HRSA Evaluation Brief

Year 4 Evaluation Summary Data

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

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Cooperative Agreement: #U22MC24100
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Executive Summary

This is the fourth year that the NCC/RC system has used a framework of common evaluation data elements related to Health Resources and Services Administration’s (HRSA) priorities and to Healthy People 2020 objectives that focus on care coordination (e.g., medical home and transition from pediatric to adult care); telegenetics; newborn screening capacity and long-term follow-up. 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).

This year, the Evaluation Workgroup focused on the national needs assessment activities, providing input on the survey instruments used to reach consumers and providers. Each monthly Evaluation Workgroup agenda has time for the RC evaluators to share their local needs assessment plans and projects with a goal toward finding common metrics for a redesigned NCC/RC system. The Workgroup also reviewed and the NCC submitted comments on the MCH Discretionary Grant Information System (DGIS) measures.

In 2015-16, the 7 RCs continued with year 4 of their 5-year funding cycle. As part of its new funding cycle (which is offset from the 7 RCs), the American College of Medical Genetics and Genomics (ACMG), as the National Coordinating Center (NCC) for the seven federally-funded Regional Genetic Service Collaboratives (RCs), focused its efforts on a national needs assessment to improve access to genetic services. Designed to collect information from consumers and providers, this effort informs HRSA’s development of innovative models of support centers for regional genetic and genomic services and resources that will be useful for the next 5 to 10 years.

The NCC established a Regional Support Service Model Workgroup (RSSM WG) and an Advisory Committee (AC); both were chaired by Celia Kaye, M.D, PhD. Their 10-months of deliberations culminated on March 31st with a set of recommendations based on eight models, potential outcome and process measures, and additional features for HRSA to consider in the redesign of the NCC/RC system. The RSSM WG’s deliberations were informed by listening sessions and national surveys. The Genetic Alliance-led (as a subcontractor to the NCC) National Genetics Education and Consumer Network (NGECN) took the lead on the design of the consumer survey. The NCC Evaluation Workgroup and other NCC workgroups provided input on both the consumer and provider surveys. As a result of this work, some of the NCC/RC system priorities have shifted.

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.
KEY REGIONAL FINDINGS 2015-2016

- RCs worked with more than 3,900 consumers, health and public health professionals and had 376 family, disease-specific and advocacy organizations on their mailing lists.

- All RCs offered education and training sessions; these reached more than 7,814 participants, 61% of whom were in-person meetings; 30% in webinars and 9% by teleconferences. The number of training and education sessions has increased every year from 145 sessions in year 1; 249 sessions in year 2; 411 sessions in year 3 to 427 sessions in year 4.

- All RCs had websites that addressed the HRSA priorities of medical home, newborn screening, and the Patient Protection and Affordable Care Act (ACA). These websites had more than 81,460 unique visits and 121,298 home page visits.

- All RCs use social media; they reported having 1,898 Facebook followers as of May 31, 2016. Three RCs (NEGC, SERC and Region 4 Midwest) and the NCC use Twitter with 554 followers and two RCs (Heartland and WSGSC) and the NCC use You Tube with 4,635 video views.

- All RCs addressed medical home. Using the HRSA Performance Measure #41 Medical Home, the RCs recorded annual increases in their average scores from 18 in year 1 to 31 out of a possible 72 in year 4 – a 72% increase.

IMPACT OF THE RC ACTIVITIES

- A consistent method for screening and reporting of CCHD across New England has been established. Over 42,000 children have been screened since the project started, three children were diagnosed with CCHD.

- The NYMAC-funded project at Children’s Hospital of Pittsburgh (CHP) Pediatric Hematology successfully conducted telemedicine visits with patients with sickle cell disease. They increased the frequency of visits (from twice yearly to quarterly visits) and improved compliance with hydroxyurea therapy (73% of pediatric patients).

- A collaboration between SERC, Genetic Metabolic Dietitians International (GMDI) members, parent organizations, and the Agency for Healthcare Research and Quality (AHRQ) resulted in PKU guidelines being completed. These evidence-based guidelines were published in a peer-reviewed journal and posted on a web-based portal and MSUD guidelines were accepted and released through the AHRQ.

- The Care Coordination training continues to impact families across the country. Region 4 Midwest trained an additional 99 caregivers in five trainings offered in collaboration with the Michigan (MI) Department of Health and Human Services. The Heartland Collaborative trained approximately 80 caregivers across 11 partnering agencies. To date, over 400 caregivers across the country have been trained.

- The Region 4 Newborn Screening: What Prenatal Care Providers Need to Know Online Course trained 102 prenatal care providers, child birth educators, and other health care professionals from across the U.S.

- 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 being 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.

- Newborn Screening Parent Fact Sheets for the disorders included on the Recommended Uniform Screening Panel (RUSP) are developed, updated, and maintained by the WSGSC on www.newbornscreening.info. A Pompe fact sheet is under expert review; a XALD is in development. The website has 30,000 unique visitors per month. The Spanish language fact sheets were recently updated.
Through the leadership of the Heartland and with participation from WSGSC, MSGRC, NYMAC, Region 4 Midwest, and SERC, 32 States have completed the Genetic Systems Assessment tool (past and current versions).

KEY NATIONAL FINDINGS 2015- 2016

- The Needs Assessment Listening Sessions (see Evaluation Brief Year 3 provided to HRSA November 2015 for details) concluded in July 2015. More than 250 participants, including providers, consumers, and public health, contributed their perspective on how genetic services should be defined; affirmation of the dearth of genetics professionals in all settings with related geographic disparities; and solutions that focused on provider education, improved coordination with families; and increased use of distance technologies to increase access to care.
- A national provider survey was conducted in the fall 2015. A total of 924 genetic professionals and related providers described their current practice including wait times and workloads and had input on how to address unmet needs. A manuscript is under development.
- A national consumer survey, entitled Understanding Access to and Quality of Genetic Services: The Individual/Family Perspective, had responses from 1,355 affected individuals/parents. A manuscript about the survey findings was submitted in April 2016 and is under review by the Journal of Maternal and Child Health.
- NGECN staff conducted 18 semi-structured interviews with hospitals, advocacy networks, and community health centers on principles and best practices for patient navigation. This information was incorporated into a Partners in Care monograph that synthesized 14 distinct patient navigation programs into 8 scalable models, showing evidence as an intervention to improve care and impact needs expressed by families.
- Regular RC/NCC leadership calls and NCC workgroup calls (n=71) promote common approaches to HRSA priority areas and rapid dissemination of innovative ideas among the 860 participants.
- There were 9,107 unique visits and 10,875 page visits to ACT Sheets. Almost 400 units of the ACT Sheet App were downloaded by Apple and Google Play users last year.
- NCC/RC Advocate Leaders’ Mentoring Program paired consumers with genetic counselors in a mentoring program at the ACMG annual meeting; 100% of participants (n=13) agreed or strongly agreed (in a post-meeting evaluation) that the learning objectives were met and that the content was useful.

IMPACT OF THE NATIONAL NCC ACTIVITIES

- The Regional Support Service Model Workgroup (RSSM WG) report gives the Health Resources and Services Administration (HRSA) Genetic Services Branch (GSB) a framework for structuring future Federal government investments in genetic services.
- The addition of a heritable condition response category on the National Survey of Child Health (NSCH) will provide for the first time estimates of the child population affected by genetic conditions. It will also give MCH constituents, other survey users, and the MCH Genetics Services Branch critical information by which to analyze variables in the NSCH.
- The RCs are working with Leadership Education in Neurodevelopmental Disabilities Joint (LEND) grantees on projects to integrate genetics into training programs. Other collaborations with HRSA-funded projects are occurring with State Title V programs, the Catalyst Center, Telehealth Resource Centers, and the Association of Public Health Laboratories NewSTEPs program.
- Fourteen states are participating in the pilot study of the Longitudinal Pediatric Data Resource (LPDR). This project will help demonstrate the feasibility of public health teams conducting long-term follow-up data collection.
• The NCC and Heartland RC submitted public comments on the MCH Discretionary Grant Information System (DGIS) measures. “Genetics” was separated from "newborn screening" making it a distinct category in the list of MCH priorities.

• State and Territorial adoption of recommended uniform screening panel (RUSP) critical congenital heart disease (CCHD) and severe combined immunodeficiency (SCID) conditions increased. In 2016, 48 States and territories screened all newborns for CCHD and 40 for SCID.

• NGECN distributed 7,999 family health history booklets and through a national E-card campaign that had 3,798 page views.

• The June 15, 2015 Dialogue Addressing and Paying for Genetic Services in Integrated Delivery Systems meeting brought together geneticists, primary care providers (PCPs), insurers, and consumers to address access to genetic services. The summary of that meeting is available on the NCC website.

• 277,824 individuals (503,573 total pageviews) found accessible and clinically accurate condition information on DiseaseInfoSearch.org; 3,697 individuals accessed links to support groups. 48,925 individuals accessed information on genetics and health, testing, and other genetic services through GenesInLife.org
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.

As shown in the Table 1, the largest numbers of intra-regional RC activities were focused in the priority areas of NBS Capacity Building and Collaboration. Because the intra-regional activities of one region look quite different from those of another, see Supplement 3 for each RC’s activities.

Table 1. Intra-/Inter-Regional Activities and National Activities (2015-2016)

<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>12</td>
</tr>
<tr>
<td>NBS Capacity Building</td>
<td>16</td>
<td>6</td>
<td>22</td>
</tr>
<tr>
<td>Collaboration</td>
<td>15</td>
<td>9</td>
<td>13</td>
</tr>
<tr>
<td>Affordable Care Act</td>
<td>9</td>
<td>5</td>
<td>11</td>
</tr>
<tr>
<td>Long-term Follow-up</td>
<td>3</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>Transition from Pediatric to Adult Care</td>
<td>8</td>
<td>1</td>
<td>0¹</td>
</tr>
<tr>
<td>Telegenetics</td>
<td>9</td>
<td>5</td>
<td>5</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
¹ Note: NCC merged its Medical Home and Transition Workgroups 6/1/16

Source: Supplement 3 RC Intra-/Inter-Regional Activities

The following highlights some of the inter-regional activities of 2015-16:

- As a result of NEGC’s initial efforts to and follow-up collaborations, other regions (NYMAC, Region 4 Midwest, MSGRC, and WSGSC) have implemented their own survey efforts documenting family experiences with health care insurance and service access.
- NYMAC engaged staff of the HRSA Hearing Loss program, Title V, EHDI and EI programs, EHDI –AAP Chapter Champions, LEND faculty, Heartland, Region 4 Midwest, the National Center for Hearing Assessment and Management and other national/regional experts to develop a Hearing Loss Genetics Survey. The Johns Hopkins IRB approved the Hearing Loss Genetics Survey, which is currently being pilot tested by pediatricians and family medicine practitioners, will be disseminated via paper and print to health care providers with data entry closing on August 31, 2016.
- NYMAC delivered a series of six medical home webinars between January and May of 2016. Parents/advocates, physicians, allied health professionals, trainees, public health professionals, and genetics professionals attended. There were 591 unique attendees. On average, 92% of
respondents found that the webinar sessions were “very good” or “excellent”. On average, 81% of respondents found the webinars “relevant” or “very relevant” to their work, and 59% “agree” or “strongly agree” that the information presented would influence the way that they practice. These webinars were approved for and offered CMEs.

- **MI Newborn Screening Data Mapping Project** builds on the Mountain States’ provider mapping by using actual MI NBS data of metabolic, sickle cell disease, and congenital hypothyroidism positive screens to understand geographic distributions of these conditions. For metabolic patients, distance to provider metrics were calculated to understand proximity to care. This project provides an empirical base for understanding where genetics patients live and understanding how to best approach telegenetics and other access to care issues.

- The **NEGC quality improvement** learning workgroups have recruited participation by 5 clinic sites for addressing developmental delay and intellectual disability and 10 clinic sites for PKU or MCAD. Through May 2016, 2,042 individuals have been entered into the registry.

- The **Genetic Services Assessment** (GSA) is a tool developed for state-level public health programs to use in assessing the genetics systems/services in their respective states. Created by an expert panel in the Heartland RC with input and pilot testing from the WSGSC, in 2015-16, Region 4 Midwest and SERC joined NYMAC and MSGRC in administering the GSA. 32 states that have implemented the tool (past versions and current version). The quality metrics cover five domains: (1) State capacity for services; (2) Access; (3) Clinical process and quality improvement; (4) Performance reporting/improvement; and (5) Workforce.

- Five NYMAC states (NY, NJ, MD, DE and VA) are participating in the **NewSTEPS 360** program with funding from the Association of Public Health Laboratories. An assistant research scientist has been hired to conduct a survey of birth hospitals on electronic capabilities including the type of electronic medical record and existing HL7 demographic messages created by their system. 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.

- The Heartland and Western States RCs and HRSA-funded Telehealth Resource Centers in Arizona and Arkansas presented a two-day, in-person telegenetics training in August 2015, which was attended by 13 trainees (2 medical genetics residents, 1 medical geneticist, 9 genetic counselors, and 1 clinic coordinator). Three training sessions for genetics providers and one training session for program administrators are planned for August 2016.

- RCs are collaborating with **Title V leaders**. WSGSC is doing Genetic Assessments with Title V programs and other state genetic service providers through in-person or videoconference meetings to collect genetic service assessment information for each state. An assessment was completed in Hawaii in September 2015 and additional assessment meetings will begin in June 2016. NYMAC held a session with Title V directors from their region at AMCHP’s rescheduled annual meeting.

- Following the November 2015 joint meeting of the NCC/RCs with the Association of University Centers on Disabilities (AUCD) Leadership Education in Neurodevelopmental Disabilities (LEND) Directors, all 7 RCs worked closely with the **43 local LEND** programs to enhance the focus on genetics in LEND programs and to engage RCs in LEND programs. The online NYMAC GPH Fellowship provided educational and leadership opportunities to 14 genetic counselors and LEND trainees. The only LEND-audiology program in the NYMAC region joined in the HL-Genetics needs assessment efforts.
Evaluation Measure: Collaborations

Legend:  Providers: e.g., Primary care provider, genetics specialist, nurse, public health employee  
Family Organizations—e.g. Family Voices, Parent to Parent  
Consumers—individuals or family members  
Disease-specific Organization: e.g. PKU Alliance, Sickle Cell Disease Association of America

The NCC/RCs work with consumers, health and public health professionals and many organizations. In 2015-2016, the RCs’ mailing lists had 3,540 providers; 366 consumers; and had 376 family and disease-specific organizations. Of these stakeholders, 401 providers and consumers participated in the RCs’ annual meeting as did 100 of the organizations. More than 630 people served and 138 organizations worked on RCs’ workgroups and committees. The total number of providers on the RC mailing lists has declined reflecting their efforts to maintain accurate and up-to-date mailing lists. Over four years, there has been growth in the number of consumers and family organizations engaged with the RCs.

All of the RCs hold annual regional meetings that include public health genetics and NBS leaders, family advocates, genetic specialists, primary car representatives, LEND and Title V leaders convene. These meetings provide a forum for sharing information and developing regional priorities.

Two of the HRSA guidance priorities are that the RCs collaborate and partner with MCH-funded programs and that they strengthen public-private partnerships. The following table shows select NCC/RC collaborative activity highlights for 2015-2016.

Table 2. Collaborative Activities and Highlights (2015-2016)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Collaborate and partner with MCH-funded programs</td>
<td>NA</td>
<td>6</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

A meeting was convened with state Title V Director/CYSCHN coordinators at the 2016 AMCHP meeting. Seventeen representatives and/or Title V directors from DE, DC, MD, NJ, NY and VA participated. Discussion focused on areas for collaboration, the NYMAC genetics in hearing loss project, and the outcome of the Genetics System Assessment conducted in Year 3. (NYMAC)
A web-based PKU survey, developed in collaboration with genetic dietitians throughout the mid-Atlantic region, for parents/caregivers of persons with PKU, and/or persons with PKU was fielded in partnership with The National PKU Alliance (NYMAC).

In the fall of 2015, 20 individuals attended the NYMAC Consumer Collaborative Network meeting in Pittsburgh PA. The theme of the meeting was to “share your story.” The group decided to create a “Parent’s Corner” on the NYMAC website. (NYMAC)

In 2015-2016, four states (e.g. Arkansas, Oklahoma, Nebraska, and North Dakota) participated in the Heartland Collaborative Partners Project (CPP), which was established by Heartland to promote state activities that would improve one aspect of their NBS system. (Heartland)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Strengthen public-private partnerships</td>
<td>NA</td>
<td>5</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

A fact sheet on whole genome and whole exome sequencing for patients was developed in collaboration with Genetic Alliance and the NYMAC CCN and was posted on the website in May, 2016. (NYMAC)

Annual collaboration with the New England Regional Genetics Group (NEGC)

GEMSS Resource Website for Schools, Families, and their Children (NEGC)

SERC/SERGG annual meeting 2015 (SERC)

Family Forum Video Project (Region 4)

Regular RC/NCC leadership calls and NCC workgroup calls (n= 71) promotes common approaches to HRSA priority areas and rapid dissemination of innovative ideas among the 860 participants in 2015-16. Staff from NewSTEPs participate on every PD/PM call. During the past year, invited participants included the HRSA Office of Advancement for Telehealth and the National Center for Medical Home Implementation.

The NCC/RCs met with Association of University Centers on Disabilities (AUCD) LEND Directors in November to review joint activities and to discuss additional collaborations. All 7 RCs worked closely with more than 43 local LEND programs to enhance the focus on genetics in LEND programs and to engage RCs in LEND programs. The AUCD website has a specific page for genetics education.

At the 2016 ACMG Annual Clinical Genetics Meeting, over 40 people attended the NCC-sponsored Community Conversation, “Can You Hear Me Now? Empowering Relationships between Consumers and Genetics Providers through Technology”.

National Impact
Evaluation Measure: Public Information

The NCC/RCs use the internet to reach providers, consumers, and state public health agencies with information about genetic conditions and the resources they offer. In Year 4, all RCs had websites that addressed the HRSA priorities of medical home, NBS, and the ACA. These websites had more than 81,460 unique visits and 121,298 home page visits. All RCs are using social media; they reported having 1,898 Facebook followers as of May 31, 2016. Three RCs (NEGRC, SERC and Region 4 Midwest) and the NCC use Twitter with 554 followers and two RCs (Heartland and WSGSC) and the NCC used YouTube with 4,635 video views.

Table 3. 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>52541</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>
</tbody>
</table>

The GEMSS website added three new conditions (Russell Silver Syndrome, CHARGE, and Neuromuscular disorders). The Parent Ambassadors, a NH Leadership Series “Action Group” thoroughly reviewed the site and provided a range of useful recommendations. The resource was disseminated to a range of audiences locally and nationally via traditional mechanisms (e.g., newsletters; presentations at major events), social media (a dedicated Facebook page), and continued development of collaborative relationships with external partners (Global Genes). GEMSS had 25,843 home page visits.

NYMAC contracted with an advertising agency to promote its Facebook page. The total reach (e.g., the number of people who have seen any content associated with the page) was 251,898; their post total reach (the total number of people the page post was served to) was 35,954; and total engagement (the number of people who clicked anywhere in the post) was 1,352.

Region 4 Midwest’s family forum continues to produce high-quality outreach and educational products that can be widely disseminated to families affected by a genetic condition and their providers. Thousands have seen the powerful, professionally produced videos that share first-hand accounts of families. These products (including the Partnering with your Doctor: The Medical Home Approach guide and the Journey through Diagnosis guide still in development) provide an important compliment to families as they learn about their loved one’s genetic condition and how to best provide and support necessary care.
A “Sickle Cell Trait: What You Need to Know” guide was developed by the Region 4 Midwest hemoglobinopathies workgroup. Roughly 1,500 sickle cell trait educational materials in English, Spanish, and French were disseminated across the region.

Newborn Screening Parent Fact Sheets for the disorders included on the Recommended Uniform Screening Panel (RUSP) are developed, updated, and maintained by the WSGSC on our companion website, www.newbornscreening.info. CCHD, SCID and CPT1a Arctic Variant fact sheets were added. The website has 30,000 unique visitors per month.

The NCC website (http://www.nccrcg.org) is the repository of and link to the RC resources. It had 4,318 unique visits and 4,365 home page visits in 2015-16. The NCC Collaborator, the NCC/RC system newsletter, features articles from each RC, the NCC and the NGECN. In 2015-2016, one issue focused on Impacting the Education Gap and Identifying Access to Genetic Service Needs. There was also a Special Edition featuring the NCC/RC Advocate Leaders Partnership Program.

NGECN supports websites that serve as reliable consumer resources. GenesInLife.org puts health and genetics in context for individuals and their families. Some 48,925 individuals accessed information on genetics and health, testing, and other genetic services through GenesInLife.org. DiseaseInfoSearch.org has information on nearly 10,000 conditions and subtypes. There were 663 curated, patient-centered, and low literacy condition descriptions and 125 new/updated support groups added to DiseaseInfoSearch.org this year. Some 277,824 individuals (503,573 total pageviews) found accessible and clinically accurate condition information on DiseaseInfoSearch.org; 3,697 individuals accessed links to support groups.
Evaluation Measure: Workforce Development and 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 2016, the NCC/RCs offered 427 education and training sessions, which reached nearly 7,814 participants. Compared with first-year 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 427 in Year 4; Participants n = 2,110 in Year 1 and n = 7,814 in Year 4).

Table 4. RC-Sponsored Educational and Training Sessions

<table>
<thead>
<tr>
<th></th>
<th>Year 1</th>
<th>Year 2</th>
<th>Year 3</th>
<th>Year 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>In-person</td>
<td>145</td>
<td>144</td>
<td>133</td>
<td>172</td>
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<tr>
<td>Webinars</td>
<td>70</td>
<td>50</td>
<td>79</td>
<td>94</td>
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<tr>
<td>Teleconference</td>
<td>20</td>
<td>26</td>
<td>121</td>
<td>184</td>
</tr>
<tr>
<td>Total</td>
<td>411</td>
<td>249</td>
<td>427</td>
<td>411</td>
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</table>

Table 5. RC-Sponsored Educational and Training Participants (Years 1, 2, 3, and 4)

<table>
<thead>
<tr>
<th></th>
<th>Year 1</th>
<th>Year 2</th>
<th>Year 3</th>
<th>Year 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-500</td>
<td>2,110</td>
<td>7,705</td>
<td>8,680</td>
<td>7,814</td>
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<tr>
<td>6,000</td>
<td>11</td>
<td>50</td>
<td>100</td>
<td>150</td>
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<tr>
<td>7,000</td>
<td>172</td>
<td>133</td>
<td>94</td>
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<tr>
<td>8,000</td>
<td>411</td>
<td>249</td>
<td>427</td>
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</table>
Table 6. Workforce Development and Consumer Education and Training Resources Activities and Highlights (2015-2016)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Expand the pool of the genetic workforce</td>
<td>NA</td>
<td>6</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

Six medical home webinars were offered between January and May of 2016. More than 590 parents/advocates, physicians, allied health professionals, trainees, public health professionals, and genetics professionals attended. There were 591 unique attendees. These webinars were approved for and offered CMEs. (NYMAC)

The EHDI Program Exchange brings state coordinators together to identify their needs and link with states where there are innovative, best practices. (Heartland)

NCC/RC Advocate Leaders’ Mentoring Program paired consumers with genetic counselors in a mentoring program at the ACMG annual meeting; 100% of participants (n=13) agreed or strongly agreed (in a post-meeting evaluation) that the learning objectives were met and that the content was useful.
**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 CCHD and SCID RUSP (Years 1-4)**

<table>
<thead>
<tr>
<th>Year</th>
<th>CCHD</th>
<th>SCID</th>
</tr>
</thead>
<tbody>
<tr>
<td>Year 1</td>
<td>26</td>
<td>13</td>
</tr>
<tr>
<td>Year 2</td>
<td>36</td>
<td>7</td>
</tr>
<tr>
<td>Year 3</td>
<td>42</td>
<td>6</td>
</tr>
<tr>
<td>Year 4</td>
<td>48</td>
<td>2</td>
</tr>
</tbody>
</table>

*CCHD: Kansas is screening almost 100% without a mandate: SCID: Kansas will start pilots summer of 2016, Missouri will have population based pilot in Fall 2016, North Dakota will mandate screening 7/1/16

**Table 8. Building State Public Health Department Capacity Activities and Highlights (2015-2016)**

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Build state public health department capacity</td>
<td>NA</td>
<td>7</td>
</tr>
</tbody>
</table>

*Select NCC/RC Activity Highlights 2015-2016*

A newborn screening timeliness webinar series was planned and speakers have been scheduled. (NYMAC)

A collaboration with GMDI members, parent organizations and AHRQ resulted in PKU guidelines being completed. These evidence-based guidelines were published in a peer-reviewed journal and posted on a web-based portal. MSUD guidelines were accepted and released through AHRQ. (SERC)

A lunch and learn series event was organized in October 2015 and May 2016 to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students on the topic of newborn screening for Pompe disease. 36 participants (in-person and remote) in October and 27 participants in May, reported a 124% increase in knowledge about Pompe disease (October) and 183% increase in knowledge about MPS I disease (May) as evidenced by results from the pre- and post-test survey administered at this event. (SERC)
An on-line course, *Newborn Screening: What Prenatal Care Providers Need to Know*, teaches providers ways to heighten expectant parents’ awareness of newborn screening. Some 102 prenatal care providers, child birth educators, and other health care professionals from across the U.S. have been trained. (Region 4 Midwest)

Technical assistance regarding the addition of Lysosomal Storage Disorders to the RUSP occurred through one in-person RUSP Implementation meeting and 5 NBS Laboratory webinars. (Region 4 Midwest)

Results of the Heartland Collaborative Partners Project (CPP), include: 77% (10) of the midwives have purchased equipment to begin pulse oximetry screening and the other 23% have identified a system for referral (OK); new state procedures will be adopted to disseminate parent brochures based on pilot results (NE); training resulted in improved NBS specimen and collection and decreased rejection rates (ND); and improved knowledge of NBS and NICU NBS policies (AR). (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. Heartland supported additional states in the region to convene and extend the CoIIN project across the region. States (Kansas, Iowa, Arkansas, Missouri, North Dakota, Oklahoma) convened CoIIN teams of newborn screening stakeholders and met in Kansas City May 5 - 6, 2016. All participants ranked the training as valuable or very valuable. (Heartland)

Parent fact sheets for the disorders included on the RUSP are developed, updated, and maintained. The Spanish language fact sheets were recently updated. A Pompe fact sheet is under expert review; XALD is in development. (WSGSC)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Address emergency preparedness</td>
<td>NA</td>
<td>3</td>
</tr>
</tbody>
</table>

**Select NCC/RC Activity Highlights 2015-2016**

Beth Vogel participated as an Advisory Committee Member on an AMCHP-led effort to update the U.S. Department of Health and Human Services Newborn Screening Contingency Plan. The final draft of the Plan was submitted to CDC and HRSA on June 30. (NYMAC)

SERC was invited participated in meeting at APHL and on numerous conference calls to complete the HRSA/APHL 2016 Update to Contingency Plan for Newborn Screening draft in June 2016. (SERC)

February 29, 2016 - APHL Newborn Screening & Genetic Testing Symposium: Regional Newborn Screening Laboratory Backup Planning: 2015 Snapshot (poster, Andersson & Perry) (SERC)
**Evaluation Measure: Long-Term Follow-Up (LTFU)**

The NCC in partnership with the Newborn Screening Translational Research Network (NBSTRN), convened an in-person meeting to develop 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 introducing the tool in 14 states with the help of public health teams.

The NCC/RCs engaged in activities to expand state and regional collaborative systems for conducting and collecting/analyzing data on long-term monitoring, follow-up, and treatment for individuals identified with genetic conditions through NBS. The Inborn Errors of Metabolism Project Information System (IBEM-IS) 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. During 2015-16, several RCs (six centers in the NYMAC region were funded NYMAC, 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, SERC and MSGRC) participated.

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objectives</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
</table>
| Expand state and regional systems for long-term monitoring and follow up | MICH-32.2. Increase the proportion of screen-positive children who receive follow-up testing within the recommended time period  
MICH 32.3. Increase the proportion of children with a diagnosed condition identified through newborn screening who have an annual assessment of services needed and received. | 6 |

**Select NCC/RC Activity Highlights 2015-2016**

The Inborn Errors of Metabolism Information System (IBEMC-IS) projects are supported by many RCs through their direct support of metabolic centers in each region. NYMAC supported 6 centers, SERC is supporting state participation, Region 4 Midwest funded an additional two centers, Heartland funded 2 centers when federal funding lapsed, and MSGRC funds its metabolic consortium’s participation. (NYMAC, SERC, Region 4 Midwest, Heartland, MSGRC)

The RCs are also working closely with the NBSTRN-led Longitudinal Pediatric Data Resource (LPDR) in a project co-sponsored by NCC: SERC has 4 participating states, Region 4 Midwest has 2 participating states, Heartland has 1, and Western States has 1 state participating. The goal is to demonstrate the feasibility of public health teams to conduct LTFU data collection. (SERC, Region 4 Midwest, Heartland, MSGRC, and WSGSC)

The CPT 1A Arctic Variant 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. (WSGSC)

The **Newborn Screening Translational Research Network (NBSTRN)** is a project of ACMG funded by the Eunice Kennedy Shriver NICHD/NIH (contract #HHSN275201300011C). The RCs have supported the development of the NBSTRN Longitudinal Pediatric Data Repository, Virtual Repository of Dried Blood Spots, and the Laboratory Performance Database. NBSTRN, with the collaboration of the RCs, developed core resources to support NBS research; this research continues to be supported by NIH, CDC, and HRSA. The NBSTRN enables investigators and public health teams to systematically collect, analyze and
share data across the research community. This information system uses consensus standardized data sets, case report forms, secure data collection, sharing and management.

**NewSTEPS** (Newborn Screening Technical assistance and Evaluation Program) a HRSA-funded resource for state data on NBS, is a program of the Association of Public Health Laboratories (APHL). With active RC collaboration, NewSTEPS has developed common definitions for NBS disorders, created web resources, and begun data collection on NBS disorders diagnosed within states.
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 4. The RCs use the HRSA Performance Measure (PM) #41 in this annual progress report to HRSA. Over the course of the four-year evaluation, there has been a steady rise in the RCs’ report of medical home activities on the PM #41 from an average score of 18 to 31—a 72% increase.

Table 10. RC Self-Report on Medical Home Activities using HRSA PM #41

(Data points show high, low, average based on a rating scale of N/A= not applicable, 0= no activity; 1= partially met; 2= mostly met; and 3= fully met)

<table>
<thead>
<tr>
<th>Year 1</th>
<th>Year 2</th>
<th>Year 3</th>
<th>Year 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>18</td>
<td>25</td>
<td>28</td>
<td>31</td>
</tr>
</tbody>
</table>

Table 11. Medical Home and Access Activities and Highlights (2015-2016)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objectives</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Treat in the context of a medical home</td>
<td>MICH-30. Increase the proportion of children, including those with special health care needs, who have access to a medical home. MICH 31. Increase the proportion of children with special health care needs who receive their care in family-centered, comprehensive, coordinated systems.</td>
<td>7</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

Four webinars were held aimed at primary care providers and associated health care professionals. Based on satisfaction surveys collected across the 4 webinars held, the 225 participants agreed that the educational session: enhanced their competence (97%, N=74); influenced their practice (90%, N=70); and impacted patient outcomes (90%, N=70). More details on the webinar series (including recordings) are accessible here: http://www.negenetics.org/work-groups/medical/products_pubs_mh. (NEGC)

Two quality improvement workgroups 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. 5 clinic sites participate on the developmental disability/intellectual disability (DD/ID) workgroup (entering data) and 10 clinic sites participate on the PKU / MCAD workgroup. Through May 2016, 2,042 individuals have been entered into the registry. (NEGC)

The NEGC Health Care Access and Financing workgroup successfully launched a new survey among families living with children who have genetic health conditions. This survey had 255 respondents. (NEGC)
After large expansion efforts within Region 4 Midwest and into the Heartland region Care Coordination: Empowering Families Training, Region 4 Midwest analyzed training fidelity among new facilitators with a new fidelity of implementation tool. Feedback from 19 new facilitators rated their experience across four domains on a scale from 1 to 9 (1-3 “Needs Work”, 4-6 “Acceptable”, 7-9 “Good Work”) and then provided qualitative evidence to support their rating. Ratings range from a mean of 7.45 to 8.15, indicating relatively high degrees of fidelity overall. (Region 4 Midwest)

The Hispanic Access Advisory Council (HAAC) has as its goal 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. In the first year of the project 20 conversations with key stakeholders were completed in an effort to identify the issues surrounding access and provision of culturally competent medical genetics services. Staff at three FQHCs were part of the interview process and helped to identify families for the interviews. HAAC met to review the findings and determine implementation priorities to improve improved access to genetic service. (Heartland and MSGRC)

MSGRC sponsored the attendance of 6 consumers and 1 family physician to the Texas Primary Care and Home Health Summit. This was the first year the summit had consumers in attendance (MSGRC)

In the Parent Partner Project, 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. The Parent Partner Project is currently in eight pediatric practices in Montana and is expanding to a reservation site and air force base both in Wyoming. (MSGRC)

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Improve insurance coverage policy and reimbursement</td>
<td>NA</td>
<td>6</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

A Research Brief: Promoting and Improving the Health and Well-Being of People with Inherited Conditions was released in May of 2016 highlighting some of the major findings of the work of the NEGC Health Care Access and Financing Group. Families, advocacy organizations, and the Catalyst Center are involved. (NEGC)

The results of the consumer benefits survey were published in Exceptional Parent Magazine in November 2015. (NYMAC)

Access to medical foods was identified as a potential area of need during the Year 3 consumer benefits survey. During year 4, a group of metabolic dietitians engaged in discussions about barriers and solutions. The group developed a survey for patients with PKU regarding medical foods. 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 and insurance coverage. (NYMAC)

The ACA—Through the Life Course Website explains the ACA to families in relation to their life stages. The website was recently updated with input from families, public health and health providers in AK, CA, HI, ID, OR, WA, and Guam. (WSGSC)

An ACA Parent Survey was modeled after similar surveys developed by NEGC and NYMAC and administered in Hawaii to families of children with special health needs regarding essential health benefits and financing of health care. (WSGSC)
The June 15, 2015 Dialogue Addressing and Paying for Genetic Services in Integrated Delivery Systems meeting brought together geneticists, PCPs, insurers, and consumers to address access to genetic services. The meeting proceedings were posted on the NCC website for use by decision makers and advocates to address emerging issues in the payment of genetic services. Work continues in three subgroups focused on Identifying Genetics Services; Coverage of Genetic Services; and the Consumer Role in Genetic Services.

The white paper entitled “Sharing Exemplary Practices from Primary and Specialty Care Providers” was finalized by the NCC Care Coordination Workgroup and posted on the NCC website.

NGECN distributed 7,999 family health history booklets.
**Evaluation Measure: Transition from Pediatric to Adult Care**

In 2015-6, six RCs engaged in activities related to transition from pediatric to adult care as shown below:

<table>
<thead>
<tr>
<th>HRSA RC Guidance 12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other – Transition</td>
<td>DH-5 - Increase the proportion of youth with special health care needs whose health care provider has discussed transition planning from pediatric to adult health care.</td>
<td>6</td>
</tr>
</tbody>
</table>

**Select NCC/RC Activity Highlights 2015-2016**

The “Health Survey in Adults Living with Genetic Conditions” was administered to young adults with genetic conditions. To date, 85 condition-oriented and 52 control surveys have been collected. The survey continues in multiple sites to document potential disparities in health and well-being among those with and without targeted conditions. (NEGC)

The NEGC continues to support the Face2Face Camp’s efforts to educate and train youth in becoming more effective self and group advocates. Eighteen youth ages 16 -24 participated in the three-day 2015 camp. Across 7 sessions rated by participants, satisfaction scores averaged 4.3 on a five-point "Awesomeness" scale (5=Awesome). When participating youth were asked whether they would be willing to advocate for themselves or others living with their condition, pre-post-tests increased from 78% agreement to 92%, and there was a 26% increase in respondent perceptions on how they could achieve their personal goals. (NEGC)

Dr. Waisbren successfully developed and implemented a transition policy at Children’s Hospital of Boston. The transition policy has also been adopted by a clinic in Vermont and is made available for use for other interested clinics in the New England region at [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/). (NEGC)

SERC is conducting a Delphi study on transition care success utilizing SERC consumers to gain a more in-depth understanding on what transition success means to them. The long-term outcome is for health professionals to utilize the results of this study to create better transition care programs for individuals and families affected by genetic disorders. (SERC)

The Healthcare Transition for individuals with genetic conditions project was to determine the role of the genetics team in healthcare transition. To date, focus groups, interviews, a survey and a learning collaborative strategy have been used to gather baseline information. From these data, it was determined that the needs were for tools/resources, interventions/protocols, and information/education. In the past project year, all of the work has been conducted in South Dakota. Tools: Transition Engagement Guide and Doc Talk Interventions: Transitions Clinic Education: Adolescent Health Rotation for Pediatric Residents (Heartland)
Cluster Measure: Telegenetics/Distance Strategy
To improve access to genetic services, all seven RCs began pursuing telegenetics and distance strategy projects during this grant year. Activities and highlights of 2015 -2016 follow:

Table 13. Telegenetics/Distance Strategy Activities and Highlights (2015-2016)

<table>
<thead>
<tr>
<th>HRSA RC Guidance12-138 Priority</th>
<th>Healthy People 2020 Objective</th>
<th>RCs with Activities as of May 2016</th>
</tr>
</thead>
<tbody>
<tr>
<td>Telegenetics</td>
<td>NA</td>
<td>7</td>
</tr>
</tbody>
</table>

Select NCC/RC Activity Highlights 2015-2016

NEG is actively supporting the launch of the Genetic Metabolic Center for Education (GMCE). GMCE partners developed a HIPAA compliant telemedicine system for hosting conferences between specialists and other care providers. Launched as a pilot effort with three clinical sites from across the New England region, GMCE completed 19 consultations with providers. (NEGC)

NYMAC collaborated with the Ferre Institute as part of the NYMAC Needs Assessment to better understand access to genetic services of pediatricians, oncologists and OB/GYNs, and pilot a help line and telegenetics for healthcare providers with genetic questions. (NYMAC)

NYMAC funded a project at Children’s Hospital of Pittsburgh (CHP) Pediatric Hematology. During the last funding year, CHP successfully conducted telemedicine visits with patients with sickle cell disease to increase the frequency of visits (from twice yearly to quarterly visits) and to improve compliance with hydroxyurea therapy (monthly visits). (NYMAC)

The Michigan Newborn Screening (MI NBS) Mapping Project uses MI NBS data of metabolic, sickle cell disease, and congenital hypothyroidism positive screens to understand geographic distributions of these conditions. For metabolic patients, distance to provider metrics were calculated to understand how far patients travel for care. The MI Department of Health and Human Services Newborn Screening Program and geographers from Michigan State University produced a series of maps illustrating the degree of clustering and spatial correlation with the total population. (Region 4 Midwest)

The Heartland and Western States RCs and HRSA-funded Telehealth Resource Centers in AZ and Arkansas presented a two-day, in-person telegenetics training in August 2015, which was attended by 13 trainees (2 medical genetics residents, 1 medical geneticist, 9 genetic counselors, and 1 clinic coordinator). Three training sessions for genetics providers and one training session for program administrators are planned for August 2016. (Heartland and WSGSC)

In collaboration with Cook Children’s MSGRC is in the initial stages of a case study to understand the implementation of telemedicine in a genetics clinic. The purpose of the study is to demonstrate outcomes and effectiveness for clinicians, patients, and payers as a basis for “convincing” geneticists to utilize telemedicine in their own clinics. (MSGRC)

Completed literature review and wrote framework for approach necessary to measure outcomes of genetic services, including information specific to telegenetics. (WSGSC)

As the NCC/RC system enters into its final year of funding, it is anticipated that the HRSA MCHB Genetics Services Branch will be redesigning this system to address the ongoing misdistribution of genetics resources, to promote the translation of genetic medicine into public health, and to improve access to genetic services. NCC has led the effort through its RSSM Workgroup to review other regionalized models of health care and to offer recommendations for how a regionalized genetic infrastructure in the United States might look like in the future to improve access to care for individuals with genetic conditions.
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### Supplement 1: Summary of RC Priorities - May 31, 2016

<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>
</tr>
</thead>
<tbody>
<tr>
<td>Treat in the context of a medical home</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Cultural competency and diversity in outreach projects</td>
<td></td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Expand the pool of the genetic service workforce</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
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<tr>
<td>Build state public health department capacity</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
</tr>
<tr>
<td>Strengthen public-private partnerships</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Collaborate and partner with HRSA MCHB-funded programs that promote the scaling up of effective practices</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
</tr>
<tr>
<td>Improve insurance coverage policy and reimbursement</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
</tr>
<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>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
</tr>
<tr>
<td>Address emergency preparedness</td>
<td></td>
<td>X</td>
<td></td>
<td>X</td>
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<td></td>
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<tr>
<td>Others not in RFA</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
<td></td>
</tr>
</tbody>
</table>
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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 2015/2016, staff were involved in two peer reviewed publications and 14 invited oral presentations.
- **GEMSS**: Continued to support scope and depth of the GEMSS website by adding 3 new conditions (Russell Silver Syndrome, CHARGE, and Neuromuscular disorders), and disseminating the resource to a range of audiences locally and nationally via traditional mechanisms, social media, and continued development of collaborative relationships with external partners (Global Genes).
- **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 (developing plans for implementation).
- **Medical Home**: Primary care providers, and associated health care professionals who attended the NEGC Medical Home Work Group sponsored webinars educating primary care practices and other care providers around concepts of creating a shared plan of care, documenting family history, incorporating genetics in the management of children in the primary care office, and holding family centered conversations.
- **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 developing a learning module supporting the use of Genetics Education Materials for School Success (GEMSS) website content in higher education, and the third is analyzing data from the second family survey about health insurance access, expenses, coverage and reimbursement. NEGC continues to collaborate with the AUCD-LEND working group on genetics education for LEND programs. This year the group is proposing a presentation on the genetics of congenital deafness for the annual AUCD LEND meeting in December 2016.
- **The Health Care Access and Financing work group** successfully launched a new research survey among families living with children who have genetic health conditions. The survey touches on a range of critical topics, including experiences working with insurance providers, cost and affordability of care, and barriers to care.
- **Transition**:  
  - The “Health Survey in Adults” was administered to young adults with genetic conditions at multiple sites, with 85 condition-oriented and 52 control health surveys collected to date.
  - The NEGC continues to support the Face2Face Camp's efforts to educate and train youth in becoming more effective self and group advocates. Eighteen youth participated in the 2015 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 is supporting a young adult’s efforts to coordinate a homocystinuria/metabolic support group. Eight participants joined in the last meeting.
  - Dr. Waisbren successfully developed and implemented a transition policy at Children’s Hospital of Boston. The transition policy has also been adopted by a clinic in Vermont and is made available for use for other interested clinics in the New England region at [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/).
- **CCHD**: An electronic resource with psychosocial and other supports for families affected by CCHD was developed ([www.necongenitalheartresources.org](http://www.necongenitalheartresources.org)) and went live in May 2016. 500 resource bags were distributed to pediatric cardiology practices throughout New England. In addition, the project evaluation report was completed, two manuscripts were submitted to peer-reviewed journals and two additional manuscripts are in preparation.
- **Genetic Metabolic Center for Education (GMCE)**: The NEGC entered into a pilot stage with its new 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.
  - Broadened effective distribution of GEMSS via use of Parent Ambassadors, a NH Leadership Series “Action Group” which thoroughly reviewed the site and provided a range of useful recommendations, dedicated
Facebook pages; publications about GEMSS in various newsletters; presentations at major events, and
collaboration with partners involved in similar missions.

- Dissemination efforts resulted in another year’s increase in reach of the GEMSS website (15,000 in 2014,
20,000 reached in 2015)
- 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 May 2016, 2,042
individuals have been entered into the registry.
- Medical Home: Of the 4 medical home webinar sessions held, over 90% of attendees indicated that the sessions were
good or excellent at providing a valuable and useful resource.
- NEGC / LEND: Three Leadership Education in Neurodevelopmental Disorders (LEND) trainees successfully completed
three distinct projects, presented their work in a variety of settings, and received positive feedback from partners.
- HAF: The second survey administered by the HAF group reached 255 respondents. In collaboration with a LEND
trainee, the results were analyzed and presented in both a poster board presentation and a policy brief. Core topics
raised include: understanding health insurance coverage, commonly denied benefits and services, services not
covered by insurance, access to services, and impacts on families.
- Transition:
  - The Adult Health Survey has been administered to 85 condition oriented and 52 control adults. Preliminary
results were presented by Dr. Farrah Rajabi at the 2016 NEGC annual meeting.
  - The Face2Face annual camp: Across 7 sessions rated by participants, satisfaction scores averaged 4.3 on a five
point "Awesomeness" scale (5=Awesome).
- GMCE: The NEGC announced 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, three sites are participating in the pilot phase of the metabolic consultations.

Was anyone better off?
- GEMSS: During the past year, over 20,000 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 62,000 users from across the world - 42,000 from the US and
20,000 from 179 countries.
  - 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 nearly
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.
- MH: Based on satisfaction surveys collected across the 4 webinars held, participants agreed that the educational
session: enhanced their competence (97%, N=74); influenced their practice (90%, N=70); and impacted patient
outcomes (90%, N=70). When asked what they will do differently, responses included: improving the implementation
of care coordination planning in their offices, more consistent / expanded use of family histories in their practices,
using knowledge gained concerning genetic testing in their practices, seek to improve communications with families.
- NEGC/LEND: Trainees involved in the NEGC/LEND projects produced a range of useful materials, including a summary
report on the HAF project, an analysis of stakeholder perspectives on the NEGC, and substantive new information for
the GEMSS project that will lead to multiple improvements in this critical resource.
- Transition:
  - Face2Face Annual Camp: When participating youth were asked whether they would be willing to advocate for
themselves or others living with their condition, pre-post tests increased from 78% agreement to 92%, and
there was a 26% increase in respondent perceptions on how they could achieve their personal goals.
- GMCE: Has provided 19 consultations in the New England area.
New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC)
Highlights 2015-16

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

1. A Pompe Disease/MPS1 Symposium provided 44 regional participants from NYMAC with increased knowledge about the diagnosis and treatment of these disorders. A discussion of barriers to implementation of screening for MPS1 and Pompe disease added to the overall NYMAC needs assessment and identified future workgroup priorities.
2. A newborn screening timeliness webinar series was planned and speakers have been scheduled.
3. A newborn screening social media campaign to pediatricians and parents had a total reach of 129,251.
4. An assistant research scientist was hired to conduct a survey of birth hospitals on electronic capabilities including the type of electronic medical record and existing HL7 demographic messages created by their system. The survey is under development.
5. The video from the Year 3 ALD meeting was distributed to other regions.

Addressing Primary Care Linkages and Strategies

1. NYMAC conducted a medical home webinar series. Between all six webinars in the series, a total of 591 unique participants attended. Responses to evaluation forms were analyzed and presented to the NYMAC AC Meeting.
2. NYMAC collaborated with the Ferre Institute as part of the NYMAC Needs Assessment to better understand access to genetic services of pediatricians, oncologists and ob/gyns, and pilot a help line and telegenetics for healthcare providers with genetic questions.
3. NYMAC funded a project at Children’s Hospital of Pittsburgh (CHP) Pediatric Hematology. During the last funding year, CHP successfully conducted telemedicine visits with patients with sickle cell disease to increase the frequency of visits (from twice yearly to quarterly visits) and to improve compliance with hydroxyurea therapy (monthly visits). Nine pediatric patients at the Erie location and two pediatric patients at the Johnstown location completed 94 and 7 telemedicine visits respectively before Dec, 2015. The proportion of patients receiving hydroxyurea was 73%. Patients/families reported improved satisfaction owing to crucial time and saving of resources saving. Two half-day workshops were conducted at the CHP on March 15th and April 12th with pediatric hematologists from DE, NJ, NY, and VA.
4. Addressing the Capacity of the Public Health Programs
1. NYMAC continued to develop meaningful relationships with the Title V/CYSHCN directors during Year 4. A meeting was convened with state Title V Director/CYSHCN coordinators at the 2016 AMCHP meeting. Seventeen representatives and/or Title V directors from DE, DC, MD, NJ, NY and VA participated. Discussion focused on areas for collaboration, the NYMAC genetics in hearing loss project, and the outcome of the Genetics System Assessment conducted in Year 3.
2. Literature review, stakeholder discussions, and data from the NCC/ACMG genetics provider survey informed the designing of a Hearing Loss (HL)-genetics needs assessment survey in collaboration with the National Center for Hearing Assessment and Management (NCHAM). The survey has been approved by the Johns Hopkins IRB and is currently being distributed to pediatricians, family medicine practitioners, nurse practitioners, and physician assistants. Stakeholders included HRSA HL program, Title V, EHDI and EI programs, EHDI –AAP Chapter Champions, LEND faculty, Heartland, Region 4, and other national/regional experts such as Arti Pandya and Kathleen Arnos. NYMAC exhibited a poster session at the EHDI 2016 Annual Conference (March 2016),
presented a poster at the ACMG 2016 Annual Meeting (March 2016), presented a webinar for the AAP-EHDI PA Chapter (May 2016), and participated in the Heartland EHDI meeting.

3. NYMAC continued to participate in periodic LEND –Genetics Workgroup conference calls. A HL –Genetics panel discussion session proposal was planned for the AUCD 2016 conference in December.

4. NYMAC Genetics/Genomics Public Health (GPH) online Fellowship included 18 LEND/Genetic Counseling fellows (1 outside the NYMAC region) in Year 4. Three webinars and three LinkedIn discussions on topics related to CADASIL, special health care needs, public health genomics, communication and advocacy were completed. Two fellows presented GPH experiences in their classes, and one fellow wrote a blog about the GPH fellowship. The final GPH evaluation is in progress. Overall, 10/13 fellows (as of 06/29) agreed or strongly agreed that expectations regarding educational opportunities were met and 13/13 agreed or strongly agreed that expectations regarding time/work commitment were met. NYMAC made a presentation on the GPH Fellowship at the Transnational Alliance for Genetic Counselors (TAGC) Meeting through Skype.

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

1. A webinar was conducted to share the results of the Year 3 consumer benefits survey with a total of 35 attendees. The results of the consumer benefits survey were also published in Exceptional Parent Magazine in November 2015.

2. Access to medical foods was identified as a potential area of need during the Year 3 consumer benefits survey. During year 4, a group of metabolic dieticians were engaged in discussions about barriers and solutions. The group developed a survey for patients with PKU regarding medical foods. The survey was approved by the IRB and is currently being distributed through the NYMAC Facebook Page, PKU parent organizations and participating dietitians. The survey results will be used for advocacy, but also to identify the techniques used by patients who are successful in obtaining coverage.

Strengthening NYMAC’s Capacity

1. We contracted with an advertising agency to promote our Facebook page. Our total reach (the number of people who have seen any content associated with the page) was 251,898; our post total reach (the total number of people our Page post was served to) was 35,954; and our total engagement (the number of people who clicked anywhere in the post) was 1,352.

2. We established a Regional Genetics Education Network (RGEN). We advertised the network to promote participation using our Facebook page, NYMAC News emails, and a mailing to genetic counselors in the region. There are currently 56 individuals enrolled in the RGEN.

3. We started an educational campaign for fragile X syndrome. We identified more than 1,000 groups for education and outreach including autism support groups, local chapters of the American Academy of Pediatrics and developmental therapy training programs. These groups will receive an email to offer educational events provided by RGEN members.

4. We developed an educational postcard on fragile X syndrome. The postcard was mailed to 10,032 Pediatricians in the NYMAC Region in the beginning of Year 5.

5. Twenty individuals attended the NYMAC Consumer Collaborative Network Meeting.

6. A fact sheet on whole exome sequencing for patients was developed in collaboration with Genetic Alliance and the NYMAC CCN and was posted on the website in May, 2016.

7. We initiated the consumer column in the NCC Collaborator and remain ongoing contributors.

8. We are planning the October, 2016 Summit on “Bridging the Gaps in Genetics and Newborn Screening Services.”
Southeast Regional NBS & Genetics Collaborative (SERC) Highlights 2015-16

SERC team members adapted the Heartland Collaborative’s Genetics Needs Assessment Survey for the SERC states. This survey was approved through the Emory IRB and was administered via SurveyMonkey. A qualitative component to the survey was also developed to collect supplemental information through telephone interviews with state genetics coordinators or their representatives. 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. Seven states in our region have responded to the survey and the results will be shared at the upcoming SERC regional meeting.

Currently, SERC is working on conducting a Delphi study on transition care success utilizing our SERC consumers as the experts in order to gain a more in-depth understanding on what transition success means to them. We are working on identifying consumer leaders in every state that can identify other consumers within the state, especially Sickle Cell consumers. We will work on facilitating focus groups via telephone conference calls to gather more questions for the question pool. The SERC transitioning workgroup’s short-term outcome is to work alongside the SERC consumer alliance group to conduct the Delphi study and publish the results in the upcoming year. The long-term outcome is for health professionals to utilize the results of this study to create better transition care programs for individuals and families affected by genetic disorders. It is our hope that the dissemination of results from the Consumer Transition Delphi study will improve transition care and quality for consumers and families affected by genetic disorders.

The National Coordinating Center for the Regional Genetic Service Collaboratives (NCC), in partnership with the Newborn Screening Translational Research Network (NBSTRN), worked together to develop a set of questions and answers and a data collection 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 introducing the tool in various states through public health teams. 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. This will potentially be a national model.

The SERC Consumer Alliance released its first quarterly newsletter. It includes events geared toward families with genetic and metabolic disorders from each state in the southeast region. Medical foods legislations have been passed in Georgia and a new non-profit organization related to newborn screening has been developed in Alabama. The SERC consumer alliance group continues to hold conference call meetings where consumers give state updates, plan awareness events, voice opinions, and provide assistance to one another. The SERC Consumer conference calls bring together 36 consumers while the SERC Transitioning regional call is comprised of 9 Core staff members; 8 from Georgia; 12 from North Carolina; 7 from South Carolina; 7 from Florida; 14 from Tennessee; 3 from Mississippi; 8 from Alabama; 12 from Puerto Rico; 1 from Louisiana; 1 from Virgin Islands, and 14 outside of our region. Collaborations among consumers have led to successful sharing of advocacy activities.

Our team is planning activities to expand workforce capacity in our region. One of the upcoming activities includes a web-based learning system which will include user participation in case studies, video conferences and online group discussions. The Emory Nutrition Academy (eGNA) is being developed as a three-phase online course in Genetic Metabolic Nutrition Education. The team will pilot test Phase I of the educational project, which aims to
enhance clinical expertise and applied research skills of clinicians in the area of inherited metabolic disorders (IMDs), specifically disorders requiring lifelong nutritional management as the mainstay of therapy.

Two SERC lunch and learn series events were organized to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students. The first event held in October 2015 focused on the topic of “Pompe Disease: Newborn Screening Initiative in the US.” A total of 25 individuals participated in this event in–person, while 11 individuals participated via webex throughout the region. The Pre and Post-test administered at this event showed a 124% increase in knowledge about Pompe disease among the participants. The second event held in May 2016 covered the topic of “MPS I disease: phenotypes and considerations for newborn screening.” A total of 14 individuals participated in this event in–person, while 13 individuals participated via webex throughout the region. The Pre and Post-test administered at this event showed a 183% increase in knowledge about MPS I disease among the participants.

SERC and Genetic Metabolic Dietitians International (GMDI) have developed toolkits designed to provide a practical and useful guide to assist clinicians implementing appropriate nutrition management of individuals with maple syrup urine disease (MSUD) and Phenylketonuria (PKU). These practical companions for clinicians are based on the nutrition management guidelines for these disorders, and are now available to practitioners on the SERC and GMDI websites.

The SERC SERGG Annual Meeting 2015 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 2015 annual meeting was held from July 16-18 at the Renaissance Hotel in Asheville, North Carolina. The meeting was attended by 176 healthcare professionals with an interest in genetics and public health. Fourteen consumers also attended. Thirty one platform and twenty three 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 Pompe’s disease in the US, a telemedicine demonstration project, a report on evidence and rational for newborn screening for MPSI along with transitioning to adult healthcare, and building family communities to enhance knowledge and improvement of patient care for rare genetic disorders. Other topics covered were genomics and newborn screening, metabolomics, national nutrition management guidelines, diagnosis of creatinine deficiency syndrome and newborn screening for lysosomal disorders. The Consumer Alliance group included a strategy session on emergency preparedness communication, medical home and transitioning, medical food equity act, and a scavenger hunt to educate consumers on available low protein food products in regular grocery stores. In addition, a special session for families with urea cycle disorder was organized. Plans are currently underway to hold the next annual meeting from July 14 – 16th, 2016 in Ponte Vedra Beach, Florida.
How much did you do?

Major Region 4 Midwest projects in year 4.

Sickle Cell Trait Follow-up Initiatives. Disseminated roughly 1,500 sickle cell trait educational materials in English, Spanish, and French across the region. “Sickle Cell Trait: What You Need to Know” guide was developed by the Region 4 hemoglobinopathies workgroup. Facilitated in-person meeting of the hemoglobinopathies workgroup to finalize 18 sickle cell trait short term follow up recommendations and began interviews with families and professionals to seek feedback on those recommendations.

MI Newborn Screening Data Mapping Project. Project builds on last year’s replication of Mountain States’ provider mapping by using actual MI NBS data of metabolic, sickle cell disease, and congenital hypothyroidism positive screens to understand geographic distributions of these conditions. For metabolic patients, distance to provider metrics were calculated to understand proximity to care. This project provides an empirical base for understanding where genetics patients live and understanding how to best approach telegenetics and other access to care issues.

Care Coordination: Empowering Families Training. Continued to partner with HRSA-funded projects and family advocacy groups 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 who play a major role in coordinating care for their child. In Year 4, Region 4 Midwest trained 99 caregivers in five trainings in collaboration with the MI Department of Health and Human Services and MI Family 2 Family. To date, over 400 caregivers across the country have been trained.

Metabolic Foods and Formulas Insurance Coverage. Coordinated information gathering and sharing efforts around insurance coverage for medical foods and formulas throughout the region. Partnership between the Michigan Department of Health and Human Services and Region 4 Midwest’s Public Health Long Term Follow-up Workgroup established to work towards understanding coverage for metabolic foods and formulas and helping consumers navigate new payment systems throughout the region.

Family Forum Videos and Journey through Diagnosis Guide. Completed and distributed the video Care Coordination: The Family Perspective. This video highlights Region 4 family partners discussing the definition and importance of care condition. This is the second video in a series aimed at the family experience. Together, the videos have been viewed by 2,264 people. The Family Forum began the development of the Journey through Diagnosis guide for families that have recently been diagnosed with a genetic condition.

Lysosomal Storage Disorders Screening Implementation Support. Facilitated technical assistance among the Region 4 states regarding the addition of Lysosomal Storage Disorders to the Recommended Uniform Screening Panel through one in-person RUSP Implementation meeting and 5 NBS Laboratory webinars. Attendees from NBS programs and NBS laboratories participated in discussions and heard presentations from Minnesota and Illinois.

Genetic Systems Assessment Facilitation. Region 4 staff coordinated with Dr. Ann Chou and Heartland Collaborative colleagues to develop a protocol to introduce the tool at the 2015 Regional Meeting. Stakeholders from each state engaged in a dedicated discussion of the state of genetic services in their state.

Regional Meeting and Workgroup Facilitation. Hosted 69 diverse stakeholders from October 19-21, 2015 for annual meeting. The meeting included five in-person Workgroup/Forum Meetings, four multi-presenter plenary panel presentations, ten break out promising practice/new idea sessions, and stakeholders meeting by state to complete the Genetic Systems Assessment tool. Region 4 Midwest staff continue to host a variety of webinars and in-person facilitation to achieve the goals of the collaborative.

Website and Sharepoint Updates for Information Dissemination and Collaboration. Continued to maintain and expand the Region 4 Midwest

![Map of the United States showing Region 4 Midwest](image)
website as an important resource for stakeholders across the region and beyond. In Year 4, there were 8,932 visits to the Region 4 Midwest homepage, with 6,553 unique visitors. Region 4 Midwest staff also launched a new sharepoint site for collaborative members to more easily access and collaborate on important workgroup efforts. **How well did you do it?**

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

**MI Newborn Screening Data Mapping Project.** We successfully established a Data Use Agreement with the MI Dept. of Health and Human Services Newborn Screening Program to share data from the three most prevalent conditions. Professional geographers performed spatial analyses to understand correlations to the state’s population distribution. Analyses of distance to provider allows for empirical estimates of reasonable access to care. Urban access issues were also explored (e.g., driving time vs. public transit time).

**Care Coordination: Empowering Families Training Fidelity of Implementation.** After large expansion efforts within Region 4 Midwest and into the Heartland region, we were able to analyze training fidelity among new facilitators with new fidelity of implementation tool. To date, we have feedback from 19 new facilitators who rate their experience across four domains on a scale from 1 to 9 (1-3 “Needs Work”, 4-6 “Acceptable”, 7-9 “Good Work”) and then provide qualitative evidence to support their rating. Ratings range from a mean of 7.45 to 8.15, indicating relatively high degrees of fidelity overall. The tool also provides concrete feedback about successes and challenges for new facilitators that Region 4 staff can incorporate when planning future assistance.

**Regional Meeting and Collaboration.** Our 2015 Regional Meeting was attended by 69 stakeholders and received positive evaluations, with 100% of respondents agreeing that the objectives were met, the content was useful, they were satisfied with the meeting, and that they felt confident they could apply what they learned (based on 54% response rate). Stakeholders who completed Regional Meeting evaluations described how they will apply the knowledge they gained after the meeting. Qualitative data was organized and coded into four main themes: Collaboration, Long Term Follow-up, Telemedicine/Access to Care, and Patient-centered Care.

**Was anyone better off?**

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

**Sickle Cell Trait Follow-up Initiatives.** Early follow-up allows children to receive needed education and provides an opportunity for families to receive genetic counseling. Follow-up for SCT in state newborn screening programs across the region varied greatly, resulting in children and families with SCT receiving different levels of education in each state. To add to the variability of follow-up among states, there are currently no published guidelines for newborn screening programs on sickle cell trait follow-up. The hemoglobinopathies workgroup has developed recommendations and is in the process of gathering consumer and expert feedback. In the meantime, approximately 1,500 educational materials in three languages have been disseminated to increase awareness of SCT.

**Family Forum Videos and Journey through Diagnosis Guide.** Region 4 Midwest’s family forum continues to produce high-quality outreach and educational products that can be widely disseminated to families affected by a genetic condition and their providers. Thousands have seen the powerful, professionally produced videos that share first-hand accounts of families. These products (including the Partnering with your Doctor: The Medical Home Approach guide and the Journey through Diagnosis guide still in development) provide an important compliment to families as they learn about their loved one’s genetic condition and how to best provide and support necessary care.
Collaborative Partners

What did we do?
In 2015-2016, four states (e.g. Arkansas, Oklahoma, Nebraska, and North Dakota) participated in the Heartland Collaborative Partners Project (CPP), which was established by Heartland to promote state activities that would improve one aspect of their NBS system. Each state selected a community participant (e.g. pediatrician) who was interested in learning more about newborn screening (NBS) and perceived him/herself as change agents who could work with their state NBS coordinator to make a difference. Each state chose a NBS project that became the focus of their work, as well as, attended the American Public Health Laboratories (APHL) Newborn Screening Genetic Testing Symposium (NBSGTS) to meet with the NBS representatives from the Heartland states, increase their knowledge of newborn screening, and provide an opportunity for networking and exchanging of information and resources. Projects included: improving their system of distributing parent information (NE); supporting midwives to adopt pulse oximetry screening for home deliveries (OK); providing NICU nurses at their children’s hospital information on NBS, the appropriate follow-up steps after a NBS is completed, and discussing new NICU policies related to NBS (AR); and improving proper NBS specimen collection and rejection rates (ND).

How well did you do it?
State participants found the APHL conference to be a very positive experience that increased their awareness of the NBS process. As one participant noted, “I came out of my comfort zone in the world of pediatrics and learned about the impact of NBS.” Others sited the parent panels as very insightful. With respect to the state activities, those that implemented a training component received positive ratings from the participants. The result of the satisfaction survey found that the participants rated (strongly agreed) the presentation very highly with high percentages (93%) reporting that they gained new information.

Was anyone better off?
As a result of their work, each project will result in improved changes in NBS systems of care:

- 77% (10) of the midwives have purchased equipment to begin pulse oximetry screening and the other 23% have identified a system for referral (OK);
- new state procedures will be adopted to disseminate parent brochures based on pilot results (NE);
- training resulted in improved NBS specimen and collection and decreased rejection rates (ND); and
- improved knowledge of NBS and NICU NBS policies (AR).

EHDI Exchange

How much did you do?
The EHDI Program Exchange was previously implemented by the EHDI coordinator visiting another state program to learn from their EHDI processes. A modification to the approach was made that included an interactive workshop. The agenda for these workshops were informed by the participants based on their prioritized needs. For the past two years, an EHDI Information Exchange workshop was conducted. The workshops were structured so there was time for a networking exchange among the participants. A national consultant, Dr. Karl White, the Director of the National Center for Hearing Assessment and Management, participated in both workshops. In 2016, the second workshop had a series of state presentations. Six EHDI coordinators identified targeted objectives for discussion at the networking section as the workshop was based on a self-assessment process. A sample of the content from these identified objectives included: how to better interface follow-up with Early Head Start or Early Intervention programs; how best to address “state border” infants; strategies to increase communication with primary care physicians; strategies to improve follow-up with home-birth populations; exploration of parent liaisons to support
other parents through the process; using texting as a communication strategy for follow-up, resources for parents, and exploration whether other states are beginning to test for CMV.

How well did you do it?

Overall, the participants indicated that having an interactive workshop with content targeted specifically for their group was very valuable and resulted in higher engagement of the participants. Aspects of the format that contributed to its success included: ample time for networking with individuals who were addressing similar challenges; an agenda that was informed by the participants; and topics that applied to their practices. They indicated that this format benefited them and facilitated their work better than the traditional conference format (e.g., the EHDI national conference). It provided an important avenue for networking that continued outside of the workshop venue. Several commented that the networking opportunity with a national caliber presenter was invaluable and they would be using many of the resources that he shared. They all recommended that this technical assistance strategy be continued.

Was anyone better off?

Based on interview and survey data, participants reported that they learned from other state EHDI coordinators, both what worked and did not work. It allowed them to “share ideas and share resources.” The EHDI coordinators reported many concrete examples of changes in practices or additions of resources that were adopted or are in the process of being developed by their states as a result of the information exchanged at these workshops. Examples include: system changes (defined a state process for follow-up with physicians, development of a text process for notifying parents); resource development (modified parent resource materials developed by another state); and support services (e.g., family to family match program).
Who We Serve

With over one million square miles extending from Canada to Mexico, the Mountain States Regions constitutes a land area of almost one-third of the entire United States and a population of more than 48 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. In the three states along the US-Mexico border, 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. Texas’s projected net population increase is over eight million, Arizona over two million, and Colorado nearly one and a half million. Approximately 685,000 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. The key assumptions underlying our strategic efforts are:

- **Collaboration** is essential to deliver genetics services to unique populations, culturally and linguistically distinct from majority populations. The Mountain States Region has large concentrations of Hispanic/Latino populations and Native Americans, as well as geographic diversity with a high percentage of rural and frontier counties.

- **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.

- **Quality services with innovation**: Innovation is essential for strengthening NBS 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, 2015, MSGRC hosted or sponsored 49 collaborative sessions using multiple venues (in-person meetings, webinars, and teleconferences). These collaborative sessions facilitated information sharing among 723 consumers and professionals about critical issues in genetics services (medical home, NBS capacity building, collaboration, Affordable Care Act implementation, NBS long-term follow-up, telehealth, and transition from pediatric to adult care. Through these collaborative sessions, MSGRC formally conducted the first phase of a regional needs assessment, focused specifically on determining the priority needs of the region by engaging stakeholders, facilitating input on regional needs, and assessing provider strengths and capacity to meet identified needs. Using web and social media, MRGRC reinforced and broadened the impact of the meetings, webinars, reports and conferencing. The MSGRC website and Facebook published highlights, shared resources, encouraged collaboration, and directed consumers, genetic counselors, genetics providers, medical geneticists, other specialists, primary care physicians and other healthcare providers, social service providers and public health professionals to relevant genetics resources. Through 5,121 unique website visits, consumers and professionals viewed information related to medical home, newborn screening capacity, long term follow-up, collaborations, ACA implementation and transition from pediatric to adult care. MSGRC currently has 417 “likes.”

MSGRC conducted and participated in regional and national projects related to ACA implementation and insurance coverage for genetic disorders (including website posting of the MSGRC ACA policy brief), inborn errors of metabolism, long-term follow-up of newborn screening, use of parent partner navigators in pediatric care for families of children with heritable conditions, telegenetics case study planning and provider recruitment, genetics services mapping, genetic services needs assessment, and the evaluation of regional models as genetic resource centers.

MSGRC supported the Parent Partners Project, which directly improved medical home services to children with special healthcare needs and their families in Texas, Wyoming and Montana.

How well did you do it?

The five MSGRC Workgroups (Consumer Advocacy, Newborn Screening, Telegenetics, Emergency Preparedness, Medical Home) and two special interest groups (Metabolic Consortium and Hemoglobinopathies) convened via
calls and an annual conference focused on strategic initiatives, projects and deliverables assigned to each workgroup. 

Members of the MSGRC Workgroups reported high levels of effective collaboration (i.e., timely topics, strong structures, staffing and membership, good process and results achieved) as measured by the annual Working Together Survey.

MSGRC successfully focused attention, expanded knowledge, and made recommendations aimed at improving access and reducing barriers to quality genetics services. Specifically, the Workgroups and MSGRC leadership:

- Increased diverse consumer involvement in MSGRC Workgroups, and regional and national committee work;
- Held regional needs assessment and priority setting sessions via in-person focus groups and conference calls;
- Hosted meetings and regional panel presentations to share effective practices for use in newborn screening labs, testing, and data exchange;
- Offered continued support to the Parent Partners Project which successfully expanded to multiple new sites via state funding through Montana and Wyoming;
- Enhanced the networking and information-sharing among consumers in the Mountain States through the MSGRC Facebook, which provides credible, practical and difficult to find resources and information.

MSGRC strengthened partnerships with stakeholders, including governmental and community-based organizations and other HRSA MCHB-funded programs. MSGRC staff have national leadership roles on the key committees. Dr. Celia Kaye, MSGRC consultant, chaired the NCC Regional Support Services Model (RSSM) Workgroup and Dr. Kathryn Hassell is a workgroup member. Dr. Janet Thomas, MSGRC Associate Project Director, chairs the NCC Identifying Genetic Services Small Group. Dr. Hassell co-chairs the NBSTRN Clinical Integration Group and is on the Steering and NBS Definitions Committees for Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). Drs. Hassell and Kaye serve on the ACHDNC Follow-Up and Treatment workgroup and Joyce Hooker serves on the ACHDNC Education workgroup. Through strong leadership and partnership, MSGRC is able to leverage resources, test innovative models for improving services and clinical practice, and support national medical home priorities.

Was anyone better off?

Through innovative mini-projects and the work of the Hemoglobinopathies Interest Group, MSGRC tested innovative medical home-centered approaches to improve access to quality genetic services and coordinated care for families with heritable disorders. MSGRC is implementing these approaches in pediatric practices (e.g., use of Parent Partner navigators in Wyoming and Montana practices), in state newborn screening laboratories (e.g. best practices for transferring blood samples from birth hospitals to state lab, newborn screening follow-up programs (e.g. increasing awareness of new sickle cell disease guidelines) and in specialty practices (e.g., transition program development at the Colorado Sickle Cell Center). These projects directly benefit the lives of families affected by genetic disorders, as well as improve the capacities of clinical practices, hospitals and public health laboratories to provide genetics services to individuals and families in geographically and culturally diverse communities.

Regionally and nationally, the MSGRC uniquely contributes to improving the care of individuals with sickle cell disease. Through the leadership of Dr. Kathryn Hassell, the Hemoglobinopathies Interest Group and national collaborators are increasing national visibility and raising awareness about sickle cell diagnosis, treatment, case reporting and long term follow-up guidelines. The group is assessing the feasibility of Sickle Cell patient data collection as part of Title V or other Children with Special Healthcare Needs program reporting. In the next year, we will assess needs and develop a strategic plan for the Mountain States Regional Genetics Resource Center. We will build on our strengths - collaboration, diverse consumer involvement, strong networks of genetics service providers and public health systems, and innovation.
Western States Genetic Services Collaborative (WSGSC) Highlights 2015-2016

How much did you do?

Major Projects

- Affordable Care Act Family Survey
- 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
- Genetic Services Outcomes
- Unity GC – Needs Assessment of Adolescents with Genetic Conditions
- Newborn Screening Survey
- Centralized Authority on the Prior Authorization of Genetic Services
- Genetic Services Outcomes

How well did you do it?

Affordable Care Act Family Survey – survey designed in conjunction with other regional genetics collaboratives enabled both a regional and national level understanding of current status of health care and insurance needs for population living with genetic conditions.

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 Outcomes
UNITY-GC Survey – documented needs of adolescents living with genetic conditions, results to be used regional in program planning.  
Newborn Screening Survey – assessing parent opinion on preferences for notification of risk related to age of symptom onset from positive newborn screen, findings available July 2016.  
Centralized Authority on the Prior Authorization of Genetic Services – shared information about this innovation model for improving reimbursement with other regions, model includes a Genetics Advisory Board consulting to Medicaid on insurance reimbursement issues.  
Genetic Services Outcomes – convened teleconference of regional stakeholders to share information about current work in measuring outcomes of genetic services, conducted literature review related to outcomes of genetic services, shared with national stakeholders, wrote framework for approach to designing genetics services outcomes measurement

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 Outcomes – see outcomes below

UNITY-GC Survey – findings from this survey are used by family advocate organizations and state programs to plan interventions for adolescents in the health care system  
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.  
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.
## New England Genetics Collaborative Regional Activities

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<tr>
<th>Intra-Regional</th>
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<tr>
<td><strong>Title:</strong> Medical Home Webinar Series</td>
<td><strong>Medical Home</strong></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>
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<tr>
<td><strong>Accomplishments:</strong> Held 4 webinars (225 total participants) educating care providers around concepts of creating a shared plan of care, documenting family history, incorporating genetics in the management of children in the primary care office, and holding family centered conversations. Based on satisfaction surveys collected across the 4 webinars held, participants agreed that the educational session: enhanced their competence (97%, N=74); influenced their practice (90%, N=70); and impacted patient outcomes (90%, N=70). More details on the webinar series (including recordings) are accessible here: <a href="http://www.negenetics.org/workgroups/medical/products_pubs_mh">http://www.negenetics.org/workgroups/medical/products_pubs_mh</a>.</td>
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### NBS Capacity Building

| **Title:** Establishing a common framework for CCHD Screening | **NBS Capacity Building** |
| **Description:** Established a consistent method for the screening and reporting of CCHD across a diverse array of health centers | |
| **Who with:** 8 Birthing Centers across Maine, New Hampshire, Vermont, Rhode Island and Connecticut | |
| **Accomplishment:** Established a consistent method for screening and reporting of CCHD across New England. Over 42,000 children screened since the project started, 3 children diagnosed with CCHD. An electronic resource was developed for families on psychosocial supports and went live in May 2016 ([www.necongenitalheartresources.org](http://www.necongenitalheartresources.org)). 500 resource bags were | |
distributed to 4 pediatric cardiology practices for distribution to families that receive a CCHD diagnosis.

**Collaboration**

**Title:** Quality Improvement Workgroup  
**Description:** 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 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 May 2016, 2,042 individuals with DD/ID have been entered into the registry.

**Title:** GEMSS Resource Website for Schools, Families, and their Children  
**Description:** Provides users with a broad range of condition specific information on how to best support children with a genetic condition in a school setting.  
**Who with:** families, consumers, & advocacy organizations from across the US  
**Accomplishment:** Provided a high quality and useful resource to nearly 20,000 individuals during the 2015/2016 project year. Since its launch in 2012, GEMSS has provided critical information to over 62,000 people across the world with 20,000 from outside the US.

**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:** Presentations by NEGC key leaders (Mark Korson, Rosemarie Smith, Sheila Upton) ~75 participants.

**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 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. There are ongoing discussions about a possible “core curriculum” and/or specific performance measures without a specific recommendation of either at this time. The AUCD website has a specific page for genetics education that includes the genetics education modules for programs to share and to contribute to.

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**Affordable Care Act**

**Title:** Research Brief: Promoting and Improving the Health and Well-Being of People with Inherited Conditions

**Description:** The Health Care Access and Financing Group of the NEGC continued its efforts to document the experiences of families and individuals living with a genetic condition. The focus of this year's efforts was on implementing an expanded survey among families in the New England region who cared for an individual with a genetic condition and summarizing the results.

**Who with:** Families, Advocacy Organizations, Catalyst Center, NEGC Staff

**Accomplishment:** Initial findings were shared with members of the HAF workgroup, a posterboard created for an annual presentation of

**Title:** NCC Health Care, Access, and Financing Group

**Description:** Ongoing collaboration with members of the NCC to review ongoing policy developments and field research. Co-authored a new publication by the Genetic Alliance reviewing national survey work on individuals living with a genetic condition.

**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.
LEND trainee work, and a research brief was released in May of 2016 highlighting some of the major findings of the work.

### Transition

<table>
<thead>
<tr>
<th>Title: Face Forward 2015 Summer Conference for Youth</th>
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<tbody>
<tr>
<td><strong>Description:</strong> Three day retreat experience for youth with genetic conditions, designed to explore transition issues, including “difficult conversations; sponsored by Next Step, Inc.</td>
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<tr>
<td><strong>Who with:</strong> youth and young adults ages 16-24 with metabolic conditions, Neurofibromatosis, type 1, mitochondrial disease or other genetic conditions.</td>
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<tr>
<td><strong>Accomplishment:</strong> Eighteen youth and young adults from across the country gained invaluable experiences propelling them toward self-advocacy. In a pre and post comparison of participant perceptions concerning whether or not they would be willing to advocate for themselves or others living with their condition, the percentage increased from 78% to 92%.</td>
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<table>
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<tr>
<th>Title: Health Surveys of Adults Living with Genetic Conditions</th>
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<tbody>
<tr>
<td><strong>Description:</strong> Continuing to collect adult health survey data to document potential disparities in health and well-being among those with and without targeted conditions.</td>
</tr>
<tr>
<td><strong>Who with:</strong> Members of the NEGC and NCC transition workgroups.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> To date, survey data has been collected from 85 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>
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<tr>
<th>Title: Homocystinuria/Metabolic Support Group</th>
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<tr>
<td><strong>Description:</strong> Dr. Waisbren provided support to a young man in his efforts to coordinate a support group for individuals living with genetic conditions.</td>
</tr>
<tr>
<td><strong>Who with:</strong> Individuals living with homocystinuria and metabolic conditions.</td>
</tr>
</tbody>
</table>
### Accomplishments: 8 individuals participated in the support group (met 1x).

### Title: Implementation of Transition Policy
**Description:** Dr. Waisbren worked with a range of partners to develop a transition policy based on best practices to help ensure appropriate levels of care for individuals living with genetic conditions.
**Who with:** Children's Hospital Boston, Area Metabolic Clinics
**Accomplishments:** 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 [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/).

### Telegenetics

**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 developed a HIPAA compliant telemedicine system for hosting conferences between specialists and other care providers, launched a pilot effort with three clinical sites from across the New England region and completed 19 consultations with providers.
New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC) Regional Activities

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<tr>
<th>Intra-Regional</th>
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<tr>
<td><strong>Title:</strong> Medical Home Webinar Series</td>
<td><strong>Title:</strong> NCC Care Coordination Workgroup</td>
</tr>
<tr>
<td><strong>Description:</strong> Webinar series</td>
<td><strong>Description:</strong> Two parent advocates, a primary care provider, and NYMAC team members continue to participate in the NCC Care Coordination Workgroup.</td>
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<tr>
<td>NYMAC delivered six webinars surrounding care coordination for providers, families, and advocates at a rate of one per month between January and May of 2016. Participants were able to register via separate registration links per webinar; there was no cost to participate. CME credits were offered.</td>
<td><strong>Who with:</strong> Parents/advocates and representatives from different regions and national partners.</td>
</tr>
<tr>
<td><strong>Who with:</strong> Parents/advocates, physicians, allied health professionals, trainees, public health professionals, and genetics professionals.</td>
<td><strong>Accomplishments:</strong> NYMAC contributed to the exemplary practices document developed by the NCC Care Coordination Workgroup.</td>
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<tr>
<td><strong>Accomplishments:</strong></td>
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<tr>
<td>Between the six webinars, there were 591 unique attendees, with a maximum of six webinars per attendee. The webinar topics, speakers, number of attendees, and number of completed evaluation forms were as follows:</td>
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<tr>
<td>1. Practice team huddles, led by Robert Ostrander, MD.</td>
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<tr>
<td>• 104 attendees, 51 completed evaluations</td>
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<tr>
<td>2. Integrating behavioral and mental health in the medical home, led by Mitzi Glass, LCSW, and Nicole Pratt, MA.</td>
<td></td>
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<tr>
<td>• 120 attendees, 46 completed evaluations</td>
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<tr>
<td>3. Improvement models, led by Ruth Gubernick, PhDc, MPH, PCHM, CCE.</td>
<td></td>
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<tr>
<td>• 103 attendees, 47 completed evaluations</td>
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<tr>
<td>4. Cultural humility, led by Joann Bodurtha, MD, FAAP, FACMG, Maria Isabel Frangenberg, BA, and Helen Dao, MHA.</td>
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<tr>
<td>• 92 attendees, 41 completed evaluations</td>
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<tr>
<td>5. Resources on care coordination, led by Malia Corde, BA, and Shama Khan, MS, CGC.</td>
<td></td>
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<tr>
<td>• 90 attendees, 45 completed evaluations</td>
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<tr>
<td>6. Patient centered coordination, led by Renee Turchi, MD.</td>
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<tr>
<td>• 82 attendees, 33 completed evaluations</td>
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On average, 92% of respondents found that the webinar sessions were “very good” or “excellent;” 97% of respondents “agree” or “strongly agree” that the speakers were knowledgeable about the subject. On average, 81% of respondents found the webinars “relevant” or “very relevant” to their work, and 59% “agree” or “strongly agree” that the information presented would influence the way that they practice. Respondents offered insight via open-ended questions after each webinar, which were shared with NYMAC AC Members at the annual meeting.

### NBS Capacity Building

<table>
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<tr>
<th>Title</th>
<th>Description</th>
<th>Who with</th>
<th>Accomplishments</th>
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<tbody>
<tr>
<td>MPS1/Pompe Symposium</td>
<td>An educational meeting for Newborn Screening for MPS1 and Pompe disease occurred over November 18-19th in Philadelphia.</td>
<td>NYMAC States, IL, MN, APHL &amp; PE; Attendees were providers, public health professionals, and consumers.</td>
<td>Educated 44 participants on clinical considerations, laboratory screening, molecular testing, follow-up on late-onset disorders, ACHDNC, and barriers/solutions to implementation. Post-test evaluations showed that all participants described the information presented as useful to their job, and most providers planned to implement the activities described in the symposium between 2016 and 2017.</td>
</tr>
<tr>
<td>IBEMC Project</td>
<td>Funding Metabolic Centers to Enter Data into IBEMC</td>
<td>Six centers in the NYMAC Region were funded</td>
<td>Contributed to National NBS Data Collection Effort</td>
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<tr>
<th>Title</th>
<th>Description</th>
<th>Who With</th>
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<tr>
<td>Dietitian Group Meeting</td>
<td>A NYMAC meeting was held in conjunction with the Genetic Metabolic Dietitians International Conference on Metabolic Nutrition in the 21st Century in Scottsdale, AZ in April, 2016. Content included an in-depth presentation about “Medical Foods: Getting Products to Patients” by Kathryn Camp, MS, RD, CSP of the NIH, followed by a discussion addressing reimbursement issues.</td>
<td>Metabolic dietitians across the NYMAC region.</td>
</tr>
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</table>
**Accomplishments** – This was the first in-person meeting of this group; the meeting helped to improve understanding of what is a medical food as defined by statute, medical foods coverage and who pays, the role of federal agencies in coverage, and reimbursement of medical foods. There is currently discussion of next steps for the group, including an upcoming conference call to discuss preliminary analysis of the medical foods survey results.

<table>
<thead>
<tr>
<th>Title: NewSTEPS</th>
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<tr>
<td><strong>Description:</strong> An application for additional funding was submitted under the NewSTEPS360 program. Five NYMAC states (NY, NJ, MD, DE and VA) are participating.</td>
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<tr>
<td><strong>Who With:</strong> NewSTEPS and the Association of Public Health Laboratories.</td>
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<tr>
<td><strong>Accomplishments:</strong> NYMAC was the only region to receive funding for a project through NewSTEPS. Individuals from all 5 member states participated in the kick-off meeting in mid-January. Monthly calls are being coordinated and conducted. An assistant research scientist has been hired to conduct a survey of birth hospitals on electronic capabilities including the type of electronic medical record and existing HL7 demographic messages created by their system. The survey is under development and hospital contacts will be identified.</td>
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<tr>
<th>Title: NBS Follow-up Group Meeting</th>
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<tr>
<td><strong>Description:</strong> Meeting on short-term follow-up which was held in conjunction with the MPS1/Pompe Symposium occurred on November 18 in Philadelphia.</td>
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<tr>
<td><strong>Who With:</strong> State NBS Follow-up staff in the NYMAC Region.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> The meeting included discussion of the future direction of the group (data-based studies vs. white papers vs. parent materials), timeliness activities – NYMAC’s current projects, the NewSTEPS application (Borderline and Invalid Specimen study), medical foods projects, NCC Long-term Follow-up Study, and NBS</td>
</tr>
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</table>
Program collaboration with other Department of Health Programs (vital records, newborn hearing screening).

**Collaboration**

**Title:** NYMAC Regional Genetics Education Network  
**Description:** The RGEN is an organization of volunteer patients, advocates, providers, and genetic counselors who are willing to provide in-person or webinar-based education on genetics and newborn screening.  
**Who with:** Patients, providers, public health and genetic counselors  
**Accomplishments:** NYMAC advertised via our Facebook page, NYMAC News emails, and a mailing to genetic counselors in the region. There are currently 56 individuals registered for the RGEN.

**Title:** NCHAM collaboration & Hearing Loss survey  
**Description:** Collaborators (see “Who with”) engaged in discussions to identify needs for genetic assessment in hearing loss. A survey of pediatricians was developed and has been approved by the institutional review board at Johns Hopkins.  
**Who with:** HRSA HL program, Title V, EHDI and EI programs, EHDI – AAP Chapter Champions, LEND faculty, Heartland, Region4, and other national/regional experts such as Arti Pandya and Kathleen Arnos.  
**Accomplishments:** The Hearing Loss Survey is currently being pilot tested by the pediatricians and family medicine practitioners at Accountable Health Partners NY and IGM. The Hopkins IRB approved survey, which will be disseminated via paper and print to health care providers in the region for 6-8 weeks with data entry closing on August 31st, 2016.

**Title:** Genetic Alliance- Fact Sheet  
**Description:** A fact sheet on whole exome sequencing was developed in collaboration with the Genetic Alliance and the NYMAC CCN.  
**Who with:** Genetic Alliance, NYMAC CCN, Patients  
**Accomplishments:** The fact sheet was developed and distributed via the NYMAC Facebook page, NYMAC website and sent to the other Regional Collaboratives. The CCN and an MD geneticist provided critical review.

**Title:** Consumer Collaborative Network meeting in Pittsburgh  
**Description:** In the fall of 2015, 20 individuals attended the NYMAC CCN in Pittsburgh PA. The theme of the meeting was to “share your story.”  
**Who with:** Consumer Collaborative Network; NYMAC, parents/advocates  
**Accomplishments:** NYMAC continued to collaborate with parent groups. The group decided to create a “Parent’s Corner” on the NYMAC website, which allows for stories to be shared. In response to a critical review of consumer-focused educational materials, the decision was made to create fact sheets in collaboration with Genetic Alliance.

**Title:** Title V Meeting at AMCHP – April 8, 2016  
**Title:** NCC Regional Model Recommendations
**Description**: A webinar occurred on Feb 29th with representatives from all states/district in the NYMAC region. A special interest session was held on April 8 at the rescheduled AMCHP 2016 Annual Conference with 17 representatives and Title V Directors from DC, DE, MD, NJ, NY, and VA, HRSA, Heartland, and WSGRC.

**Who with**: Representatives from states and State Title V Director/CYSCHN coordinators

**Accomplishments**: NYMAC continued to develop meaningful relationships with the Title V/CYSHCN directors, as representatives from six NYMAC states attended the meeting. Discussion focused on areas for collaboration, the NYMAC genetics in hearing loss project, and the outcomes from the Genetics System Assessment (GSA) conducted in Year 3. A matrix of GSA findings, Title V performance measures, and possible NYMAC activities was prepared by our evaluation team to facilitate the discussion and was disseminated during the AMCHP – NYMAC Session. NYMAC established contacts with state Medicaid programs to compile information on Medicaid policies on genetics services.

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**Description**: A Regional Support Service Model Workgroup reviewed existing models of regional care and developed recommendations for a model or models for care.

**Who with**: National Coordinating Center, NYMAC’s Joann Bodurtha was on an advisory committee to the workgroup, and Michele Caggana was on the workgroup.

**Accomplishments**: Members of NYMAC helped contribute to the NCC Regional Model Recommendations through workgroups and advisory committees.

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**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. This survey had a maximum of 98 questions, with skip logic allowing participants to answer only the questions relevant to their experience with management of 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 and insurance coverage. The results of the survey will be published, and used to create advocacy materials and resources to help individuals and families obtain coverage.
**Who with:** Genetic dietitians in NYMAC region, individuals with PKU, and their parents and/or caregivers.

**Accomplishments:** After pilot testing, the survey was distributed via surveymonkey.com in May of 2016. NYMAC distributed the survey to the National PKU Alliance, who then distributed the survey to its membership in June. By Sunday, June 26th, the survey had gathered 227 responses.

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**Telegenetics**

**Title:** Expansion of telemedicine for sickle cell disease (SCD) patients

**Description:** NYMAC funded Children’s Hospital of Pittsburgh to conduct telemedicine training sessions for pediatric hematologists from across the region.

**Who with:** Children’s Hospital of Pittsburgh (CHP), pediatric hematologists and their team members in the NYMAC region

**Accomplishments:** Nine pediatric patients with SCD at the Erie location and two pediatric patients with SCD at the Johnstown location completed 94 and 7 telemedicine visits respectively before Dec, 2015. The proportion of patients receiving hydroxyurea was 73% with improved adherence (monthly visits). The frequency of visits increased from twice yearly to quarterly visits. Patients/families reported improved satisfaction owing to crucial time and saving of resources. Two half-day workshops were conducted at the CHP on March 15th and April 12th with pediatric hematologists from DE, NJ, NY, and VA. The benefits (access to care and hydroxyurea, increased workforce capacity) and barriers and strategies to implementing telemedicine programs were discussed in these workshops.

NYMAC presented a poster and conducted a special interest telemedicine session with Dr. David Flannery and Dr. Debra Cohen at the ASPHO2016 Annual Meeting in May’16.

NYMAC participated in the ATA2016 to learn the existing landscape of telemedicine services and explore telegenetics opportunities. A public health-telegenetics needs assessment is underway.

**Title:** NCC Telegenetics Workgroup

**Description:** A genetic counselor and NYMAC team members continue to participate in the NCC Telegenetics Workgroup.

**Who with:** Representatives from different regions and Telehealth Resource Centers

**Accomplishments:** NYMAC continues to contribute to the NCC Telegenetics Workgroup efforts including work on telegenetics outcomes.
### Title: Hospital Engagement Needs Assessment and Demonstration Project / Genetics Help Desk

**Description:** The project was carried out over three phases.

- **Phase 1** took place between December (2015) to January (2016), and consisted of a baseline needs assessment in which 56 rural community hospitals in the NYMAC region were surveyed and researched to assess their access to genetics. Hospitals were grouped to be offered the provider reference service or reference services and telegenetic services.

- **Phase 2** is currently in place and has been so since February of 2016. This phase involves hospital and provider invitation and engagement. Letters, phone calls, mailings, faxes, emails and meetings were employed to engage providers and gather feedback.

- **Phase 3**, the genetic help desk utilization phase, has been in place since April.

**Who With:** The Ferre Institute (Luba Djurdjinovic and Alissa Bovee Terry), providers in the NYMAC region.

**Accomplishments:** Twenty-nine (OBGYN/PCP/oncology) practices were surveyed via a scripted interview. Data was collected regarding their genetic testing and referral practices, capacity, and others. Many lessons were learned from this activity for future efforts to engage hospitals and providers regarding genetics consultation and support for telegenetics.

### Other

**Title:** Provider Needs Assessment

**Description:** In the fall of 2015, the NCC conducted a provider needs assessment survey to better understand national and regional genetic needs. The survey covered a variety of areas, including current conditions and gaps in genetic practices, areas of unmet service needs, and desired components of a future regional genetic resource.

**Title:** Genetics/Genomics in Public Health Fellowship

**Description:** Based on the NCC Advocate leaders program model, NYMAC provided this online Fellowship opportunity to LEND and Genetic Counseling trainees (referred to as fellows). Eighteen Fellows (1 outside the NYMAC region) participated in three webinars and three LinkedIn discussions on topics related to CADASIL, special
center. Each RC was also able to include some questions specific to their region; NYMAC contributed 15 additional items to the survey.  
**Who with:** Health care providers in the NYMAC region.  
**Accomplishments:** The survey garnered 143 maximum responses per question. The NYMAC evaluation team analyzed the provider survey data and presented the results at the NYMAC Advisory Council meeting in May, 2016.

| Title: Consumer Benefits Survey Webinar and Publication | Description: Results of the consumer benefits survey were shared in a webinar on September 9th, 2015.  
**Who with:** Genetics consumers in the NYMAC region.  
**Accomplishments:** Thirty-five participants attended the webinar. The results of the consumer benefits survey were published in Exceptional Parent Magazine in November of 2015. |
| --- | --- |

| Title: Fragile X Project | Description: Data from Labcorp and the Institute for Basic Research was analyzed and used to reveal opportunities for education regarding screening for fragile X syndrome.  
**Who With:** Fragile X Alliance  
**Accomplishments:** The analyzed data was presented to the AC meeting and was used to highlight needs regarding FXS screening education. In addition, a postcard offering information regarding FXS screening was sent to 10,032 offices throughout the NYMAC region. |
| --- | --- |
Southeast Regional NBS & Genetics Collaborative (SERC) Regional Activities

<table>
<thead>
<tr>
<th>Intra-Regional</th>
<th>Inter-Regional</th>
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<tbody>
<tr>
<td><strong>Medical Home</strong></td>
<td><strong>NBS Capacity Building</strong></td>
</tr>
<tr>
<td><strong>Title:</strong> Web-based evidence and consensus based guidelines</td>
<td><strong>Description:</strong> 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</td>
</tr>
<tr>
<td><strong>Description:</strong> 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</td>
<td><strong>Who with:</strong> Collaboration with GMDI members, parent organizations and AHRQ</td>
</tr>
<tr>
<td><strong>Who with:</strong> Collaboration with GMDI members, parent organizations and AHRQ</td>
<td><strong>Accomplishments:</strong> PKU guidelines have been completed and published in a peer-reviewed journal. Web based portal with published guidelines Linking food lists with parent organizations. MSUD guidelines were accepted and released through AHRQ.</td>
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<table>
<thead>
<tr>
<th><strong>Collaboration</strong></th>
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<tbody>
<tr>
<td><strong>Title:</strong> SERC SERGG annual meeting 2015</td>
</tr>
<tr>
<td><strong>Description:</strong> 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 building with local, statewide and national partners. The 2015 annual meeting was held from July 16-18 at the Renaissance Hotel in Asheville, North Carolina.</td>
</tr>
<tr>
<td><strong>Who with:</strong> In attendance were 176 healthcare professionals with an interest in genetics and public health. Fourteen consumers also attended.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong> Thirty one platform and twenty three poster presentations were made covering current issues in all areas of genetics: molecular genetics, cytogenetics, biochemical genetics and clinical genetics. Highlights included sessions on newborn screening for Pompe’s disease in the US, a telemedicine demonstration project, a report on evidence and rational for newborn screening for MPSI along with transitioning to adult healthcare, and building family...</td>
</tr>
</tbody>
</table>
communities to enhance knowledge and improvement of patient care for rare genetic disorders.

### Affordable Care Act

<table>
<thead>
<tr>
<th>Title</th>
<th>Exploration of medical foods and Affordable Care Act</th>
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<tbody>
<tr>
<td><strong>Description:</strong></td>
<td>Strategies to include medical foods as a part of the Affordable Care Act</td>
</tr>
<tr>
<td><strong>Who with:</strong></td>
<td>Catalyst center</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong></td>
<td>A project has been identified with the catalyst center to update information related to medical foods legislation in each state.</td>
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</tbody>
</table>

### Long-Term Follow-Up

<table>
<thead>
<tr>
<th>Title</th>
<th>SERC Pilot proposal for Long-term follow-up (LTFU) utilizing Longitudinal Pediatric Data Resource (LPDR)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Description:</strong></td>
<td>SERC was awarded a grant to support the vision of the national project to build a strong infrastructure which offers definitions, tools and resources that are uniform, simple and timely to support public health and clinical data collection to further understand the impact of NBS.</td>
</tr>
<tr>
<td><strong>Who with:</strong></td>
<td>SERC will be working with NCC/NBSTRN and participating states in our region (SC, TN, LA, GA).</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong></td>
<td>This project will help demonstrate the feasibility of public health teams (within 2-3) to conduct long-term follow-up data collection.</td>
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<table>
<thead>
<tr>
<th>Title</th>
<th>Long-term follow-up (LTFU) collaboration with National Coordinating Center (NCC)</th>
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<tbody>
<tr>
<td><strong>Description:</strong></td>
<td>Face-to-face meeting</td>
</tr>
<tr>
<td><strong>Who with:</strong></td>
<td>NCC, APHL (New Steps) and NBSTRN</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong></td>
<td>The group identified minimum data elements for LTFU common to public health for quality improvement, research and new knowledge generation. Goals include to obtain feedback at the state and regional levels and create a pilot to validate the common data elements and to develop processes for integration with technology.</td>
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### Transition

<table>
<thead>
<tr>
<th>Title</th>
<th>SERC Consumer Delphi Survey</th>
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<tbody>
<tr>
<td><strong>Description:</strong></td>
<td>SERC is working on conducting a Delphi study on transition care success utilizing our SERC consumers as the experts in order to gain a more in-depth understanding on what transition success means to them.</td>
</tr>
<tr>
<td><strong>Who with:</strong></td>
<td>SERC consumer alliance group leaders in every state in our region.</td>
</tr>
<tr>
<td><strong>Accomplishments:</strong></td>
<td>It is our hope that the dissemination of results from the Consumer Transition Delphi study will improve transition</td>
</tr>
</tbody>
</table>
care and quality for consumers and families affected by genetic disorders. The long-term outcome is for health professionals to utilize the results of this study to create better transition care programs for individuals and families affected by genetic disorders.

Telegenetics

**Title:** Doxy.me Telegenetics Technology Collaboration  
**Description:** Doxy.me is a simple, secure, HIPAA-compliant and free telemedicine available to any clinician in the world. Clinicians can use the platform to provide care to their patients, for clinical research or clinical trials.  
**Who with:** Regional healthcare providers. This new technology was demonstrated to various healthcare providers who participated in the Telegenetics session at the SERC SERGG meeting.  
**Accomplishments:** The group feedback indicated that the use of this technology can potentially revolutionize the delivery of healthcare by making it more convenient and accessible for patients to access qualified healthcare professionals, and reducing unnecessary expenses. Application of this technology will be further explored in our region.

**Title:** Heartland Genetics Services Collaborative Training in Nashville, TN.  
**Description:** The Western States and Heartland Regional Genetics Collaborative have developed a telegenetics training program which includes distance learning and hands-on training components in collaboration with national Telehealth Resource Centers.  
**Who with:** The training program will introduce practicing and trainee geneticists and genetic counselors to the world of telegenetics. A member of our region has been approved to attend this meeting.  
**Accomplishments:** To increase access to genetic services and education nationwide

Other

**Title:** Lunch and Learn Series: “Pompe Disease: Newborn Screening Initiative in the US.”  
**Description:** A lunch and learn series event was organized in October 2015 to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students on the topic of Newborn screening for Pompe disease.  
**Who with:** A total of 25 individuals participated in person and 11 individuals participated via webinar throughout the region.  
**Accomplishments:** The lunch and learn series resulted in a 124% increase in knowledge about Pompe disease evidenced by results from the pre and post-test survey administered at this event.

**Title:** Lunch and Learn Series: “MPS I disease: phenotypes and considerations for newborn screening.”  
**Description:** A lunch and learn series event was organized in May 2016 to educate healthcare providers, laboratory personnel, clinicians, genetic counselors, public health staff, dietitians and students on the topic of phenotypes and considerations for newborn screening of MPSI disease.  
**Who with:** A total of 14 individuals participated in person and 13 individuals participated via webinar throughout the region.  
**Accomplishments:** The lunch and learn series resulted in a 183% increase in knowledge about MPS I disease evidenced by results from the pre and post-test survey administered at this event.
## Region 4 Midwest Genetics Collaborative Regional Activities

<table>
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<tr>
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<tr>
<td><strong>Medical Home</strong></td>
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</tbody>
</table>
| **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 99 caregivers in five trainings offered in collaboration with the MI Department of Health and Human Services. | **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:** Heartland trained facilitators  
**Accomplishments:** The Care Coordination training continues to impact families across the country. Heartland Collaborative trained an approximate 80 caregivers across 11 partnering agencies. |
| **NBS Capacity Building** | **NBS Capacity Building** |
| **Title:** Hemoglobinopathies Workgroup—Sickle Cell Trait  
**Description:** Hemoglobinopathies Workgroup held an in-person meeting to finalize sickle cell trait short term follow-up recommendations.  
**Who with:** 13 workgroup members, the Midwest Sickle Cell Demonstration Project (STORM) families, clinicians, community based organizations, and public health  
**Accomplishments:** Finalized 18 short term follow-up recommendations and developed a plan to get feedback from consumers and experts on the recommendations before incorporating them into a publication for a peer-reviewed journal. Plans include evaluating implementation of these recommendations within the region. | **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 102 prenatal care providers, child birth educators, and other health care professionals through online course available on Region 4’s website. |
| **Title:** Sickle Cell Trait educational materials dissemination  
**Description:** Disseminated sickle cell trait educational material across the region. *Sickle Cell Trait: What You Need To Know* was developed by the Region 4 hemoglobinopathies workgroup.  
**Who with:** Region 4 NBS Follow-Up programs, community-based sickle cell organizations, clinical providers, genetic counselors. | **Title:** ACHDNC Participation  
**Description:** Regional representation and participation on the Secretary’s Advisory Committee  
**Who with:** George Dizikes, Mei Baker, Sue Berry  
**Accomplishments:** Regional support for participation on ACHDNC workgroups and committees |
**Accomplishments:** Distributed two types of educational materials (*Sickle Cell Trait: What You Need To Know* brochure and a sickle cell trait flyer) to about 1500 stakeholders across the region. Materials were translated into French and Spanish and distributed in three languages.

**Title:** Lysosomal Storage Disorders Implementation  
**Description:** Facilitated technical assistance and information sharing around Lysosomal Storage Disorders screening  
**Who with:** Region 4 NBS Laboratory Forum, RUSP Implementation Workgroup, NBS program representatives  
**Accomplishments:** Facilitated technical assistance among the Region 4 states regarding the addition of Lysosomal Storage Disorders to the Recommended Uniform Screening Panel through one in-person RUSP Implementation meeting and 5 NBS Laboratory webinars. Attendees from NBS programs and NBS laboratories participated in discussions and heard presentations from Minnesota and Illinois.

**Collaboration**

**Title:** LEND Collaboration  
**Description:** Joint LEND/Region meetings  
**Who with:** Region 4 staff and 4 LEND programs within the region.  
**Accomplishments:** Participated in the joint RC/LEND meeting in conjunction with the NCC annual meeting. Developed an action plan for the coming year. Last year’s action plan resulted in LEND representation at Region 4 Midwest’s Regional Meeting.

**Title:** Family Forum Video Project  
**Description:** Produced a second video in a series of 7 parents talking about their experience receiving a genetic diagnosis and moving forward from that diagnosis to managing their child’s care and life.  
**Who with:** Seven Region 4 parents from 5 states representing children with PKU, Sickle Cell Disease, Syndrome without a Name, Cystic Fibrosis, and other genetic conditions.  
**Accomplishments:** Disseminated video, *Receiving a Diagnosis: From Cope to Hope* professionally produced in Year 2 and second video.

**Title:** Care Coordination: Empowering Families training Fidelity of Implementation Analyses  
**Description:** Analysis and review of Fidelity of Implementation ratings for new Care Coordination: Empowering Families training facilitators.  
**Who with:** Heartland Region  
**Accomplishments:** With the expansion, a new fidelity of implementation tool has been used to get feedback from facilitators about their experience in the training. To date, we have feedback from 19 new facilitators who rate their experience across four
**Care Coordination – A Family Perspective** in Year 4. Videos had 2,264 views this year. Videos available on Region 4 Midwest website: [www.region4genetics.org/families](http://www.region4genetics.org/families)

Domains on a scale from 1 to 9 (1-3 “Needs Work”, 4-6 “Acceptable”, 7-9 “Good Work”) and then provide qualitative evidence to support their rating. Mean ratings range from 7.45 to 8.15, indicating a high level of fidelity. Staff continue to monitor this feedback to understand where additional supports may be necessary.

**Title:** 2015 Regional Meeting  
**Description:** Annual meeting held from October 19-21, 2015  
**Who with:** 69 stakeholders from seven states  
**Accomplishments:** Five in-person Workgroup/Forum Meetings, 4 multi-presenter plenary panel presentations, 10 break out promising practice/new idea sessions, and stakeholders meeting by state to complete the Genetic Systems Assessment tool.

**Title:** Heartland Care Coordination Webinar  
**Description:** Technical Assistance Webinars for newly trained facilitators  
**Who with:** Heartland Collaborative  
**Accomplishments:** Provided three technical assistance webinars for the Heartland Genetics Services Collaborative Care Coordination: Empowering Families certified facilitators. The focus of the webinars was on Registration & Recruitment, Evaluation, and follow-up, respectively. The follow-up webinar also included Region 4 Midwest facilitators.

**Title:** Journey through Diagnosis guide for families  
**Description:** Family Forum began 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 is being 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.

**Affordable Care Act**

**Title:** Metabolic Foods & Formulas ad-hoc workgroup  
**Title:** Medicaid Coverage for Genetic Testing and Services
| Description: The Public Health Long-Term Follow-Up Workgroup formed an ad-hoc group to address regional issues related to the financing and access to medical foods and formula. | Description: Performed an inventory across regional Medicaid programs for current policy and coverage related to genetic testing and services. |
| Who with: MI Department of Health and Human Services | Who with: NCC Health Care Access and Financing Workgroup, Regional state Medicaid programs |
| **Accomplishments:** The in-person meeting included an interactive session where a work plan was developed. During this process, attendees assessed the group’s strengths, weaknesses, opportunities and threats. Key actions were discussed, and three action items were selected: Create a position statement on minimal safety net for medical foods and formula coverage, create a toolkit for states and families, and collect outcome data from the group as programs are implemented. A project charter has been created for the medical foods and formula project. | **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. |

| **Title:** MDHHS Diet for Life Metabolic Food and Formula Stakeholder Workgroup | |
| **Description:** Consultation, facilitation, and dissemination of information and stakeholder input regarding insurance coverage of metabolic foods and formulas. | |
| **Who with:** MI Department of Health and Human Services Genomics and Genetics Disorders Section, Children’s Special Health Care Services | |
| **Accomplishments:** Consulting with staff from MDHHS Genomics and Genetics Disorders Section to update department on feedback and input from families regarding barriers faced in accessing insurance coverage for metabolic foods and formula. Consulted with Children’s Special Health Care Services staff regarding eligibility requirements and guidelines regarding enrollment and coverage for families needing metabolic formula. Consulted on the development of a toolkit for families regarding how to navigate insurance for metabolic formula, and other options for coverage. Shared resources and lessons learned with all states in Region 4. |
| 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.” |
|---|---|
| Title: Care Coordination: Empowering Families insurance data analyses | Description: Analysis of a dataset that includes 234 training participants across the region who answered our extended items about insurance.  
Who with: Kathy Neville, Region 4 Midwest insurance advocate  
Accomplishments: Results continue to confirm that those with public only insurance (compared with private only or a blend of public and private) reporting the fewest concerns about insurance costs (co-payments and deductibles). We have explored other questions with this dataset including any trends for those reporting gaps in insurance, notable trends in coverage before and after the ACA deadline, and whether demographic factors influence coverage or concern about costs. Two of the more interesting findings that have emerged to date involve the relationship between medical complexity (as measured by the number of conditions parents report for their child) and insurance. |

**Long-Term Follow-Up**

| 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 |
<table>
<thead>
<tr>
<th>Accomplishments: 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:** Michigan Newborn Screening Mapping Project  
**Description:** Project using MI NBS data of metabolic, sickle cell disease, and congenital hypothyroidism positive screens to understand geographic distributions of these conditions. For metabolic patients, distance to provider metrics were calculated to understand how far patients need to travel for care.  
**Who with:** MI Department of Health and Human Services Newborn Screening Program, geographers from Michigan State University  
**Accomplishments:** Project resulted in a series of maps illustrating geographic distribution of patients with positive screens. Analyses included degree of clustering and spatial correlation with the total population. Mapping real instance data helps understand the
geographic realities of providing care to these patients and that access to care issues may be condition specific. This project addresses important implications for access to care for both rural and urban patients.

<table>
<thead>
<tr>
<th>Needs Assessment</th>
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<tbody>
<tr>
<td><strong>Title</strong>: Implementation of Genetic Systems Assessment Tool</td>
</tr>
<tr>
<td><strong>Description</strong>: Region 4 staff coordinated with Dr. Ann Chou and Heartland Collaborative colleagues to develop a protocol to introduce the tool at the 2015 Regional Meeting.</td>
</tr>
<tr>
<td><strong>Who with</strong>: Heartland Region, Dr. Ann Chou</td>
</tr>
<tr>
<td><strong>Accomplishments</strong>: Dr. Chou traveled to the Regional Meeting to present the tool and help facilitate state teams as they worked on discussing and completing high impact measures. Copies of the full instrument were sent to state leads to complete and review. Final data were submitted to Dr. Chou for reports. Region 4 staff received insightful and constructive feedback about the tool. It was especially interesting to hear the discussion of service provision from the perspective of multiple stakeholders (state public health officials, consumers, primary care providers).</td>
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</table>
Heartland Genetics and Newborn Screening Collaborative Regional Activities

### Intra-Regional

| Title | Care Coordination Training / Empowering Families | Training Description: In Project Year 3, the Heartland RC partnered with Region 4 Midwest RC to offer the Care Coordination Training: Empowering Families. In Project Year 4, those trained were to provide at least one training in their state by November 2015. In order to facilitate the success of their trainings, Heartland offered small grants to help cover the costs. Seven of the eleven organizations received an award and agreed to conduct another training by May 31, 2016. | 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. In the first year of the project twenty conversations with key stakeholders were completed in an effort to identify the issues surrounding access and provision of culturally competent medical genetics services. Staff at three FQHCs were part of the interview process and helped to identify families for the interviews. |
| Who With: ND, SD, NE, KS, MO, IA | Accomplishments: Eleven organizations participated in the training resulting in 24 certified facilitators. As of May 31, 2016, 135 parents have completed the in-person training and pre/post assessments. | 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. |

### Inter-Regional

| Title: Collaborative Partners Project | Description: The purpose of the Collaborative Partners Project’s was to promote NBS education throughout the region. This year the states had the flexibility to select their participants based on their ability to contribute to their respective NBS programs. The selected participants met with the Heartland NBS work group representatives at the American Public Health Laboratories (APHL) Genetic Testing and Newborn Screening Symposium held in 2014. The participants agreed to 1) to attend the APHL conference to learn more about newborn screening; 2) to provide a written statement describing their experience and how they felt it will impact their state; and 3) to complete a state project. | Title: Genetic Services Assessment (GSA) Project | Description: The Genetic Services Assessment (GSA) is a tool developed for state level public health programs to use in assessing the genetics systems/services in their respective states. The quality metrics cover five domains: (1) State capacity for services; (2) Access; (3) Clinical process and quality improvement; (4) Performance reporting/improvement; and (5) Workforce. Parallel to the continuous refinement of the GSA tool, Heartland has focused on creating cross collaborative opportunities for implementation. The tool was implemented with each of the participating regions to facilitate a continuous improvement process in their state. |
| Who with: Arkansas, Oklahoma, Nebraska, and North Dakota. | Who with: NYMAC, Mountain State, Region 4, Heartland |
### Accomplishments:
All five participants completed their action plans that resulted in dissemination of NBS information through education of colleagues; completion of a policy statement to increase NBS state infrastructure support and services; and development of a process to determine the feasibility of expanding a pulse oximetry screen on newborns in home births.

### Accomplishments:
Participation in the GSA process resulted in states completing a needs assessment that identified their strengths and needs related to genetic service delivery in their state. For some of the regions, this was the first time of administration and these results were used to frame a continuous improvement process. The information reported as aggregate and tailored for each state was used as a tool for the states to engage in further needs assessment, cost-benefit analyses, and program design and implementation to enhance access to genetic services, particularly prevention. Heartland is also using the GSA to monitor progress. Compared to the data collected from three years ago, the Heartland made great strides overall, but especially in accomplishing a number of elements that were targeted for improvement at the time of the initial assessment.

### Title: EHDI Exchange Program
**Description:** Since the EHDI coordinators have distinct roles within their states it is necessary for them to reach outside of their state borders for technical assistance and training. The Heartland network provided a natural mechanism to bring state coordinators together in order to maximize their learning from colleagues in similar roles across states.

**Who with:** Nebraska, Iowa, Kansas, Oklahoma, Arkansas, South Dakota, North Dakota, Missouri

**Accomplishments:** A targeted technical assistance approach allowed for states to identify their needs and link with states where there are innovative, best practices that they can learn from and adopt in their state. A regional approach allows for states to take advantage of states who have dealt with similar issues.

### Title: Collaborative Improvement and Innovation Network for Timeliness in Newborn Screening
**Description:** 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.
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:** Kansas, Iowa, Arkansas, Missouri, North Dakota, Oklahoma

**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. We do not yet know how many states will apply for the next round of awards due in August 2016, but several indicated that they planned to do so. All participants ranked the training as valuable or very valuable.

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**Long-Term Follow-Up**

**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. Two centers (SD and MO) were offered continued support when funding from a federal grant lapsed.

---

**Transition**

**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. To date, focus groups, interviews, a survey and a learning collaborative strategy have been used to gather baseline information. From these data, it
was determined that the needs were for tools/resources, interventions/protocols, and information/education. Moreover, the theoretical foundations of self-determination, supported employment and social capital were applied to the identified problems/potential solutions in the healthcare transition process for people with genetic conditions.  

**Who with:** In the past project year, all of the work has been conducted in South Dakota.  

**Accomplishments:**  
Tools: Transition Engagement Guide and Doc Talk  
Interventions: Transitions Clinic  
Education: Adolescent Health Rotation for Pediatric Residents

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**Telegenetics**  

**Title:** Telegenetics  
**Description:** Educated genetics residents, genetic counseling students and those in practice less than five years in partnership with Western States region.  
**Who with:** Heartland and Western States  
**Accomplishments:** Conducted two trainings involving 15 participants, which included, physicians, genetic counselors, special health care needs program, genetic counseling students, a metabolic nurse practitioner, and one director of another telehealth resource center.
### Mountain States Genetics Regional Collaborative (MSGRC) Regional Activities

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<td><strong>Title:</strong> Texas Primary Care and Home Health Summit <strong>Description:</strong> MSGRC was able to sponsor the attendance of MSGRC members (6 consumers and 1 family physician) to the summit. Surveys were distributed before attendance to gather insight on consumer perspective on medical home. <strong>Who with:</strong> MSGRC members and Marilyn Brown, Project Manager <strong>Accomplishments:</strong> Pre- and post- attendance surveys have been disseminated and are in process of being collected. This was the first year the summit had consumers in attendance and MSGRC members actively participated in discussions after presentations and workshops.</td>
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</table>

| **Title:** Parent Partner Project **Description:** 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. **Who with:** Project Lead, Brad Thompson, Hali Project; Montana Health Department, Wyoming Health Department **Accomplishments:** The Parent Partner Project is currently in eight pediatric practices and is expanding to a reservation site and air force base site, both in Wyoming. The project in Montana is now being 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. |                 |

| **Title:** State Updates Submission **Description:** Prior to the annual meeting, MSGRC’s state health department contacts are asked to complete a questionnaire addressing updates in the prior year in their state NBS program. These updates are compiled into a packet and distributed to NBS | **Title:** NCC ACT Sheet Workgroup **Description:** ACT sheets and their algorithms give quick overviews on conditions, diagnoses, and next steps for patients. **Who with:** Dr. Celia Kaye is an expert member of the panel. **Accomplishments:** As new conditions are added to the NBS panel, |
WG members at the Annual Meeting.  
**Who with:** State health partners.  
**Accomplishments:** For 2015-2016, 7 of 8 states in our region submitted a completed form. This serves as an opportunity for states to share potential resources and information.

additional materials are added. MSGRC has continued to support development and use of the ACT sheets in our region.

**Title:** Hemoglobinopathies Interest Group (HIG)  
**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.  
**Who with:** Kathryn 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.

**Title:** Participation in Advisory Committee on Heritable Disorders in Newborn Children (ACHDNC)  
**Description:** In addition to the MSGRC staff attendance of quarterly meetings. MSGRC staff Joyce Hooker (Regional Outreach) is a member of the education committee.  
**Who with:** All MSGRC leadership staff  
**Accomplishments:** MSGRC staff attended all ACHDNC meeting in Y4. 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 PKU care guidelines. Additionally, at the last meeting in April 2016, members from several state health departments brought several case studies for discussion. Dr. Mark Korkson from Genetic Metabolic Center for Education (GMCE) also attended, to educate attendees on his group’s work and purpose and gather feedback.

**Collaboration**

**Title:** In-person Meetings, Jan. 2016 Focus Groups and April 2016  
**Title:** NCC Regional Support Service Model (RSSM)
### Annual Meeting

**Description:** MSGRC held two in-person meetings this year. In January 2016, Focus Groups were held to inform our members of data collected through regional and national needs assessment and allow an opportunity for our stakeholders to participate in open forums and MSGRC Priority Setting Sessions. Outcomes were used to drive the content of the April 2016 Annual Meeting where all five workgroups (Consumer Advocacy, Emergency Preparedness, Medical Home, Telegenetics, and Newborn Screening) met.

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

**Accomplishments:** MSGRC collected information on where our future resources and focus should be directed. These outcomes were shared with our mailing list with an opportunity for feedback. Several new members were recruited to the Focus Groups. The groundwork done at the Focus Groups created a groundwork to identify new projects at our Annual Meeting. Two of these, the Cook Children’s Case Study and Extended Genetic Services Mapping Project have already completed initial planning and work.

### Workgroup

**Description:** The workgroup, chaired by Dr. Kaye, was charged with reviewing existing regional structures 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:** To date, a final brief of a proposed model has been 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 comment period.

### Newborn Screening Translational Research Network (NBSTRN)

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

**Description:** Kathryn Hassell is a member of the NBSTRN Clinical Integration Workgroup which facilitates implementation of the LPDR and integrity of tools and projects of the NBSTRN.

**Who with:** Kathryn Hassell and other workgroup members

**Accomplishments:** Dr. Hassell co-chairs the Clinical Centers WG along with Dr. Susan Berry.

### Affordable Care Act

**Title:** Affordable Care Act webpage on MSGRC website

**Description:** MSGRC developed an ACA Café in Year 3 that disseminated consumer-focused information and resources on the ACA.

**Who with:** Led by Kristi Wees with input from MSGRC members

**Accomplishments:** The content is available on the MSGRC website and is archived on our Facebook for continued reference. The ACA webpage had 55 visits in Year 4.

**Title:** MSGRC Policy Brief

**Description:** In 2014, NEGIC released an ACA Policy Brief. In 2015, MSGRC undertook an adaptation of the brief to the mountain states. The adapted brief was completed in October 2015.

**Who with:** MSGRC staff and UNI PhD student, Christine Cardinal. Meg Comeau, Catalyst Center, also acted as a consultant during the final review process.
| **Title:** Long-term Follow-up in Metabolic Consortium  
**Description:** Dr. Janet Thomas has used the Metabolic Consortium as a venue to share the work of the IBEMC with clinicians in the MSGRC region.  
**Who with:** MSGRC MC members  
**Accomplishments:** Long-term Follow-up continues to be reviewed at Annual In-person Meetings. For the April 2016 Annual Meeting, members of the Metabolic Consortium were able to recruit young adult PKU patients to attend our Annual Meeting as consumers and attend a special young adult transition session. | **Title:** ACHDNC Long-Term Follow-Up  
**Description:** Both Kathryn Hassell and Celia Kaye are members of the ACHDNC Long-Term Follow-up and Treatment Workgroup.  
**Who with:** Drs. Kathryn Hassell and Celia Kaye and other national participating members  
**Accomplishments:** Dr. Hassell has been able to attend and represent MSGRC at all In-Person ACHDNC meetings. Dr. Kaye continues to participate through webinar attendance. |
| --- | --- |
| **Title:** Metabolic Consortium Participation in IBEMC  
**Description:** MSGRC funds the participation of the Metabolic Consortium in the IBEMC. Information on long-term clinical progress on patients in our region is being collected through this funding and entered into the IBEMC.  
**Who with:** Dr. Janet Thomas and metabolic providers in region  
**Accomplishments:** To date, legacy data on patients previously consented is being obtained to enter into new system and new patients have been consented with clinical outcomes being tracked. | **Title:** Transition Discussion Forum  
**Description:** MSGRC actively recruited young adults to the 2016 MSGRC Annual Meeting. An open forum was hosted in which young adults shared their experiences and the resources that are lacking in transition. The group discussed how MSGRC could effectively serve as a resource for young adults.  
**Who with:** Discussion was led Project Director Kathryn Hassell and Consultant Celia Kaye.  
**Accomplishments:** This was the first year that MSGRC has been able to bring young adults to the annual meeting and host a special session for them. One young adult that came has continued to be active since... |
then and recently attended the Texas Primary Care and Home Health Summit as a MSGRC representative and consumer.

### Telegenetics

**Title:** Telegenetics Lunch Session  
**Description:** In lieu of the regular Telegenetics Workgroup session, MSGRC hosted a lunch speaker, Dr. Dale Alverson, who reviewed telehealth resources and the incorporation of telemedicine into genetics clinical practice. The latter half of the session served as an opportunity for MSGRC members to plan potential projects for Year 5.  
**Who with:** Speaker: Dr. Dale Alverson, immediate Past President of the American Telemedicine Association, and MSGRC TG WG member; Moderator: Dr. Celia Kaye; all MSGRC Annual Meeting attendees  
**Accomplishments:** This session has created a foundation for recently initiated MSGRC TG projects. To date, we have created the project outline for a telemedicine case study at Cook Children’s and consulted with the CO School of Public Health on expanding our previous mapping project of genetic services in the region. This would serve as a basis on what areas are in highest need of TG services.

**Title:** Recruitment of Providers for Telegenetics Training  
**Description:** Western States and Heartland have developed a curriculum for telegenetics training and is now leading training sessions in Nashville and Phoenix. MSGRC is sponsoring the attendance of clinicians in our region.  
**Who with:** MSGRC staff and members, Heartland Collaborative, Western States Collaborative  
**Accomplishments:** MSGRC currently has five clinicians that will be attending telegenetics training. MSGRC has discussed the potential of having attending clinicians guide a “mini” telegenetics training at the MSGRC 2017 Annual Meeting.

**Title:** Cook Children’s Telemedicine Case Study  
**Description:** MSGRC in collaboration with Cook Children’s is in the initial stages of a case study on implementation of telemedicine in a genetics clinic. The purpose of the study is to demonstrate outcomes of wide use of telemedicine as a basis for “convincing” geneticists to utilize telemedicine in their own clinics. The case study will look retrospectively to understand the implementation process and growth to present day at Cook Children’s. The purpose is to be informational for institutions or clinics interested in the uptake of telemedicine and demonstrate effectiveness for clinicians, patients, and payers.
**Who with:** Dr. Mary Kukolich (Geneticist) and Kim Epperson (Telemedicine Director) of Cook Children’s. Project team consultants include Dr. Dale Alverson (past ATA president), Andy Rohrwasser (UT NBS Director), Patricia Carroll (MSGRC TG Co-Chair), and Christine Barth (GC currently using telemedicine).

**Accomplishments:** Cook Children’s has agreed to serve as a site for the case study and offered to go back retrospectively to collect data for the study. A project outline has been distributed to the project workgroup.

**Title:** Genetic Services Mapping Project  
**Description:** MSGRC previously mapped the location of genetic clinics and providers in its eight state region. Practices were interviewed by phone and information collected on types of patient seen (i.e. cancer only, children, pregnant women, etc.), wait times, numbers of new patients seen. The project is now being expanded to collect additional information such as socio-economic characteristics of patients served, catchment areas, and use of telemedicine.

**Who with:** MSGRC staff and Colorado School of Public Health  
**Accomplishments:** MSGRC work has been presented at conferences (ACMG) and other national meetings (PD/PM meeting). The project was also used as a basis for a mapping project conducted in Region IV Midwest. In the project expansion, we are partnering with the Colorado School of Public Health to use more sophisticated software mapping. Additionally, we have additional questions in the practice questionnaires that will better quantify catchment areas and socioeconomic factors. MSGRC will also identify clinics that are currently using telemedicine services.

**Other**

**Title:** Hispanic Advisory Access Council  
**Description:** Qualitative interviews where held in Heartland with Hispanic families on access to genetic services. MSGRC has participated as part of their access council.  
**Who with:** Dr. Janet Thomas, Dr. Margarita Saenz, Marilyn Brown, and others recruited regionally and nationally by
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<th>Heartland</th>
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<td><strong>Accomplishments</strong>: MSGRC staff has participated in conference calls and in-person meeting. Dr. Thomas and Dr. Saenz are both filling needed roles of geneticists on the Advisory Council.</td>
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# Western States Genetic Services Collaborative (WSGSC) Regional Activities

## Intra-Regional

### 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:** 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 is still under expert review, XALD is in development.  
**Who with:** AK, CA, HI, ID, OR, WA, and Guam partners.  
**Accomplishments:** The website has 20,000 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:** Established contact with each state Title V program and will begin additional assessment meetings in June 2016. Assessment was completed in Hawaii in September 2015.

**Title:** Newborn Screening Survey  
**Description:** Parent opinion about notification preferences  
**Who with:** WSGSC region  
**Accomplishments:** survey complete, analysis underway

### 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 was recently updated and has been well received.

**Title:** ACA Parent Survey  
**Description:** Online and paper based survey of families of children with special health needs regarding essential health benefits and financing of health care.  
**Who with:** WSGSC survey was modeled after similar surveys developed by NEGC and NYMAC.  
**Accomplishments:** Administered in Hawaii.

### 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:** Community engagement activity planning and collaboration with key stakeholders are established. Project underway.

### Transition

**Title:** UNITY-GC Survey  
**Description:** Online and paper based survey of teens/young adults with genetic conditions assessing needs related to health insurance, barriers to care and frequency of specialty provider visits.
<table>
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<tr>
<th>Title: Technical assistance for telegenetics</th>
<th>Title: Telegenetics Education and Training</th>
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<tbody>
<tr>
<td>Description: Providing technical assistance for the states in our region to increase services and education provided by telegenetics.</td>
<td>Description: Two-day, in-person training sessions for genetic counselors on the use of telemedicine in clinical genetics practice.</td>
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<td><strong>Who with:</strong> Washington Genetics Program, California and Alaska NBS programs.</td>
<td><strong>Who with:</strong> Heartlands Regional Genetics Collaborative and HRSA-funded Telehealth Resource Centers in Arizona and Arkansas.</td>
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<td><strong>Accomplishments:</strong> Provided telegenetics protocols and information to help the development of telegenetics activities in the other states.</td>
<td><strong>Accomplishments:</strong> The second training session was successfully held in August 2016 and attended by genetic counselor trainees. The third training session is planned for August 2016.</td>
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SUPPLEMENT 4. NCC and NGECN Highlights

In 2015, HRSA awarded the ACMG a two-year competitive continuation renewal to continue the work of the NCC to:

- Develop a framework for regional genetic care centers that reflect the needs of diverse regions;
- Provide an infrastructure that strengthens communication and collaboration between the RCs, offers technical and clinical expertise as needed, promotes and disseminates outcomes of RC activities of national significance, facilitates partnerships with federal and non-federal entities, and evaluates the impact of RC activities; and
- Implement a NGECN that provides access to genomic information and resources that cover the lifespan for consumers and the public.

This competitive continuation provided a shift in direction, focus, and activities. As a result, NCC/NGECN data will be slightly altered from that of the rest of the RCs. The NCC focused its efforts on a national needs assessment to improve access to genetic services. Designed to collect information from consumers and providers, this effort informs the Health Resources and Services Administration’s (HRSA) development of innovative models of support centers for regional genetic and genomic services and resources that will be useful for the next 5 to 10 years.

How much did you do?

Regular RC/NCC leadership calls and NCC workgroup calls (n=71) promote common approaches to HRSA priority areas and rapid dissemination of innovative ideas among the more than 860 participants. (See NCC summary for a table depicting NCC Workgroup activities.) Staff from NewSTEPS participate on PD/PM calls; NCC staff participate on NewSTEPS workgroups/committees and other national centers. NCC staff also participate on telehealth resource network advisory councils.

The NCC/RCs met in-person with the Association of University Centers on Disabilities (AUCD) LEND Directors in November 2015 to review joint activities and to discuss additional collaborations.

The June 15, 2015 Dialogue Addressing and Paying for Genetic Services in Integrated Delivery Systems brought together geneticists, PCPs, insurers, and consumers to address access to genetic services. 82 individuals participated in-person and 80 participated remotely. The meeting summary with next steps and gaps was posted on the NCC website.

NCC leadership (PI, PM, and Medical Director) participated in and made presentations at all seven RC annual meetings. The NCC staff also presented and exhibited at the ASHG, APHL Newborn Screening and Genetic Testing Symposium, AMCHP, and ACMG annual meetings. Additionally, NCC participated in a North Dakota newborn screening meeting to share national work with a local program.

The NCC conducted 12 listening sessions with more than 250 stakeholders to develop the framework for HRSA’s new regional genetic care center models. These listening sessions engaged genetic professionals, family leaders, public health staff and others in 36 states. Sessions occurred at the NGECN Advisory Committee meeting, the ACMG annual clinical meeting and with the American Academy of Pediatrics (AAP) Committee on Genetics and the AAP Section on Genetics and Birth Defects Executive Committee, and at four RCs’ annual meetings/summits (NYMAC, NEG, Heartland, and MSGRC). Teleconference listening sessions occurred with Family Voices, Region 4 Midwest, and WSGSC stakeholders.
NCC-supported 7 genetic counseling students and 10 advocate leaders from 6 RCs who participated in the Advocate Leaders’ Mentoring Program at the ACMG Annual Clinical Genetics Meeting, to continue the successful peer-mentoring program between emerging professionals and consumers.

Over 40 people attended the NCC-sponsored Community Conversation, “Can You Hear Me Now? Empowering Relationships between Consumers and Genetics Providers through Technology” at the ACMG Annual Clinical Genetics Meeting.

How well did you do it?
During 2015-16, 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. A special workgroup and advisory committee was convened to achieve the work outlined through Goal 1: regional support service model development.

NCC/RC Advocate Leaders’ Mentoring Program, offered as a consumer—genetic counseling mentor program at the ACMG annual meeting. 100% of participants (n=13) agreed or strongly agreed (in a post-meeting evaluation) that the learning objectives were met and that the content was useful.

In the care of patients with genetic conditions, ACT Sheets are being utilized by clinicians. There were 9,107 unique visits and 10,875 page visits to ACT Sheets. Over 398 units of the ACT Sheet App were downloaded by Apple and Google Play users last year.

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). In 2016, 48 States and territories screened all newborns for CCHD and 40 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.

The Regional Support Service Model Brief contained a review of over 25 regionalized structures and a set of 8 proposed models. The brief was developed and submitted within the 10-month timeframe and has provided information to HRSA on potential options for the future of regionalized genetics programming. This brief was made available publicly and disseminated to the seven RCs in April 2016.
### NCC Workgroup 2015-2016 Activity Highlights

<table>
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<tr>
<th>NCC Workgroup</th>
<th>Mission</th>
<th>2015-2016 Activities</th>
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<tbody>
<tr>
<td>Regional Support Model Workgroup</td>
<td>Review models</td>
<td>Reviewed 25 models to inform its recommendations. RSSM WG developed a list of priorities based upon the results of the needs assessments. Regional Models Recommendation Brief was submitted to HRSA on March 31, 2016.</td>
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<td>ACT Sheets</td>
<td>Clinical resources</td>
<td>Worked on revised ACT Sheets and new Algorithms related to Lysosomal Storage Disorders.</td>
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<td>Healthcare Access and Financing</td>
<td>Improve insurance coverage</td>
<td>Held a June 15, 2015 Dialogue Addressing and Paying for Genetic Services in Integrated Delivery Systems – a meeting which brought together geneticists, PCPs, insurers, and consumers to address access to genetic services. The proceedings from the meeting were posted on the NCC website. Work continues in three subgroups focused on Identifying Genetics Services; Coverage of Genetic Services; and the Consumer Role in Genetic Services.</td>
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<td>Care Coordination</td>
<td>Promote medical home transition, and family health history</td>
<td>The white paper entitled “Sharing Exemplary Practices from Primary and Specialty Care Providers” was finalized and posted on the NCC website. Reviewed data from the 2015 Access to Care Survey; commented on the questions in the 2016 consumer survey; and conducted two webinars to determine future directions.</td>
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<td>Long-term Follow-up</td>
<td>Build public health capacity building and use of the NBS Recommended Uniform Screening Panel (RUSP)</td>
<td>14 States have joined in the pilot of the Longitudinal Pediatric Data Resource. 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 Advisory Committee on Heritable Disorders in Newborn Children (ACHDNC).</td>
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<tr>
<td>Telegenetics</td>
<td>Improve access to genetic services</td>
<td>Promoted training in the use of telemedicine for genetic service professionals.</td>
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Conducted a literature review to inform the selection of outcome measures for genetic services delivered through distance strategies.

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<th>Evaluation</th>
<th>Assess NCC/RC impact</th>
<th>Conducted listening sessions to inform the development of regional genetic care service center models.</th>
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<td>Reviewed and commented on the design and content of the consumer and provider surveys.</td>
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<td>Promoted the Genetics Services Assessment (GSA) tool.</td>
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<td>Served as a forum for sharing regional needs assessment activities.</td>
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<td>Commented on the MCH Discretionary Grant measures.</td>
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<tr>
<th>Project Directors/Managers</th>
<th>Promote inter-RC collaborations; Partnerships with other HRSA funded programs;</th>
<th>Held an NCC/AUCD LEND meeting in November.</th>
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<td>Supported an Advocate Leaders’ Mentoring Program held at ACMG’s Clinical Genetics Meeting in March.</td>
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<td>Participated in the RSSM Workgroup’s deliberations, as well as other NCC Workgroups.</td>
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This, the first year of a two-year project continuation, focused on ensuring the voices of individuals with genetic conditions and their families were heard in regional and national discussions on the future of genetic services. Working closely with families and family support groups, the NGECN developed and disseminated a national needs assessment to gather actionable data on needs related to access and quality; data used by NCC and the RSSM workgroup to recommend models for future regional/national efforts. We focused on promoting patient-centered educational materials and care practices so that families are better supported as they navigate service systems. 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
- Hosted 2 community engagement webinars (202 total attendees) with consumers on genetic services needs
- Designed and fielded a 34-question national needs assessment entitled *Understanding Access to and Quality of Genetic Services: The Individual/Family Perspective* (responses from 1355 affected individuals/parents) on access to care and support
- Conducted 18 semi-structured interviews with hospitals, advocacy networks, and community health centers on principles and best practices for patient navigation as a scalable intervention
- Presented consumer needs assessment data and field survey results at 2 national and 1 regional meeting to experts tasked with designing measurable interventions to address gaps in services
- Facilitated continuing discussions on family engagement and genetic services with 49 advocates/advocate leaders across HRSA-funded programs (RCs, LEND, Title V) in Genetic Alliance Family Advocates Network (7 conversations)
- Added 1,663 curated, patient-centered, and low literacy condition descriptions and 125 new/updated support groups to DiseaseInfoSearch.org
- Served as an advisor to Heartland’s Hispanic Access Advisory Group (2 calls, 1 in-person meeting), strategizing ways to address barriers to care experienced by Hispanic families

How well did you do it?

- Manuscript submitted and under review (as of 4/27/16) by the *Journal of Maternal and Child Health* on appropriate referral to genetic services and supports based on findings of national consumer needs assessment
- Active participation in community engagement webinars demonstrated by 120 discussion points and follow up from 25 participants
- *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
- 48,925 individuals accessed information on genetics and health, testing, and other genetic services through GenesInLife.org
• Family-friendly *Guide to Genetic Counseling* integrated into genetics training workshop for 100 nurses
• 7,999 family health history booklets were requested (100% delivered) by organizations and family members and through a national E-card campaign (3,798 total pageviews)

Was anyone better off?
• Data from consumer needs assessment prioritized by RSSM Workgroup; findings integrated into the effort as foundation for recommendations to improve services
• *Partners in Care* monograph effectively catalyzed discussions among stakeholders on the potential for standardized outcomes collection, policies, and practices in patient navigation
• 277,824 individuals (503,573 total pageviews) found accessible and clinically accurate condition information on DiseaseInfoSearch.org; 3,697 individuals accessed links to support groups
• 3 culturally appropriate, family-driven recommendations and 6 strategies for implementation devised as part of strategic plan for reducing barriers to care for Hispanic families
• 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