family Forum Videos and Journey through Diagnosis Guide.** Over 1,800 guides have been distributed to families to date. Region 4 states Minnesota and Michigan plan to provide the guidebooks to newly diagnosed families in their states. The guide was conceived of, created, and reviewed by Region 4 families, who were proud to produce a high-quality product whose supportive advice will ripple through the Midwest as it is broadly disseminated. *I was very happy with how well our guidebook was so well received. And that family is definitely recognized as a valued & valuable members of the team.*—Family Forum Member
Mentorship Project

How much did you do?
The goals of the mentoring project were to develop advocacy skills in youth who have a genetic/chromosomal condition. Ultimately, we anticipate that the advocacy training would make a difference not only in the lives of the youth, but in their states, condition-specific advocacy organizations, and Heartland advocacy efforts.

A core activity of the project was to develop and implement a mentoring curriculum. The curriculum was developed in collaboration with the Midlands Mentoring Partnership, a member of the national MENTOR network. In September 24, 2016, the Heartland sponsored six volunteer mentors to attend a training on mentoring concepts and skills in Omaha, Nebraska. Prior to this training, five young people living in the Heartland Region were identified as interested participants in the Mentoring Project as mentees. Nearly all of the participating youth and trained mentors attended the Annual Conference in Little Rock in October 2016. The mentoring activities were initiated at the Heartland conference. Following the conference, the process of matching mentors to youth was initiated. Mentoring was initiated in January 2017. A mentoring agreement was signed and goals for each youth is in the process of being established.

Related to the mentoring project, the Heartland Collaborative also sponsors the travel expenses for youth to attend the Face Forward Conference. The conference focus is to provide young adults ages 16-24 who have been diagnosed with PKU, metabolic, mitochondrial or other genetic conditions the opportunity to attend a four-day camp program to develop leadership, advocacy, and other skills to achieve a positive, healthy, and fulfilling lifestyle. Two of these Face Forward Heartland participants currently are in the Mentoring Project.

How well did you do it?
The mentoring training was received positively by the six participants. They indicated they gained knowledge and felt the training prepared them with the skills they needed to be a mentor. Overall, they reported feeling motivated to begin the mentoring process with their assigned youth. To facilitate the first meeting between mentors and mentees, Heartland provided a networking lunch prior to the start of the 12th annual conference in Little Rock, AR. This facilitated the first stage of the matching process as mentors and mentees had a chance to become acquainted with one another and exchange common interests.

Although initially a challenge, the project leaders were able to recruit strong youth advocates that have all consistently participated in mentoring activities. Recruitment initially began through contact with advocacy agencies (e.g., Family Voices). It was found that a multi-faceted recruitment process, which included word of mouth, was an effective approach. The initial training and mentor sessions received
positive feedback from both the youth and the mentors. There was evidence of strong commitment from both groups.

Was anyone better off?
The two youth who participated in the Face Forward Conference found it to be a positive and life changing experience. They were energized and focused on moving forward with their lives as a person with health conditions. The mentoring process is just beginning, so there are only preliminary findings. Mentors report that relationships are being developed and Smart Goals were established that would help to frame the mentoring content.

Individualized Health Plan

What did we do?
Individual Healthcare Plans (IHP) are written plans used in the school setting to communicate students’ health conditions and their care/accommodation needs. They are written for a variety of health conditions and make it easier for school teams to determine appropriate healthcare interventions. The Heartland Genetic Services Collaborative conducted a focus group in which many parents, school nurses, healthcare professionals, school administrators and advocates recognized that a consistent, standardized practice was not in place for healthcare plan development or implementation. This prompted Heartland to team with the University of South Dakota (USD) Center for Disabilities to design a project to improve the IHP process.

A team from the USD Center for Disabilities created a standard procedure and materials to serve as a “toolkit” of best practices for developing and utilizing IHPs. This team then trained ten participants, six school nurses, two program nurses, one nurse supervisor and one state Department of Education representative on effective methods for creating and implementing IHPs in order to better meet students’ individual needs. The participants in the training received hands-on practice using the “toolkit” in order to effectively use this model for schoolwide implementation. All training participants also took part in ongoing follow-along support throughout the school year.

How well did you do it?
Participants were very positive regarding the training and follow-up that was provided. They strongly felt that this was a need and the training provided them with the skills to improve the processes in their communities.

Was anyone better off?
All ten participants reported an increase in knowledge and understanding following the training, even those who rated themselves as having expert and advanced level knowledge prior to the training. Throughout the school year, those ten participants have trained nearly 400 additional school nurses in the IHP process. The participants have also helped develop over 4,800 Individual Healthcare Plans since the initial training. Following the training, the number of IHPs connected to IEPs or 504 plans have more than doubled in the schools served by one of the participants. Trained nurses have also noted that parents and school staff have demonstrated greater participation in the development of plans as well as increased satisfaction in the IHP process.

Based on the success of the first several months of implementation, the Center for Disabilities has received a request for an additional training opportunity for school nurse representatives in one state in the Heartland region in April 2017.
Who We Serve

With over one million square miles extending from Canada to Mexico, the Mountain States Region constitutes a land area of almost one-third of the United States and a population of approximately 50 million people. The Mountain States Region is richly diverse with large concentrations of Hispanic and Latino populations, Native Americans, and other culturally and linguistically distinct population groups. The region is also geographically diverse, with a high percentage of rural and frontier counties. In four states, a language other than English is spoken at home in one quarter to one third of all households. Each of the eight Mountain States is among the top twenty fastest growing states in the US. More than 700,000 births occur annually within the Mountain States, with an estimated 21,000 infants born with a birth defect each year.

Collaboration, Information Sharing, and Quality Services with Innovation

The mission of the MSGRC is to collaborate to ensure that individuals with heritable disorders and their families have access to quality care and appropriate genetics expertise and information in the context of a medical home. The three core values driving MSGRC priorities are regional collaboration with involvement from diverse populations, information sharing to improve quality of care, and innovation in identifying and managing treatable disorders and strengthening capacity of state public health departments. **Collaboration** is essential to deliver genetics services to unique populations, **Information sharing** is critical to strengthening the systems of care in which newborn screening and genetics services are accessible, family-centered, continuous, comprehensive, coordinated, compassionate, culturally competent, and maintained throughout the life course. **Innovation** is essential for strengthening newborn screening capacity in state public health departments, and for providing genetic treatment services for a diverse and growing population.

How much did we do?

Since June 1, 2016, **MSGRC hosted or sponsored 48 collaborative sessions using multiple venues** (in; person, webinar, conference calls). These collaborative sessions facilitated information sharing and project planning among our members (clinicians, public health professionals, and consumers).

Using **web and social media**, MSGRC reinforced and broadened the impact of the meetings, webinars, reports and conferencing. The MSGRC website and Facebook page published highlights, shared resources, encouraged collaboration, and directed consumers, genetics providers, other specialists, primary care providers, other healthcare providers, social service providers, and public health professionals to relevant genetics resources. The MSGRC website recorded 3,696 unique website visitors access a large variety of available information. The number of “likes” on the MSGRC Facebook page also continued to grow, increasing from 417 to 466.

**MSGRC conducted and participated in regional and national projects** related to inborn errors of metabolism, long-term follow-up of newborn screening, quality measures for long term follow-up of newborn screening, health care access and finance of genetic services, Hispanic access to genetic services, use of parent partner navigators in pediatric care for families of children with heritable conditions, telegenetics case study project and provider recruitment, genetics services mapping, and genetic services needs assessment. The MSGRC needs assessment, started in Year 4, was finalized and presented to stakeholders during a November 2016 meeting in Denver, Colorado.

MSGRC expanded its previous genetics services mapping work to look specifically at pediatric genetic service providers in each state and identify their capacity to serve both urban and rural patients. To date, twelve clinics have completed the survey to help MSGRC identify the number of practicing genetics professionals, the number and location of outreach clinics, and the use of telemedicine to serve patients. In addition, MSGRC is collecting data on the geographic residence of patients served by these clinics to map clinic catchment areas and identify areas of the region not currently served by an existing clinic. Completed
maps describing these findings will be available before the end of Year 5.

**How well did you do it?**

**MSGRC successfully focused attention, expanded knowledge, and made recommendations aimed at improving access and reducing barriers to quality genetics services.** The Workgroups and MSGRC leadership: 1) Increased *diverse consumer involvement* in MSGRC workgroups, and regional and national committee work; 2) Hosted a *regional meeting* to assess regional needs and develop plans for a regional genetics network; 3) Conducted a *case study* of a successful telegenetics program to identify elements and resources needed for developing telegenetics (accepted as an oral presentation at the ACMG Annual meeting); 4) Built on the previous Genetic Services *Mapping Project* to capture additional information on outreach sites, telegenetics, and patient demographics; and 5) Enhanced the *networking and information-sharing* among consumers through the MSGRC website and Facebook page.

**MSGRC strengthened partnerships with stakeholders, including governmental and community-based organizations and other HRSA MCHB-funded programs.** Members of the MSGRC team have national leadership roles on key committees. Dr. Janet Thomas, MSGRC Associate Program Director, chairs a NCC Healthcare Access and Finance Workgroup Small Group. Dr. Hassell, MSGRC Program Director, co-chairs the NBSTRN Clinical Integration Group, of which Dr. Thomas is also a member. Dr. Hassel is also on the Steering and NBS Definitions Committees for Newborn Screening Technical assistance and Evaluation Program (NewSTEPS). Dr. Celia Kaye, MSGRC Consultant serves on the NCC’s ACT sheet workgroup. Drs. Hassell, Thomas, and Kaye serve on the ACHDNC Follow-Up and Treatment workgroup, and Joyce Hooker serves on the ACHDNC Education workgroup. Dr. Thomas and Marilyn Brown, MSGRC Program Coordinator, also serve on the Hispanic Access Advisory Committee for the Heartland Genetics Services Collaborative. Through strong leadership and partnership, MSGRC has leveraged resources, tested innovative models for improving services delivery, and supported national medical home priorities.

**Was anyone better off?**

MSGRC has a strong history of engagement and collaboration with genetics professionals, consumer advocates, public health leaders, and other HRSA funded programs. As part of our activities in Years 4 and 5, MSGRC has attracted many new integral members, expanded our connections to new genetic centers, and increased the diversity of our consumer members. Members of our HIG and MC benefit from a forum in which they can discuss and learn from difficult cases and review and update care plans, leading to better patient care. Providers have also received training in telegenetics which has resulted in introduction of this model in at least one additional center in Colorado. In the 11/16 Strategic Planning Meeting, our key stakeholders and leaders met to plan expansion of MSGRC to better engage genetic centers in our region and identify underserved sites in which to target future activities.

MSGRC celebrated many successes in the last year. The *needs assessment*, which was initiated in Year 4, was completed and presented at the November meeting. The needs assessment helped guide MSGRC program priorities for Year 5 and in planning for future activities. *Parent Partners* has continued to expand to new sites in the last year, including American Indian communities and a military base. MSGRC’s evaluation of the project has shown positive outcomes, and was integral in the state funding of this project in both Montana and Wyoming. This project demonstrates MSGRC’s ability to expand mini; projects into sustainable programs. MSGRC also initiated the *Cook Children’s Hospital case study*, a project aiming to describe characteristics of a successful telegenetics model that may inform implementation in other regional centers. The expanded mapping project has allowed MSGRC to more closely identify what areas of the region are in highest need of expanded genetics resources and services. This information will allow us to continue to allocate our resources strategically, targeting our highest need populations. MSGRC is currently planning our April 2017 *Annual Meeting*, focused on collaboration and resource-sharing throughout the region. In this, MSGRC intends to celebrate the successes of our past five years and foster the sustainability of MSGRC achievements.
How much did you do?

**Major Projects**

- Affordable Care Act through the Life Course Webpages
- Annual Regional Stakeholder Partners Summit
- CPT1 Arctic Variant Community Project
- Educational and Training Webinars, Teleconferences
- Genetic Visit Outreach Outcomes
- Genetic Services Assessment by State Title V Leaders
- Newborn Screening Parent Fact Sheet Maintenance, Development and Distribution
- Telegenetics Education and Training Project
- Telegenetics and Genetic Services Outcomes
- Newborn Screening Survey
- Coordination of newborn screening activities
- Genetic information for Family to Family and Family Voices organizations
- Collaboration with LEND programs in the region

How well did you do it?

**Affordable Care Act through the Life Course Webpages** – webpages on WSGSC website devoted to a family-friendly explanation of what the ACA is, benefits, how to obtain information about ACA based upon life course model and developmental milestones.

**Annual Regional Stakeholder Partners Summit** – state public health genetics and newborn screening leaders, state family organization leaders, representatives from state specialty and primary care provider groups convene to both share information about best practices and to learn about current events and topics related to genetics services and newborn screening. Stakeholders consistently report satisfaction with and knowledge gained during this annual event.

**CPT1 Arctic Variant Community Project** - The project seeks to improve newborn detection, management, and long-term follow-up of children with carnitine palmitoyl transferase deficiency, type 1 (CPT-1A) Arctic variant.

**Educational and Training Webinars, Teleconferences** – numerous webinars and teleconferences devoted to information sharing and educational training sessions related to telegenetics practice, newborn screening current topics and outcomes of genetic services.

**Genetic Visit Outreach Outcomes** – assuring access to genetic specialty services via outreach clinics in Alaska and Hawaii, documenting outcomes of visits as perceived by both patients and genetic counselors.

**Genetic Services Assessment by State Title V Leaders** – teleconference focused conversations with state Title V leaders seeking their perceptions of current status of genetic services using Genetic Systems Assessment survey in conjunction with other regional collaboratives.

**Newborn Screening Parent Fact Sheet Maintenance, Development and Distribution** – ongoing expert updating and national distribution of fact sheets related to information about conditions detected via newborn screening.

**Telegenetics Education and Training Project** – inter-regional project to develop and offer training about telegenetics to increase understanding of and uptake of use of telemedicine among genetic specialists.

**Telegenetics and Genetic Services Outcomes** – completed and shared framework for measuring genetic services with all RCs, working on process for expert development of health outcomes.

**Newborn Screening Survey** – assessing parent opinion on preferences for notification of risk related to age of symptom onset from positive newborn screen, findings available May 2017.

**Coordination of newborn screening activities** – the WSGSC coordinates activities of concern to the
newborn screening programs in our region. This includes: providing information about the ACHDNC to the states and providing information from the states back to the ACHDNC: linkages with the NBSTRN, NewSTEPs, and Baby’s First Test; and bringing together NBS stakeholders for discussions when common issues arise (e.g. implementing X-ALD screening).

**Genetic information for Family to Family Health Information Centers and Family Voices organizations** – the WSGSC staff provide information about genetics and newborn screening issues when requested by the F2F HICs and Family Voices organizations. These include referral information, articles for their websites or newsletters, and parent support organization information.

**Collaboration with LEND programs in the region** – information from the WSGSC is shared with the LEND leaders in our region to be used in their training activities.

**Was anyone better off?**

**Affordable Care Act Family Survey** – families throughout the Western region have the opportunity to engage in sharing their needs and current status related to access to and quality of genetic services, findings from the needs assessment will guide improved policy and program development.

**Affordable Care Act through the Life Course Webpages** – annually, hundreds of page views of the ACA pages in the Western states and throughout the nation reflect the interest in users learning about the impact and specifics of the ACA. Anecdotally we have heard from many providers and family advocates who use and share the website with their patients or clients.

**Annual Regional Stakeholder Partners Summit** – key to a regional approach to improving access to genetics services, the annual regional summit provides the only opportunity available for all genetics/NBS stakeholders in the region to come together to learn about best practices, provide input to national leaders and to plan for regional genetics/NBS needs.

**CPT1 Arctic Variant Community Project** – a unique opportunity for families in the region living with CPT1 Arctic variant to understand their risks as well as treatment and management of this regional genetic condition. Public health geneticists and researchers may use this project as a model for improving understanding and treatment for rare genetic disorders.

**Educational and Training Webinars, Teleconferences** – hundreds of genetics/NBS stakeholder, including consumers/families have received education and information about genetics they would not otherwise have had access to.

**Genetic Visit Outreach Outcomes** - hundreds of families in Alaska and Hawaii have received genetic services for diagnosis, ongoing treatment and management they would not otherwise have had access to.

**Genetic Services Assessment by State Title V Leaders** – regional families will benefit when programs are planned based upon knowledge shared by Title V leaders who know what is happening in their states regarding access to genetic specialty care within or outside of medical homes.

**Newborn Screening Parent Fact Sheet Maintenance, Development and Distribution** – hundreds of families and providers have access to accurate, easily understood information about newborn screening conditions.

**Telegenetics Education and Training Project** – a foundation for increasing access to genetic services is being built by training new providers to incorporate telemedicine as part of routine practice. This effort has been expanded to other regions who are now offered this training.

**Telegenetics and Genetic Services Outcomes** – regional and national dissemination of current practices related to measuring outcomes, documented framework of approach to measuring outcomes will ultimately prove the worth of genetic services for families and improve reimbursement.

**Newborn Screening Survey** – findings from this survey will be used by regional and possibly national NBS policy makers to develop appropriate policy for when to notify parents of risks related to positive newborn screen for later onset conditions.

**Coordination of newborn screening activities** – the state NBS programs benefit by being able to jointly discuss issues and participate in national activities. This leads to all babies in each of the states benefiting from better NBS activities.

**Genetic information for Family to Family Health Information Centers and Family Voices organizations** – the F2F HICs and Family Voices organizations and the families they serve benefit by having up-to-date and accurate genetic and newborn screening information.

**Collaboration with LEND programs in the region** – the LEND trainees benefit by having genetics and newborn screening information as part of their training.
# New England Genetics Collaborative Regional Activities

## Intra-Regional

<table>
<thead>
<tr>
<th>Medical Home</th>
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<tbody>
<tr>
<td><strong>Title:</strong> Medical Home Webinar Series</td>
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<tr>
<td><strong>Description:</strong> Continued web-based educational supports to improve integration of genetics and primary care medicine.</td>
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<tr>
<td><strong>Who with:</strong> Members of the NEGC Medical Home Workgroup, including representatives from Maine, New Hampshire, and Vermont Pediatric Improvement Partnerships.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> Developing a new webinar focused on building collaboration between Family outreach groups and pediatric providers. The new webinar will be hosted 3/30/17. More details on the webinar series (including recordings of past events) are accessible here: <a href="http://www.negenetics.org/work-groups/medical/products_pubs_mh">http://www.negenetics.org/work-groups/medical/products_pubs_mh</a>.</td>
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## NBS Capacity Building

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<th>NBS Capacity Building</th>
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<tr>
<td><strong>Title:</strong> CCHD Screening and Related Resources</td>
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<tr>
<td><strong>Description:</strong> Hosted a range of information for families impacted by CCHD following a successful screening project across 8 birthing centers in Maine, New Hampshire, Vermont, Rhode Island and Connecticut</td>
</tr>
<tr>
<td><strong>Who With:</strong> UNH, Institute on Disability, Project Partners</td>
</tr>
<tr>
<td><strong>Accomplishment:</strong> The NEGC continues to support an electronic resource for families on psychosocial supports (live in May 2016 <a href="http://www.necongenitalheartresources.org">www.necongenitalheartresources.org</a>). During the current year, 81 users have accessed the resource page.</td>
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## Collaboration

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<th>Collaboration</th>
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<tr>
<td><strong>Title:</strong> Quality Improvement Workgroup</td>
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<tr>
<td><strong>Description:</strong> The QI workgroup seeks to engage all centers in the continuous quality improvement of metabolic center care of patients with inborn errors of metabolism. One of the primary mechanisms supporting this initiative is a shared registry for individuals with</td>
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<tr>
<td><strong>Title:</strong> GEMSS Resource Website for Schools, Families, and their Children</td>
</tr>
<tr>
<td><strong>Description:</strong> Provides users with a broad range of condition specific information on how to best support children with a genetic condition in a school setting.</td>
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DD/ID, PKU, or MCAD. By entering information on the health care needs and services of participating patients and then sharing summary information in a group setting, the QI workgroup is collectively able to come to a more informed understanding of practice patterns across the region and identify potential areas for improvement.

**Who with:** Specialists, primary care providers, family advocates from the New England region.

**Accomplishment:** The quality improvement learning work groups have recruited participation by 5 clinic sites for addressing developmental delay and intellectual disability and 10 clinic sites for PKU or MCAD. Through November, 2016, 2,116 individuals with DD/ID have been entered into the registry.

| Title: Annual Collaboration with the New England Regional Genetics Group |
| Description: Co-sponsorship on telehealth session at NERGG’s annual meeting |
| **Who with:** Participants of the NERGG Annual Meeting |
| **Accomplishment:** Presentation by Jill Rinehart, MD, & Marinell Newton, MSW, “Medical Home in New England: Achieving a Shared Plan of Care for Effective Care Coordination”, at New England Regional Genetics Group, Inc. ~90 in attendance. Co-sponsored session with NEGC. |

| Title: Integration of genetics resources into regional LEND programs |
| Description: This activity seeks to integrate a range of critical information from the genetics community into the training curricula of New England LEND programs. |
| **Who with:** LEND programs representing Maine, Vermont, and New Hampshire |
| **Accomplishment:** The NEGC LEND group meets twice yearly and members have agreed to an overall educational objective for LEND training programs. Dr. Moeschler has participated on the AUCD National Evaluation Brief 38|Page 38|Page |
Genetics workgroup in planning for the Fall 2016 LEND Directors meeting and will participate in a session on implementing appropriate genetics education in LEND programs. The meetings have led to a shared understanding that many LEND programs have genetics education incorporated and that those programs have a “local approach” to genetics education. The draft core curriculum is completed and will be circulated to LEND directors for comments Spring 2017. The AUCD website has a specific page for genetics education that includes the genetics education modules for programs to share and to contribute to.

### Affordable Care Act

| **Title:** Health Care Access and Financing - Insurance Appeal Resources for Families |
| **Description:** The Health Care Access and Financing Group of the NEGC continued its efforts to support families and individuals living with a genetic condition. The focus of this year’s efforts was on documenting recommendations and best practices around filing for successful appeals on insurance claims. The results of this effort are expected to be publicly released by the time of the NEGC Annual Meeting on April 6-7, 2017. |
| **Who with:** Families, Advocacy Organizations, NEGC Staff |
| **Accomplishment:** Initial findings were shared with members of the HAF workgroup, a draft resource sheet has been created along with a useful infographic, and dissemination strategies identified. |

| **Title:** NCC Health Care, Access, and Financing Group |
| **Description:** Ongoing collaboration with members of the NCC to review ongoing policy developments and field research. |
| **Who with:** Regional Collaboratives, NCC, HRSA |
| **Accomplishments:** As a result of our initial efforts and follow up collaborations, other regions have implemented their own survey efforts documenting family experiences with health care insurance and service access and a new journal publication was created which informs the knowledge base concerning the needs of families and individuals living with a genetic condition. |

### Transition

| **Title:** Face Forward 2016 Summer Camperence for Youth |
| **Description:** Four-day, 3-night retreat experience for youth with genetic conditions, designed to explore transition issues, including “difficult conversations; sponsored by Next Step, Inc. |
| **Who with:** youth and young adults ages 16-24 with metabolic conditions, Neurofibromatosis, type 1, mitochondrial disease or other genetic conditions. |
**Accomplishment:** Twenty five youth and young adults from across the country gained invaluable experiences propelling them toward self-advocacy.

<table>
<thead>
<tr>
<th>Title</th>
<th>Description</th>
<th>Who with</th>
<th>Accomplishments</th>
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<tbody>
<tr>
<td>Health Surveys of Adults Living with Genetic Conditions</td>
<td>Continuing to collect adult health survey data to document potential disparities in health and well-being among those with and without targeted conditions.</td>
<td>Members of the NEGC and NCC transition workgroups.</td>
<td>To date, survey data has been collected from 93 condition oriented and 52 control adults. Early indications of increased onset of older adult health challenges among the young adult population living with genetic conditions.</td>
</tr>
<tr>
<td>Homocystinuria/Metabolic Support Group</td>
<td>Dr. Waisbren provided support to a young man in his efforts to coordinate a support group for individuals living with genetic conditions.</td>
<td>Individuals living with homocystinuria and metabolic conditions.</td>
<td>4 individuals participated in the support group (met 1x).</td>
</tr>
<tr>
<td>Implementation of Transition Policy</td>
<td>Dr. Waisbren worked with a range of partners to develop a transition policy in 2015/2016 based on best practices to help ensure appropriate levels of care for individuals living with genetic conditions.</td>
<td>Children's Hospital Boston, Area Metabolic Clinics</td>
<td>Children's Hospital of Boston and a clinic in Vermont are implementing the policy developed by Dr. Waisbren. The policy is available for review/adoption by other clinics at <a href="http://newenglandconsortium.org/for-professionals/transition-to-adult-care/transition-to-adult-healthcare-clinicians-policy/">http://newenglandconsortium.org/for-professionals/transition-to-adult-care/transition-to-adult-healthcare-clinicians-policy/</a>. During</td>
</tr>
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the current project year, the clinician policy page has had 43 page views and the family page 16 views.

| Title: Long Distance Metabolic Education Support |
| Description: There are not enough clinicians to manage all patients diagnosed with inborn errors, whether diagnosed clinically or by newborn screening. The problem is complicated by the lack of “metabolic competence” of subspecialists who are most likely to see these patients in their clinical practice. This situation directly impacts the diagnosis and management of these patients in New England, and beyond. To address this area, the NEGC is actively supporting the launch of the Genetic Metabolic Center for Education (GMCE) |
| Who with: Genetic Metabolic Center for Education |
| Accomplishments: GMCE partners have continued to provide critical supports in the New England area for a range of conditions (including Nonketotic hyperglycinemia, Glut-1 defect, neonatal cholestasis, and neuronal ceroid lipofuscinosis) across 3 participating clinic sites in 2017. To date, GMCE has provided 97 consults in the New England area, with 63 consults focused on management, 18 on diagnosis, 13 on diagnosis & management, and 3 on counseling. |
## New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) Regional Activities

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<thead>
<tr>
<th>Intra-Regional</th>
<th>Inter-Regional</th>
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<tbody>
<tr>
<td><strong>Medical Home</strong></td>
<td><strong>Title:</strong> NCC Care Coordination Workgroup</td>
</tr>
<tr>
<td><strong>Description:</strong> NYMAC team members continue to participate in the NCC Care Coordination Workgroup.</td>
<td><strong>Who with:</strong> Parents/advocates and representatives from different regions and national partners.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> NYMAC and other regions continue to gather to share and discuss resources, challenges, and successes related to care coordination.</td>
<td><strong>Accomplishments:</strong></td>
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analytic, analytic, and post-analytic timeliness, families affected by NBS, and NBS recommendations.

<table>
<thead>
<tr>
<th>Title: NBS video</th>
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<tr>
<td><strong>Description:</strong> The NBS Specimen Collection and Timeliness Video describes the importance of NBS and the collection of NBS blood spots, including timing and timeliness, procedures, differences between invalid and valid tests, and transportation of tests.</td>
</tr>
<tr>
<td><strong>Who With:</strong> NBS Program Staff and the NBS Work Group.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> The video was piloted and distributed to hospitals. A “test your knowledge” quiz was offered to hospital staff. Differences within individuals on pre-test and post-tests indicated that viewers’ knowledge of NBS timeliness increased significantly. The average pre-test score across all participants (N=72) was 90%; the average post-test score was 94.6%. Overall scores on assessments for those 50 participants that took both the pre-test and post-test increased by 6.7% on average, a significant increase (t value = 5.04, p&lt;0.001).</td>
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<table>
<thead>
<tr>
<th>Title: Newborn Screening Hospital Survey</th>
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<tr>
<td><strong>Description:</strong> As part of the NBS timeliness project, we distributed a survey to hospitals in the region to gather information about current NBS practices. Information gathered included the staff responsible for collecting and managing blood spots, packaging and sending of specimens, and barriers to the hospital sending blood spots within 24 hours of collection. In the next phase of the survey, hospitals will be surveyed regarding their electronic capabilities including their readiness to adopt health information technology strategies.</td>
</tr>
<tr>
<td><strong>Who with:</strong> Hospitals located in the NYMAC region.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> One hundred and forty-nine hospitals have completed the survey from five states; approximately 50% of the hospitals in the region. The survey responses are being prepared for analysis.</td>
</tr>
</tbody>
</table>
| **Title:** NYMAC Regional Genetics Education Network  
**Description:** The RGEN is an organization of volunteer patients, advocates, providers, genetic counselors willing to provide in-person or webinar-based education on genetics and newborn screening.  
**Who with:** Patients, providers, public health and genetic counselors in the NYMAC region.  
**Accomplishments:** There are currently 57 individuals registered for the RGEN who are available to speak about genetics and NBS topics. Three educational events have occurred. |
|---|
| **Title:** NCAHM collaboration & Hearing Loss survey  
**Description:** A survey of pediatricians was developed, distributed and analyzed. The survey featured questions surrounding knowledge of the EHDI 1-3-6 guidelines, family referrals for hearing loss, and confidence speaking with families about genetic causes of hearing loss among others.  
**Who with:** Title V, EHDI Programs, EI Programs, Gallaudet University, the ACMG HL Guidelines Committee, and the National Center for Hearing Assessment and Management (NCHAM)  
**Accomplishments:** Over 250 physicians completed the survey. Data were presented at the NYMAC Summit and EHDI meeting. The poster won best poster award in the EHDI workforce category at the EHDI 2017 conference. A manuscript is in preparation. |
| **Title:** Deafness and Genetics  
**Description:** A patient-targeted handout and a provider-targeted handout were created and distributed. The handout for providers detailed considerations for a genetics referral/consultation for deafness or hard of hearing children, such as when to refer, what is needed, and how to refer (including CPT codes for connexin testing). The patient handout provided resources to information about genetic causes of deafness and the importance of a genetics consultation.  
**Who with:** NCHAM, HRSA, HRSA Title V Region III, EHDI programs  
**Accomplishments:** DHH-genetics handouts were prepared and distributed by NYMAC. The dissemination plan for the providers’ handout included: an upload on the NYMAC website, distribution to stakeholders through emails, NYMAC news and Facebook, advertisement in pediatrics journals, distribution at the EHDI conference, publication in the EHDI AAP newsletter, and distribution through NCHAM and EHDI coordinators networks. The dissemination plan for the patients’ handout included dissemination through Facebook, and NCHAM and EHDI coordinators. |
Title: NYMAC Genetics/Genomics in Public Health  
**Description:** Genetic counseling students from the NYMAC region participated in the GPH fellowship. Three webinars took place prior to 2/28 with 106 attendees, focusing on access to care and care coordination, the role of genetic counselors in public health, and patient-centered communication, advocacy, and cultural humility. An evaluation of the fellows’ experience will be conducted at the end of the fellowship period. Presenters included: Dana Yarbrough, Brenda Figueroa, Beth Vogel, Amy Gaviglio, Jana Monaco, and Ediomi Utuk.  
**Who with:** Thirteen Students from genetic counseling programs and LEND programs in the NYMAC region participate in the GPH Fellowship.  
**Accomplishments:** Academic centers and trainees became more engaged in NYMAC activities. NYMAC resources were disseminated to fellows, and fellows participated in three webinars regarding care coordination, public health, and patient-centered communication, advocacy, and cultural humility. These webinars also became open to the public; however, discussion forums were only available for fellows.

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Title: Affordable Care Act  
**Title:** PKU Survey  
**Description:** In collaboration with genetic dietitians throughout the mid-Atlantic region, NYMAC developed a web-based survey for parents/caregivers of persons with PKU, and/or persons with PKU. The overarching theme of the survey was history of medical formula and low protein food usage, and the availability and challenges to accessing medical formula and low protein foods, particularly surrounding costs, insurance coverage, and sources of financial assistance.  
**Who with:** Individuals with PKU and their parents and/or caregivers. Dietitians in the region helped to develop the survey.  
**Accomplishments:** The results of the survey were analyzed and shared at the NYMAC Summit and with the Secretary’s Advisory

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Title: NCC Healthcare Access and Financing  
**Description:** NYMAC team members continue to participate in the NCC Healthcare Access and Financing Workgroup.  
**Who with:** Representatives from NYMAC and other regions.  
**Accomplishments:** NYMAC continues to contribute to the NCC Healthcare Access and Financing efforts including sharing information about telegenetics services and access to treatment for PKU gathered through NYMAC surveys.
Committee on Heritable Disorders in Newborns and Children. The results will be published, and results will be used to create advocacy materials and create resources to help individuals and families obtain coverage.

**Transition**

**Title:** Summit Session on Transition  
**Description:** At the 2016 Summit meeting in Baltimore, MD, a session was dedicated to transition. There were two learning objectives, 1: “Explain models of transition from pediatric to adult medical care for individuals with genetic disease using sickle cell disease as an example.” And 2. “Discuss the interplay between a primary care provider and a specialist in the transition care process.”  
**Who with:** Providers, patients/families, public health professionals, and trainees attended the session. The session was led by Miren Blackwood, Robert Ostrander, and Debra Cohen.  
**Accomplishments:** NYMAC resources regarding transition were distributed to attendees. Attendees and presenters were asked to consider regional strategies to improve transition for individuals with special healthcare needs.

**Telegenetics**

**Title:** Telegenetics Community of Practice (TCOP)  
**Description:** Established in 2016, the TCOP aims to bring together providers with experience or interest in telegenetics. An interactive online community offers TCOP members access to webinars, slides, and discussion forums among other resources. NYMAC collaborated with the telehealth resource centers to develop the webinars.  
**Who with:** Providers in the NYMAC region who are practicing, have practiced, or are interested in telegenetics may join the TCOP.  
**Accomplishments:** Over 100 providers in the region have joined the TCOP. The TCOP has developed, coordinated, and shared webinars focusing on 1) an introduction and summary of the telegenetics landscape survey findings, 2) conducting a needs assessment for

**Title:** NCC Telegenetics Workgroup  
**Description:** A genetic counselor and NYMAC team members continue to participate in the NCC Telegenetics Workgroup.  
**Who with:** Representatives from NYMAC and other regions and Telehealth Resource Centers.  
**Accomplishments:** NYMAC continues to contribute to the NCC Telegenetics Workgroup efforts including work on telegenetics outcomes. Alissa Bovee, NYMAC team member and genetic counselor, attended the in-person meeting.
telegenetics, 3) Care coordination by telegenetics, and 4) Business planning and financial models for telegenetics.

**Title:** Telegenetics Landscape Survey  
**Description:** In the fall of 2016, a survey was distributed to genetics providers (at the program level) who were planning to use, currently using, or previously using telegenetics to provide care to patients in the NYMAC region. The survey was divided into sections including a description of the telegenetics service, program staff, patients served, program funding, care coordination, and lessons learned.  
**Who with:** Genetics programs that are practicing, have practiced, or plan to practice clinical genetic care via telegenetics and reach patients in the NYMAC region  
**Accomplishments:** Fifty-four unique telegenetics programs completed the survey. Results have been presented at the 2016 NYMAC Summit, NCC Telegenetics Workgroup meeting, during at TCOP webinar, and will be presented at the MATRC pre-summit session on telegenetics. Results of the survey are currently being prepared for publication.

**Title:** Pre-Summit Telegenetics Meeting (prior to the Telehealth Research Symposium)  
**Description:** Members of NYMAC staff will convene with other professionals interested in telehealth at the 2017 Telehealth Research Symposium in Virginia. Sessions will focus on evidence-based research, technology, funding, evaluation, and future directions. A pre-summit meeting will focus on telegenetics.  
**Who with:** The Mid-Atlantic Telehealth Resource center is hosting the event, which is open to researchers, academics, and health care professionals. NYMAC recruited 6 genetics providers from the NYMAC region to serve as speakers and moderators at the meeting.  
**Accomplishments:** To date, 70 individuals have signed up to attend the telegenetics session. Funding was made available for 13 people to attend the conference: 7 speakers and 6 attendees. Members of the NYMAC team helped to develop, coordinate, and advertise the telegenetics session, and Alissa Bovee, NYMAC team member and genetic counselor, will be moderating the session. NYMAC has applied to offer Continuing Education Units for genetic counselors.

**Title:** Western States In-Person Telegenetics Training  
**Description:** We funded a genetic counselor to participate in the telegenetics training.  
**Who with:** A genetic counselor from NYMAC region and the Western States region. The training was led by Sylvia Mann and Michelle Takemoto.  
**Accomplishments:** The genetic counselor is now an active member of our Telegenetics Community of Practice.

**Other**

**Title:** RGEN Fragile X Presentation with Therapists
Description: Over 115 direct care providers have participated in educational programs related to Fragile X Syndrome. Two in-person presentations surrounding Fragile X Syndrome were given to therapists (the first directed towards speech therapists, the second directed towards occupational and physical therapists). These presentations discussed FXS at different times during the life course, the inheritance, diagnostic process, and genetic tests for FXS, and the parent perspective of having a child with FXS among other topics. One additional presentation is scheduled.

Who with: Speech, occupational, and physical therapists

Accomplishments: Between the two presentations, 44 participants completed feedback forms (21 ST, 18 OT, and 5 PT), with very positive feedback overall. For example, all participants somewhat or strongly agreed that the presentation helped them to understand the diagnostic process; 98% somewhat or strongly agreed that the presentation helped them to understand the benefits of early diagnosis and treatment, and 73% somewhat or strongly agreed that the information will change the way they practice.
## Southeast Regional NBS & Genetics Collaborative (SERC) Regional Activities

### Intra-Regional

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<th>NBS Capacity Building</th>
<th>Inter-Regional</th>
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| **Title:** Pompe disease presentations at the 2016 SERC SERGG Annual Meeting  
**Description:** The following topics on “Cross-Reactive Immunologic Material (CRIM) Positive Infantile Pompe Disease (IPD): Characterization of Immune Responses in Patient Treated with Enzyme Replacement Therapy (ERT)” and “Prophylactic Immune Modulation in Pompe Disease Using Low-Dose Methotrexate Induction: A Safe, Inexpensive, Widely Accessible, and Efficacious Strategy” were presented at the SERC annual meeting  
**Who with:** Regional genetics and newborn screening community  
**Accomplishments:** Shared knowledge of these topics to be added to the newborn screening panel. | **Title:** Web-based evidence and consensus based guidelines  
**Description:** evidence was gathered systematically from literature and consensus was obtained from experts by utilizing Delphi surveys and nominal group meetings with representation from all 7 regions  
**Who with:** Collaboration with GMDI members, parent organizations and AHRQ  
**Accomplishments:** Guidelines for MSUD and PKU have been released on the project’s web based portal. Propionic Acidemia guidelines are currently being reviewed and will be published in 2017. PKU guidelines were accepted and released through AHRQ. |

### Inter-Regional

**Title:** Connecting Genetic Counselor State Representatives  
**Description:** SERC is leveraging the SERC website infrastructure to strengthen genetic counselor networking across the Southeast region.  
**Who with:** Inter-regional Genetics Counselors  
**Accomplishments:** Facilitating connections and discussions about licensure and telemedicine efforts as well as any other topics of interest to the group, including regional needs for both patient and professional, and how to meet them.

### Collaboration

| **Title:** The SERC SERGG Annual Meeting 2016  
**Description:** To build on and enhance the work of SERC, particularly with the emphasis on core activities, annual meetings are held for face-to-face communication between the chairs and participants of the core activities as well as collaborative network | **Title:** Collaboration with Parent Organizations (including National Organization for Tyrosinemia Advocates –NOTA)  
**Description:** Using the NBS Connect registry infrastructure, SERC has been able to identify relationships with inter-regional groups. This is being translated to provide support and identify |
building with local, statewide and national partners. The 2016 annual meeting was held from July 14–16th, 2016 in Ponte Vedra Beach, Florida. 

**Who with:** In attendance were 122 healthcare professionals with an interest in genetics and public health. Twelve consumers also attended.  

**Accomplishments:** Thirty platform and twenty one poster presentations were made covering current issues in all areas of genetics: molecular genetics, cytogenetics, biochemical genetics and clinical genetics. Highlights of the meeting included newborn screening for GAMT deficiency, a live telemedicine demonstration, a report on results from the genetic needs assessment survey and another on the Registry and Surveillance System for Hemoglobinopathies (RuSH) project. Other topics covered were genomics and newborn screening, metabolomics, national nutrition management guidelines, regional collaborative models, newborn screening for X-linked Adrenoleukodystrophy and consensus recommendations for Tyrosinemia Type 1.  

**Affordable Care Act**

| Title: Exploration of medical foods and Affordable Care Act |
| Description: Strategies to include medical foods as a part of the Affordable Care Act |
| **Who with:** Catalyst Center |
| **Accomplishments:** A project has been identified with the Catalyst Center to update information related to medical foods legislation in each state. Our region participated in updating this information. |

**Long-Term Follow-Up**

| **Title:** The SERC Consumer Alliance advocacy efforts for medical foods |
| **Description:** The SERC Consumer Alliance has been active in the passing of medical foods legislation on the national level for military families. The Medical Nutrition Equity Act proposes to mandate the |

consumer needs for families affected by rare diseases, via a face-to-face meeting in Atlanta.

**Who with:** Inter-regional families and individuals with Tyrosinemia Type I SERC is hosting a meeting “Tyrosinemia Type I: Making Connections Count Meeting” in Atlanta, GA. **Accomplishments:** The purpose of this meeting is to connect families in the US together to understand the clinical needs for medical nutritional management. This meeting will give families and individuals with Tyrosinemia Type I the opportunity to interact with one another and health professionals in the field of inherited metabolic disorders (IMDs). Furthermore, they will have the opportunity to learn from one another.
coverage of medically necessary foods through Medicare, Medicaid, and CHIP and has ongoing support and involvement from the
Southeast Region. In Georgia, the newborn screening fee was raised by $13 in order to contribute funds towards the Medical Nutrition
Therapy for Prevention Program at the Emory University Department
of Human Genetics, as well as expansion of the newborn screening
panel. Additionally, Senator Johnny
Isakson, R-GA, declared December 3rd National PKU Awareness Day.
**Who with:** The SERC Consumer Alliance in collaboration with the
National PKU Alliance, American Academy of Pediatrics, and North
American Society for Pediatric Gastroenterology, Hepatology, and
Nutrition,

**Accomplishments:** As a group, the Consumer Alliance has published
three newsletters in the past fiscal year, with information on events,
advocacy and legislation, conferences, camps, and state updates.
The group meets on a monthly basis via conference call with
approximately 10 members present/call. Currently, the SERC
Consumer Alliance has representation from the following states:
Alabama – 3; Florida – 4; Georgia – 5; Louisiana – 3; Mississippi – 1;
North Carolina – 3; South Carolina – 1; Tennessee – 1. Alabama and
Louisiana’s state PKU organizations are currently working to obtain
501(c)3 status.

### Transition

**Title:** Transitioning Delphi Study (Key Elements for, and Indicators of,
a Successful Transition: Perspectives of Youth, Family and
Caregivers with Special Health Care Needs)

**Description:** The SERC Medical Home and Transitioning Work Group
in conjunction with the SERC Consumer Alliance has developed a
Delphi study on the healthcare transitioning process. The study has
been designed to achieve a consumer perspective on transitioning.

**Who with:** Ideal participants are patients and/or caregivers of
patients age 18 and older, residents of the United States, have a
diagnosed special healthcare need, and have completed the
healthcare transition from pediatric to adult care.  
**Accomplishments:** Several members of both the work group and Consumer Alliance have played instrumental roles in the recruiting process by involving their consumer networks. As of March 1, 2017, 20 participants have completed the verbal informed consent process. The short-term goal of this project is to have a sample size of 50 participants for the first Delphi survey, and conduct up to three rounds of the survey. The study team plans to publish and disseminate the survey results among healthcare professionals to guide the development of stronger, patient-centered transition programs for emerging adults with special health care needs.

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**Telegenetics**

**Title:** Annual telegenetics meeting  
**Description:** The meeting offered several presentations of diverse interests.  
**Who with:** Open to all attendees regardless of experience level.  
**Accomplishments:** Presentations ranged from nuts-and-bolts information on setting up a new telegenetics clinic to patient testimonials about using social media in healthcare, and a presentation on training clinicians and genetics counselors for telegenetics. There was a demonstration of the doxy.me software developed by a SERC member and is freely available for use on any basic digital media (e.g., laptop, smartphone, tablet).

**Title:** SERC Genetics Needs Assessment  
**Description:** The SERC Genetics Needs assessment targets state public health officials with the hope of impacting the funding and resources given to both short and long term follow up clinics for NBS and other genetic conditions.  
**Who with:** Adapted from the Heartland Collaborative's needs assessment survey.  
**Accomplishments:** This survey was approved through the Emory IRB and was administered via SurveyMonkey. The data for the regional needs assessment was shared at the 2016 annual SERC/SERGG meeting. In addition to the regional needs assessment, an assessment of telegenetics sites in the region was conducted. Currently, there are 5 states that use telegenetics with at least one new clinic being added in each state over the next 3 years.

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**Other**

**Title:** SERC lunch and learn series.  
**Description:** A lunch and learn event was organized in October 2016 to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students on the topic of “Medical Foods Insurance Coverage.”
**Who with:** A total of 24 individuals participated in this event in-person, while 10 individuals participated via webex throughout the region.

**Accomplishments:** The lunch and learn series resulted in a 9% increase in knowledge about “Medical Foods Insurance Coverage.” evidenced by results from the pre and post-test survey administered at this event.
### Region 4 Midwest Genetics Collaborative Regional Activities

#### Intra-Regional

**Title:** Care Coordination: Empowering Families Training  
**Description:** Full day training for parents of children with genetic conditions on care coordination and related medical home topics  
**Who with:** Region 4 Midwest staff and trained facilitators  
**Accomplishments:** The Care Coordination training continues to impact families across the country. Region 4 Midwest trained an additional 84 caregivers in seven trainings offered in collaboration with the MI Department of Health and Human Services, MI Family to Family, and the Pacer Center in MN.

#### Inter-Regional

**Title:** Care Coordination: Empowering Families Training - Spanish Translation  
**Description:** Inter-regional review and edit of Spanish translated curriculum  
**Who with:** Spanish speaking trainers in Heartland and Midwest  
**Accomplishments:** Training curriculum and participant materials reviewed for cultural competency and applicability for Spanish speaking population.

#### NBS Capacity Building

**Title:** Hemoglobinopathies Workgroup — Short –Term Follow-Up Recommendations for Sickle Cell Trait follow-Up in Newborn Screening Program Manuscript  
**Description:** Developed a manuscript to publish sickle cell trait short term follow-up recommendations developed by the group.  
**Who with:** Region 4 Hemoglobinopathy Workgroup  
**Accomplishments:** Draft manuscript outlining the follow-up recommendations and the process of development.

**Title:** Newborn Screening Laboratory Technical Assistance and Resource Sharing  
**Description:** Facilitated technical assistance and information sharing on topics of importance to regional newborn screening laboratories  
**Who with:** Region 4 NBS Laboratory Forum,  
**Accomplishments:** Facilitated technical assistance among the Region 4 states newborn screening laboratories regarding the recall of CF testing kits and implementation of new testing methods, Pompe and MPS1 screening implementation, using HL7 messaging for newborn screening results, and implementation NBS into state health information exchanges.

**Title:** Newborn Screening: What Prenatal Care Providers Need to Know Online Course  
**Description:** On-line course teaching ways to heighten expectant parents’ awareness of newborn screening  
**Who with:** Course participants from across the U.S.  
**Accomplishments:** Trained 50 prenatal care providers, child birth educators, and other health care professionals through online course available on Region 4’s website this reporting period.

**Title:** ACHDNC Participation  
**Description:** Regional representation and participation on the Secretary's Advisory Committee  
**Who with:** Sue Berry  
**Accomplishments:** Regional support for participation on ACHDNC workgroups and committees.
### Collaboration

**Title:** Diet for Life Metabolic Food and Formula Video Project  
**Description:** Produced a 4 minute video about the importance for individuals with metabolic conditions to stay on diet. In the video 3 patients talk about their experience managing their metabolic diet.  
**Who with:** Partnered with the Michigan Department of Health and Human Services and the Michigan Family to Family Health Information Center  
**Accomplishments:** Video housed on Region 4 Midwest YouTube page.

**Title:** NCC Workgroups  
**Description:** Support four Region 4 Midwest staff members to participate, support an additional four regional partners to participate in 8 NCC workgroups  
**Who with:** All Regional Collaboratives, NCC  
**Accomplishments:** To be determined by the NCC

**Title:** 2016 Regional Meeting  
**Description:** Annual meeting held from October 17-18, 2016  
**Who with:** 62 stakeholders from seven states  
**Accomplishments:** Plenary panels on day one featuring speakers addressing telegenetics, genetics education, quality improvement and health care access, and health equity. On day 2, participants broke into groups addressing these same topics for facilitated strategic planning sessions to understand the priorities as the collaborative plans for the future.

**Title:** Journey through Diagnosis guide for families  
**Description:** Family Forum completed the development of a guide for families that have recently been diagnosed with a genetic condition  
**Who with:** Members of Region 4 Midwest’s Family Forum  
**Accomplishments:** The booklet was created, written, and edited with input from the Region 4 Family Forum, which is comprised of families of children diagnosed or identified with a wide variety of genetic conditions. It provides helpful tips, information and insight for families of a recently diagnosed child(ren). Topics covered include managing emotions, finding support, family relationships and challenges, identifying resources and cultivating positive parent/professional relationships within a medical home. To date,
1415 guides have been requested for distributed in print and 438 have been downloaded from the Region 4 Midwest website to date.

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<th><strong>Affordable Care Act</strong></th>
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| **Title:** Metabolic Foods & Formulas online toolkit  
**Description:** The Medical Foods and Formula Toolkit was developed to be a resource for state health departments, advocates, patients, and their family members across the United States working towards better coverage of medical foods and formulas for individuals diagnosed with inborn errors of metabolism or other conditions.  
**Who with:** Region 4 Long-Term Follow-Up Workgroup  
**Accomplishments:** Online toolkit that offers resources for state programs and resources for patients and families. |
| **Title:** Medicaid Coverage for Genetic Testing and Services  
**Description:** Performed an inventory across regional Medicaid programs for current policy and coverage related to genetic testing and services.  
**Who with:** NCC Health Care Access and Financing Workgroup, Regional state Medicaid programs  
**Accomplishments:** Working in partnership with Region 4 Midwest state leads and respective Medicaid programs, gathered state level Medicaid policy information for the reimbursement and coverage of genetic testing and genetic services. |

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<thead>
<tr>
<th><strong>Care Coordination: Empowering Families Training Insurance Module</strong></th>
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</table>
| **Title:** Care Coordination: Empowering Families Training Insurance Module  
**Description:** Insurance module of training includes information about the ACA as it relates to CYSHCN  
**Who with:** Partners in all 7 R4 states  
**Accomplishments:** Continue to include substantive module on the ACA and insurance issues within the training. Provided the Catalyst Center’s “The Affordable Care Act: A side-by-side comparison of major provisions and the implications for children and youth with special health care needs.” |

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<th><strong>Long-Term Follow-Up</strong></th>
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| **Title:** Congenital Hypothyroidism 3-Year Follow-Up Project: Results from the Region 4 Midwest Genetics Collaborative  
**Description:** Region 4 manuscript that identifies the 3-year follow-up management and education patterns of primary care clinicians and pediatric endocrinologists for children diagnosed with congenital hypothyroidism (CH) by newborn screening programs.  
**Who with:** Kupper Wintergerst and partners in all R4 states |
| **Title:** Inborn Errors of Metabolism Project—Data Entry  
**Description:** Ongoing collaboration with IBEMC and NIH funded metabolic centers in national effort to collect data on metabolic disorders  
**Who with:** IBEM-IS |
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<tr>
<th>Accomplishments: Manuscript under review with <em>Pediatrics</em>.</th>
<th>Accompishments: Financial support for IBEM-IS data entry for two additional centers in Region 4: the Medical College of Wisconsin and the University of Louisville, Weisskopf Child Evaluation Center.</th>
</tr>
</thead>
</table>
| Title: Regional Participation in the NCC LPDR LTFU Database  
**Description:** Facilitated regional recruitment in the National Coordinating Center Pilot Long-term Follow-up module in the LPDR  
**Who with:** NCC Long-term Follow-Up Workgroup, Region 4 Midwest Public Health Long-term Follow-Up Workgroup  
**Accomplishments:** Facilitated the recruitment and participation on Region 4 Midwest states in the LPDR Long-term Follow-up Pilot project through the NCC. Michigan and Minnesota moved forward with participation. |  
**Title:** Care Coordination: Empowering Families Transition Module  
**Description:** Transition module of training emphasizes the need to plan for transition for children with genetic conditions  
**Who with:** Partners in all 7 R4 states  
**Accomplishments:** Continued to highlight the importance of planning for transition to adult care within the training. Participants with older children regularly site this module as informative and important. |
| **Title:** Children’s Special Health Care Services Online Transition Course  
**Description:** Developed content for online transition course.  
**Who with:** Michigan CSHCS, Michigan F2F  
**Accomplishments:** Worked in partnership with the Michigan children’s special health care services program and Michigan family to family health information venter to develop content for an online transition course. The result was six modules of information and resources geared towards parents and young adults with special health care needs including genetic conditions. |
### Telegenetics

**Title:** Telegenetics Training  
**Description:** Provided support for three regional representatives to attend and participate in the Telegenetics Training at the South Central Telehealth Forum 2016  
**Who with:** Western States and Heartland Genetics Collaboratives, South Central Telehealth Resource Center  
**Accomplishments:** Continued partnership and support for the Western States and Heartland Regional Genetics Collaboratives telegenetics training program. This opportunity allowed the Midwest to train providers in telegenetics and to observe the curriculum for future support and implementation in the region.

### Needs Assessment

**Title:** Mapping Projects  
**Description:** Completed analyses of MI Newborn Screening mapping project and began planning for seven-state baseline maps of underserved populations to be completed by May 31, 2017.  
**Who with:** MI Newborn Screening Program; geographer consultants  
**Accomplishments:** Presented findings from the MI Newborn Screening mapping project at the 2016 Regional Meeting in October. Results indicated that not just rural patients, but any genetics patient living at a great distance from their specialist provider may be consider underserved.

**Title:** Genetic Systems Assessment Tool  
**Description:** Dr. Ann Chou delivered GSA results to Region 4 this year and presented them at the 2016 Regional Meeting  
**Who with:** Heartland Region, Dr. Ann Chou  
**Accomplishments:** Dr. Chou delivered aggregate and state-specific reports. She traveled to the Regional Meeting to present findings and discuss possible quality improvement efforts moving forward.
# Heartland Genetics and Newborn Screening Collaborative Regional Activities

## Intra-Regional

**Title:** Individualized Health Plan (IHP)

**Description:** The goal of this project is to identify ways to improve the process of developing, communicating, implementing, and enforcing an individualized student health plan.

**Who With:** MO, ND, KS, AR

**Accomplishments:** Implemented the new process with 10 individuals (school nurses, school nurse administrators) over a 5 month period resulting in nearly 400 additional schools trained in the IHP process and over 4,800 plans developed. Moreover, the number of IHP documents linked to an IEP or 504 plan has more than doubled.

## Inter-Regional

**Title:** Hispanic Access Advisory Council (HAAC)

**Description:** The goal of this project is to identify the barriers to accessing genetic services for Hispanic patients and to identify strategies to support genetic service providers in providing culturally competent quality care. Interviews (26) were conducted with 26 families across three Heartland states. These results were analyzed and finding were shared with the HAAC. Eight recommendations were identified and the top three strategies were prioritized. The HAAC has begun to identify implementation activities to address these three priority areas.

**Who With:** Heartland and Mountain states have representatives on HAAC.

**Accomplishments:** HAAC met to review the findings and determine implementation priorities to improve improved access to genetic service.

## State Public Health Capacity Building

**Title:** Collaborative Improvement and Innovation Network for Timeliness in Newborn Screening (CoIIN)

**Description:** The Iowa NBS program received an award from NewSTEPSs for the planning and implementation of a Collaborative Improvement and Innovation Network (CoIIN) for timeliness in newborn screening two years ago. Heartland supported additional states in the region to convene and extend the CoIIN project across the region. The rationale was to enable programs to engage in quality improvement activities through shared learning of evidence-based strategies for improving timeliness within their program’s newborn screening system.

**Who With:** KS, IA, AR, MO, ND, and OK

**Accomplishments:** States convened a CoIIN team of newborn screening stakeholders and met in Kansas City May 5-6,
2016. NewSTEPs facilitator presented on the quality improvement process, states reported on the specific section(s) of Quality Indicator 5 that pertains to their state project, and states shared barriers and strategies. All participants ranked the training as valuable or very valuable. After the meeting, Iowa, Kansas, Nebraska, North Dakota, and Oklahoma received awards under NewSTEPs 360 grant to improve timeliness in their NBS programs.

<table>
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<tr>
<th>Collaboration</th>
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| **Title:** Youth Advocate Mentoring  
**Description:** The goal of the mentoring project is to develop advocacy skills in youth who have a genetic/chromosomal condition.  
**Who With:** OK, NE, IA  
**Accomplishments:** 6 mentors trained; mentees identified; attendance at the annual regional meeting to meet one another; matching and mentoring in process. |

<table>
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<tr>
<th>Long-Term Follow-Up</th>
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| **Title:** IBEM-IS  
**Description:** The project’s purpose is to develop a long-term follow-up database and to track the treatments, health, and developmental outcomes of the patients with inborn errors of metabolism.  
**Who with:** Heartland and Region 4  
**Accomplishments:** Participation in these pilots will help to facilitate the states adoption of case definitions for the recommended uniform screening panel. |

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<tr>
<th>Transition</th>
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| **Title:** Healthcare Transition for individuals with genetic conditions  
**Description:** The purpose of the overall project was to determine the role of the genetics team in healthcare transition. In the current project year, the transition clinic and curriculum have continued to be implemented and refined. |
**Who with:** In the past project year, all of the work has been conducted in South Dakota.

**Accomplishments:** 13 trainees (4 residents, 1 genetic counselor, and 8 genetic counseling students) completed the curriculum. Nine patients served in the clinic. Two abstracts accepted at national meetings (Transition conference and AMCHP) for poster presentations.

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**Telegenetics**

**Title:** Telegenetics  
**Description:** Educated genetics residents, genetic counseling students and those in practice.

**Who with:** Western States (co-developed curriculum); participants for training were from SERC, Region 4 Midwest, and Mountain States

**Accomplishments:** Conducted one training involving 14 participants, which included physicians, genetic counselors, nurse practitioners, and a genetic counseling trainee.
# Mountain States Genetics Regional Collaborative (MSGRC) Regional Activities

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<thead>
<tr>
<th>Intra-Regional</th>
<th>Inter-Regional</th>
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<tr>
<td><strong>Title:</strong> Parent Partner Project</td>
<td>Medical Home</td>
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<tr>
<td><strong>Description:</strong> Parents of children with special healthcare needs are trained to assist other families with special needs to navigate the non-medical components of the medical home.</td>
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<td><strong>Who with:</strong> Project Lead, Brad Thompson, Hali Project; Montana Health Department, Wyoming Health Department</td>
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<tr>
<td><strong>Accomplishments:</strong> The Parent Partner Project is currently in eight pediatric practices and has expanding to a reservation site and Air Force base site, both in Wyoming. The project in Montana is now completely funded out of their CYSCHN budget. A second evaluation was undertaken in 2015-2016 to measure the impact the program is having on family’s abilities to identify and access resources. MSGRC is in early phases of an article for potential journal submission.</td>
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| **Title:** Texas Primary Care and Health Home Summit | |
| **Description:** In Year 4, MSGRC funded the attendance of 8 members (1 pediatrician, 7 consumers) to the health home summit. In Y5, we collected feedback from attendees. Accomplishments: One consumer member, Joe Martinec, wrote an article on his attendance that was shared through MSGRC (PD message and Facebook). It was also submitted to the steering committee of the conference. | |

## NBS Capacity Building

| **Title:** Hemoglobinopathies Interest Group (HIG) | **Title:** Participation in Advisory Committee on Heritable Disorders in Newborn Children (ACHDNC) |
| **Description:** Dr. Kathryn Hassell leads the MSGRC HIG. Dr Hassell continues to serve on the Sickle Cell Guidelines Expert Panel Membership at the NHLBI. While the final guidelines were released in 2014, MSGRC has continued to disseminate guidelines to its members in 2015-2016. The HIG is planning a meeting for April 2017 as part of the MSGRC Annual Meeting. Who with: Kathryn | **Description:** In addition to MSGRC staff attendance at quarterly meetings, MSGRC staff are active participants in ACHDNC workgroups. Joyce Hooker (Regional Outreach) is a member of the education committee and Drs. Hassel, Thomas, and Kaye are members of the Long term Follow- |

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NCC/RC National Evaluation Brief
Hassell, MSGRC members, other stakeholders

**Accomplishments:** Approximately twenty MSGRC members attended the April 2016 in-person HIG meeting. Feedback on dissemination and usefulness of the new guidelines was solicited from attendees.

**Who with:** All MSGRC leadership staff

**Accomplishments:** MSGRC staff attended all ACHDNC meeting in Y5. MSGRC Directors also attended meetings in-person when possible.

**Title:** Metabolic Consortium (MC)

**Description:** Dr. Janet Thomas directs the Metabolic Consortium. The Metabolic Consortium meets in-person every year to discuss patient care guidelines, case studies, and other regional developments.

**Who with:** Janet Thomas, MSGRC members, other stakeholders

**Accomplishments:** The MC continues to be a forum for distribution and feedback on the previously developed shared datasets. Members have the opportunity to bring case studies for discussion. Quality of newborn screening within the region is discussed including false negative tests or unique situations, new pilot disorders for population screening, and second tier laboratory testing. Dr. Mark Korson from Genetic Metabolic Center for Education (GMCE) also attended, to educate attendees on his group’s work and purpose and gather feedback. The Metabolic Consortium is planning an in-person meeting 4/20/17 as part of the MSGRC Annual Meeting.

**Title:** Strategic Planning Committee, November 2016

**Description:** MSGRC held one in-person meeting in the first 9 months of year 5. In November 2016, stakeholders from across the region convened to review the needs assessment and prioritize activities/strategies for the Mountain States Region moving forward.

**Who with:** MSGRC members and other leaders in the region.

**Accomplishments:** 17 stakeholders participated. Strategies from the meeting include engaging primary care and genetics clinics from across the region, expanding telegenetics and outreach clinics, and identifying and reaching underserved populations (rural, American Indian, and Hispanic).

**Title:** NCC Regional Support Service Model (RSSM) Workgroup

**Description:** The workgroup, chaired by Dr. Kaye, was charged with reviewing existing regional and national models of genetic services delivery as a basis to develop a model for HRSA that would be used for 2017 and beyond.

**Who with:** Celia Kaye, MSGRC staff, and representatives from other RCs and partnering organizations

**Accomplishments:** A final brief of a proposed model was developed and submitted to HRSA. Dr. Kaye’s leadership was vital in the development of this workgroup and MSGRC actively attended meetings and provided comment during the open
The work of the RSSM Workgroup provided much of the foundation for activities of MSGRC’s Strategic Planning Committee.

The MSGRC Annual Meeting is scheduled for April 2017. It will engage other federally funded partners, other RCs, and new clinical and consumer members.

**Title**: Newborn Screening Translational Research Network (NBSTRN)

**Description**: Drs. Kathryn Hassell and Janet Thomas are members of the NBSTRN’s Clinical Integration Workgroup which facilitates implementation of the LPDR and reviews the integrity of tools and projects of the NBSTRN.

**Who with**: Kathryn Hassell, Janet Thomas, and other workgroup members

**Accomplishments**: Dr. Hassell co-chairs the Clinical Integration WG along with Dr. Susan Berry. Dr. Thomas is an active member.

**Title**: Hispanic Access Advisory Council (HAAC)

**Description**: Dr. Janet Thomas, Marilyn Brown, and Dr. Margarita Saenz have all served as members on Heartland’s HAAC.

**Who with**: MSGRC representatives, Heartland Genetics Collaborative, other providers

**Accomplishments**: To date, the HAAC has developed a survey to assess needs of Early Intervention (EI) staff that work with families of children with hereditary disorders. Heartland staff will also be presenting at the MSGRC Annual Meeting.

**Title**: Adaptation of the 2015 Policy Brief

**Description**: MSGRC is in the early stages of planning an adaption of our previous policy brief, depending on changes that will be made to the Affordable Care Act.

**Who with**: MSGRC staff and Christine Cardinal, PhD, JD, Professor of Health Policy at Sam Houston State University.

**Accomplishments**: Dr. Cardinal is continuing her involvement with MSGRC and will be attending the MSGRC Annual Meeting. She had expressed her interest in leading writing of the policy brief as

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**Affordable Care Act**
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<tr>
<th>Title: Long-term Follow-up</th>
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<tr>
<td><strong>Description:</strong> Dr. Janet Thomas continues to have an active research protocol to consent children and families diagnosed with conditions via newborn screening into long term follow-up in association with Inborn Errors of Metabolism Information System (IBEM-IS). MSGRC funds the participation in the IBEM-IS.</td>
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<td><strong>Who with:</strong> Dr. Thomas is the local site principle investigator; Melinda Dodge, MSGRC consumer member, is the study coordinator.</td>
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<td><strong>Accomplishments:</strong> Patients continue to be consented into the protocol as appropriate. Focus of year 5 is data entry into the IBEM-IS.</td>
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<thead>
<tr>
<th>Title: ACHDNC Long-Term Follow-Up</th>
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<tr>
<td><strong>Description:</strong> Drs. Kathryn Hassell, Janet Thomas, and Celia Kaye are members of the ACHDNC Long-Term Follow-up and Treatment Workgroup.</td>
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<tr>
<td><strong>Who with:</strong> Drs. Kathryn Hassell, Janet Thomas, and Celia Kaye and other national participating members</td>
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<tr>
<td><strong>Accomplishments:</strong> Dr. Hassell and Dr. Thomas have been able to attend and represent MSGRC at all In-Person ACHDNC meetings. Dr. Kaye continues to participate through webinar attendance. All are active participants of the LTFU Quality Measures sub-workgroup</td>
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<tr>
<th>Title: Genetic Services Mapping Project</th>
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<td><strong>Description:</strong> MSGRC continued work on an evolving mapping project by conducting interviews with genetics providers to ask about provider capacity, delivery of outreach and telemedicine services, and geographic region served.</td>
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<td><strong>Who with:</strong> MSGRC staff, UC Denver Student (via practicum opportunity); Genetics clinics in the region</td>
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<td><strong>Accomplishments:</strong> Completed surveys of 12 clinics in the region; mapping to be completed by the end of Year 5.</td>
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<tr>
<th>Title: Recruitment of Providers for Telegenetics Training</th>
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<td><strong>Description:</strong> Western States and Heartland have developed a curriculum for telegenetics training and has offered training sessions in Nashville and Phoenix. MSGRC sponsored the attendance of clinicians in our region.</td>
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<td><strong>Who with:</strong> MSGRC staff and members, Heartland Collaborative, Western States Collaborative</td>
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<tr>
<td><strong>Accomplishments:</strong> Five clinicians from MSGRC attended the telegenetics training in Nashville, TN sponsored by Heartland Genetic Services Collaborative. Attendees include 1 geneticist, 1 medical genetics resident, and 3 genetic counselors all from University of Colorado and the Children’s Hospital Colorado. One genetic counselor attended training in Phoenix sponsored by Western States Genetics Collaborative.</td>
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<th>Title: Cook Children’s Telegenetics Case Study</th>
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<tr>
<td><strong>Description:</strong> MSGRC completed an in-depth case study of the telegenetics service line at Cook Children’s Hospital in Fort Worth, TX.</td>
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Who with: MSGRC staff, MSGRC Telegenetics Workgroup Members, Cook Children’s staff/clinicians

Accomplishments: Data collection completed via telephone interviews and email transmission of quantitative data. The Case Study was accepted for an oral presentation at the 2017 ACMG Annual Meeting (Dr. Janet Thomas presenting). A manuscript is also under development.

Other

Title: NCC PD/PM presentation on underserved populations
Description: MSGRC presented on outreach to American Indian populations.
Who with: Trish Thomas of Family Voices and Joyce Hooker with MSGRC
Accomplishments: Ms. Hooker presented on MSGRC’s past projects, such as the mHealth project in Tuba City, that have reached American Indian populations. She also discussed the listening sessions held in American Indian communities. Ms. Thomas was able to offer perspective on cultural sensitivity when working with diverse populations and best approaches.

Title: ACMG Community Conversation
Reaching Minority Populations: Genetics in the Frontiers
Description: Trish Thomas with MSGRC presented as part of the open forum section of ACMG’s Community Conversation at the 2017 ACMG Annual Meeting. She shared her experience of how the medical community can best serve American Indian populations with a culturally appropriate approach.
Who with: Trish Thomas, presenters from Heartland Genetics Collaborative, NCC
Accomplishments: The Community Conversation is open to all attendees at the ACMG conference and is well attended, with over 200 in attendance in past years.
**Western States Genetic Services Collaborative (WSGSC) Regional Activities**

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<tr>
<th>Intra-Regional</th>
<th>Inter-Regional</th>
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| **Medical Home** | **Title:** Genetic Outreach Visit Outcomes  
**Description:** Measure of patient/family perception of outcomes of genetic visits provided via outreach clinics.  
**Who with:** Public health genetics in Hawaii and Alaska.  
**Accomplishments:** Hundreds of patients have received genetic services they would not have received or would have had to travel out of state for, understanding of patient experience of outcomes of genetic services. |
| **NBS Capacity Building** | **Title:** Genetic Outreach Visit Outcomes  
**Note:** Used by other regions for their evaluation activities. |
| **Title:** Newborn Screening Parent Fact Sheets  
**Description:** Parent fact sheets for the disorders included on the RUSP are developed, updated, and maintained by the WSGSC on our companion website, [www.newbornscreening.info](http://www.newbornscreening.info). Pompe fact sheet has been added. X-ALD has been created and is under initial review.  
**Who with:** AK, CA, HI, ID, OR, WA, and Guam partners.  
**Accomplishments:** The website averages 28,000 unique visitors per month. The Spanish language fact sheets were recently updated. | **Title:** Newborn Screening Parent Fact Sheets  
**Description:** Parent fact sheets for the disorders included on the RUSP are developed, updated, and maintained by the WSGSC on our companion website, [www.newbornscreening.info](http://www.newbornscreening.info). Pompe fact sheet has been added. X-ALD has been created and is under initial review.  
**Who with:** Shared and used in numerous states.  
**Accomplishments:** The website averages 28,000 unique visitors per month. The Spanish language fact sheets were recently updated. |
| **Collaboration** | |
| **Title:** Annual Regional Summit  
**Description:** Public health genetics and NBS leaders, family advocates, genetic specialists, primary care representatives, LEND and Title V leaders convene to share information and develop regional priorities.  
**Who with:** AK, CA, HI, ID, OR, WA, and Guam partners.  
**Accomplishments:** Yearly regional genetics projects, ongoing vehicle for state to state exchange of information and best practices. |
**Title**: Genetic Assessments for Title V Programs  
**Description**: In-person or videoconference meetings to collect genetic service assessment information for each state.  
**Who with**: State Title V programs and other state genetic service providers.  
**Accomplishments**: Contacted each state Title V program to conduct assessment meetings or key informant interviews to obtain information regarding their perceived needs related to genetic services.

**Title**: Newborn Screening Survey  
**Description**: Parent opinion about notification preferences  
**Who with**: WSGSC region  
**Accomplishments**: Completed analysis and report on findings.

**Title**: Newborn Screening Survey  
**Description**: Have interest from the other regions to do same activity.

**Affordable Care Act**  
**Title**: The ACA—Through the Life Course Website  
**Description**: WSGSC web pages devoted to explaining the ACA to families in relation to their life stages  
**Who with**: Regional family, public health and health providers in AK, CA, HI, ID, OR, WA, and Guam  
**Accomplishments**: The website is routinely updated and continues to be well received.

**Title**: The ACA—Through the Life Course Website  
**Description**: Disseminated by other regions as a resource.

**Long-Term Follow-Up**  
**Title**: CPT 1A Arctic Variant Project  
**Description**: The project seeks to improve newborn detection, management, and long-term follow-up of children with carnitine palmitoyl transferase deficiency, type 1 (CPT-1A) Arctic variant.  
**Who with**: Dave Koeller, MD from the Oregon Health and Science University, the Alaskan Native Health organizations, Alaskan Native tribal groups, and the Alaska Department of Health.
**Accomplishments:** Dr. Koeller was able to take his work with the WSGSC to apply for and be awarded a NIH grant to do the long term follow-up project.

**Telegenetics**

**Title:** Technical assistance for telegenetics  
**Description:** Providing technical assistance for the states in our region to increase services and education provided by telegenetics.  
**Who with:** Washington Genetics Program, California and Alaska NBS programs.  
**Accomplishments:** Provided telegenetics protocols and information to help the development of telegenetics activities in the other states. Planned, developed and implemented ten new on-line modules for the telegenetics training.

**Title:** Telegenetics Education and Training  
**Description:** Two online webinars, a set of ten online modules and one-day, in-person training session for genetic counselors and administrators on the use of telemedicine in clinical genetics practice.  
**Who with:** Heartlands Regional Genetics Collaborative and HRSA-funded Telehealth Resource Centers in Arizona and Arkansas.  
**Accomplishments:** Two groups of genetic counselors attended the Telegenetics Training in August 2016, one in Tucson, AZ and one in Kent, WA. We also held a telegenetics training for a group of WA program administrators in Kent, WA. The next round of training is planned for April 2017.

**Other**

**Title:** Genetic Services Outcomes  
**Description:** Completed literature review and wrote framework for approach necessary to measure outcomes of genetic services, including information specific to telegenetics.  
**Who with:** NCC – all regions  
**Accomplishments:** All RCs now have access to information about current practices in outcomes measurement for genetics, foundation laid for proceeding with Telegenetics outcomes measurement, initiated process for expert development of consensus outcomes.
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SUPPLEMENT 4. NCC and NGECN Highlights

In 2016-17, the American College of Medical Genetics and Genomics completed work on the second year of its HRSA award as the National Coordinating Center (NCC) for the seven Regional Genetic Collaboratives. Because of the May 31, 2017 end to this grant cycle and with input from the HRSA Project Officer, the NCC/NGECN data in this report reflects nine months of program activity (June 1, 2016 to February 28, 2017).

How much did you do?

• As a part of its mission to strengthen communication and collaboration between the RCs, the NCC hosts regular **RC/NCC leadership calls** and **NCC workgroup calls** (n=45). These exchanges promote common approaches to HRSA priority areas and rapid dissemination of innovative ideas among the more than **420** participants. (See NCC summary for a table depicting NCC Workgroup activities.) Staff from **NewSTEPS** participate on the Project Director/Project Manager (PD/PM) calls.

• NCC staff participate in the meetings of the **Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC)**, in NewSTEPS workgroups/committees, on the Association of University Centers on Disabilities (AUCD) Leadership Education in Neurodevelopmental Disabilities (LEND) Genetics Workgroup, and several national committees of the AMA and NCQA.

• NCC leadership (PI, PM, and Medical Director) participated in and made presentations at five **RC annual meetings** (SERC in July, WSGSC in Sept., NYMAC, Heartland, and Region 4 in October). The NCC staff also presented and exhibited at the ASHG, AMCHP, and ACMG annual meetings.

• To promote the framework for regional genetic service models and to share information from the consumer and provider needs assessments, the NCC presented posters at the **American Society of Human Genetics**. These posters were entitled **Improving Access to Genetic Services- Provider and Consumer Needs Assessment Data from the National Regional Genetics Collaboratives and Regional Genetic Support service Models- What’s out there and what could the U.S. learn for future genetic services delivery models?**

• The NCC provides the RCs and our partners with monthly calendars of events and with the **Collaborator** – the newsletter of the NCC/RC system. The July 2016 edition focused on Partnering with Consumers; the November 2016 release highlighted Telegenetics.

How well did you do it?

• During 2016-17, the NCC continued to support workgroup activities in the **HRSA priority areas** of Long-term Follow up, Health Care Access, Care Coordination, and Telegenetics/Distance Strategies. An Evaluation Workgroup is also supported by the NCC.

• **ACT Sheets** are being utilized by clinicians. There were **3,717 unique visits and 4,432 page visits** to ACT Sheets. Some **567 units** of the ACT Sheet App were downloaded (Apple and Google Play users).

• Through supplemental funding from HRSA, the NCC formed a **NBS Hearing Screening Workgroup** to develop a guideline and manuscript that provides options to improve early detection of hearing loss before the age of 5.
Was anyone better off?

- More babies are being screened in States and Territories which adopted the recommended uniform screening panel (RUSP) for critical congenital heart disease (CCHD) and severe combined immunodeficiency (SCID). As of February 2017, **50 States and territories** screened all newborns for CCHD and **46 States and territories** screened for SCID.

- The integration of genetics resources into **Leadership Education in Neurodevelopmental Disabilities (LEND)** programs means that more LEND trainees are prepared to address genetic and genomic issues.

- Using the NCC/RC system resources, more consumers have access to the information they need to help themselves or their families. Providers have been trained or have NCC/RC resources to deliver up-to-date genetic services.

### NCC Workgroup 2016-2017* Activity Highlights

<table>
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<tr>
<th>NCC Workgroup</th>
<th>Mission</th>
<th>2016-2017* Activities</th>
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| ACT Sheets             | Clinical resources               | An ACT Sheet on Pompe was developed, but before releasing it, the workgroup will revisit the Pompe algorithm.  
An in-person meeting is planned on May 12-13, 2017 in Washington, D.C. in conjunction with the ACHDNC meeting. |
| Healthcare Access and Financing | Improve insurance coverage | Work continues in three subgroups:  
The Identifying Genetics Services subgroup conducted two webinars in July with presentations from Dr. Ned Calonge, President and CEO of the Colorado Trust and Chair of the EGAPP group, and Dr. Marc Williams, Director of the Genomic Medicine Institute at the Geisinger Health System.  
The Coverage of Genetic Services subgroup collected Information about state Medicaid policies on genetic services.  
The Consumer Role in Genetic Services subgroup created a Consumer Advocacy Resource Database. |
<p>| Care Coordination      | Promote medical home transition, and family health history | Developed a driver diagram to frame the issues on care coordination. To wrap up their deliberations for this grant cycle, they will produce a white paper for HRSA. |</p>
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<tr>
<th>Category</th>
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<tbody>
<tr>
<td>Used the results of webinars</td>
<td>That were conducted in April and May 2016, to determine future directions.</td>
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<td>Build public health capacity</td>
<td>14 States joined in the pilot of the Longitudinal Pediatric Data Resource and worked to refine the public health questions that LTFU should address.</td>
<td>Collaboration continues with the Inborn Errors of Metabolism Consortium on a large dataset on long-term follow-up of individuals with inborn errors. Participation on and presentations were made to the ACHDNC.</td>
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<tr>
<td>Improve access to genetic</td>
<td>Promoted training in the use of telemedicine for genetic service professionals.</td>
<td>Presented the results of a literature review to inform the selection of health outcome measures for genetic services delivered through distance strategies. Held an in-person WG meeting on October 28th to discuss telegenetics activities (regionally and nationally), and the development of health outcome measures.</td>
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<tr>
<td>Complete the HRSA Evaluation</td>
<td>Completed the HRSA Evaluation Brief: Year 4 Evaluation Summary Data and distributed it on September 28th. This report uses a framework of common evaluation data elements to demonstrate the impact of the NCC/RC system.</td>
<td>Presented posters with the consumer and provider needs assessments data at ASHG and ACMG annual meetings. Engaged the evaluators in the telegenetics health outcomes measurement work. Served as a forum for sharing regional needs assessment activities.</td>
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<tr>
<td>Project Directors/Managers</td>
<td>Participated in the annual PD/PM annual meeting in November 2016 and on NCC Workgroup calls.</td>
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<tr>
<td>Partnerships with other HRSA funded programs</td>
<td>Supported an Advocate Leaders’ Mentoring Program held at ACMG’s Clinical Genetics Meeting in March.</td>
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*Because of the May 31, 2017 end to this grant cycle and with input from the HRSA Project Officer, the NCC/NGECN data in this report reflects nine months of program activity (June 1, 2016 to February 28, 2017)*
The National Genetics Education and Consumer Network
Year 5 (Continuation), Select Data
June 1, 2016–February 28, 2017

In the final year of the two-year project continuation, the NGECN worked to finalize and disseminate the publications and products developed during the project, including the submission of a journal article, distribution of a monograph, and launch of an updated website. Project activities during the past year demonstrated success towards the goal of improved access in the following ways:

**How much did you do?**

- Served as national representative for the RC network and their family advocates workgroups, participating on monthly calls, presenting at 2 RC annual meetings, and serving as faculty for the NCC Advocates Leader Program
- Wrote and submitted an article based upon a 34-question national needs assessment entitled *Understanding Access to and Quality of Genetic Services: The Individual/Family Perspective*
- Wrote and designed a monograph titled *Next Steps for Patient Navigation: Perspectives from Partners in Care* based on 18 semi-structured interviews with hospitals, advocacy networks, and community health centers
- Presented consumer needs assessment data and field survey results at 2 national and 2 regional meetings to experts tasked with designing measurable interventions to address gaps in services
- Organized a presentation for 12 family advocates titled *Advocates Luncheon: Gene Editing* as part of the American Society of Human Genetics meeting
- Published 6 Exceptional Parent Magazine articles and 3 consumer perspective pieces in the journal *Genetic Testing and Molecular Biomarkers*
- Facilitated connections between relevant support and information resources and HRSA-funded programs, including Family Voices, Parent to Parent USA, and the Genetics and Rare Disease Information Center (3 webinars/events).
- Tagged a list of over 4,500 diseases by 55 categories to allow resource, clinical trial, and support matching
- Developed a 175 question survey, including the validated CSHN measures, for distribution through the PEER registry as part of DiseaseInfoSearch.org
- Added 168 curated, patient-centered, and low literacy condition descriptions to diseaseInfoSearch.org
- Served as an advisor to Heartland’s Hispanic Access Advisory Group (4 calls, 1 in-person meeting), strategizing ways to address barriers to care experienced by Hispanic families
- Distributed over 6,000 family health history materials to families and clinics

**How well did you do it?**

- *Partners in Care* monograph synthesized 14 distinct patient navigation programs into 8 scalable models, showing evidence as an intervention to improve care and impact needs expressed by families
• 60,904 individuals accessed information on genetics and health, testing, and other genetic services through GenesInLife.org
• 6,000 family health history booklets were requested (100% delivered) by organizations and family members and through a national E-card campaign

**Was anyone better off?**
• 4,175 individuals accessed links to support groups through DiseaseInfoSearch.org
• Continuing discussions with family advocates across HRSA-funded MCH programs has widened the breadth of engagement in efforts to distill consumer needs and priority areas for future genetic services programs