

HEARTLAND GENETICS & NEWBORN SCREENING COLLABORATIVE

Annual Evaluation Report 2008-2009



September 2009

The Heartland Genetics and Newborn Screening Collaborative is supported through a cooperative agreement from the Genetic Services Branch of the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA Grant U22MC03962).

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Program Description

The Heartland Genetics & Newborn Screening Collaborative is a network of representatives (parents, patients, clinicians, researchers, industry representatives, laboratians and public officials) from eight (8) states. This five-year-old Collaborative actively engages with its partners at a regional and national level to increase access to and improve quality of genetic services in the region and at the same time contribute to projects of national significance. This work is completed under the leadership of the Heartland Regional Coordinating Center (RCC) with planning and direction supported by the Heartland Advisory Board and four work groups [e.g., Newborn Screening (NBS), Clinical Services, Education, and Advocacy].

Evaluation Findings

The purpose of this report is to provide a summary of the evaluation findings regarding the Heartland Collaborative's implementation of program activities, its success in accomplishing program outcomes and performance on HRSA outcome performance measures. A multi-method approach, including qualitative and quantitative methodologies, was used to help inform the continuous improvement process. (See Appendix A for the 2008-2009 Evaluation Plan). Built into the evaluation process was the ongoing review of formative and summative data based on the Heartland Collaborative's four goal areas to inform program improvement through reflection and action planning. The results of the evaluation are summarized in the following:

Clinical Genetics Services

Goal 1: Facilitate access to quality clinical genetic services for the citizens of the states with the Heartland Collaborative

A number of activities were initiated to facilitate access to quality clinical genetic services. The outcomes of those projects are summarized in the following:

- 100% of the states contributed to the Region 4 Laboratory Performance Priority project.
- 50% of the states genetics' centers participated in Region 4's Inborn Errors of Metabolism Information System (IBEM-IS). Two of these centers have completed the IRB process and will begin submitting data during the 2009-2010 year.
- Missouri was the only state that used telehealth strategies to increase access to genetic services and NBS follow-up specialty visits. This year Missouri completed 238 visits.

Genetics Education

Goal 2: Promote genetics education regarding resources, clinical and laboratory services, genetic testing, teratogen services and facilitate integration of genetics education into curriculum of schools.

Utah MedHome Portal

Two modules, Prader-Willi syndrome and Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), were completed and posted. The module on fetal alcohol spectrum is in process. Providers have access to the modules, but they were not yet approved for continuing medical education (CME) credits. Evaluation of the modules is planned for 2009-2010.

Leadership Education in Neurodevelopmental and Related Disabilities (LEND) Project

Heartland Collaborative developed an educational strategy to partner with LEND programs in the region by providing their students access to the diagnosis modules on genetic disorders. These modules will be offered to the LEND programs as a practicum opportunity for LEND trainees during 2009-2010.

Newborn Screening Training Rated Positively by Participants [Pilot Project]

Missouri expanded their newborn screening from 6 to 28 disorders, including newborn hearing. The results of a needs assessment indicated that parents felt that their primary care physician informed them about newborn screening, but had not given them adequate information about the disorders. Similarly, the providers felt that they were not as informed as they would like to be in regards to the expanded disorders that are on the screening panel. Based on these findings, the Missouri Department of Health and Senior Services, Bureau of Genetics and Healthy Childhood sponsored an educational conference, "Newborn Screening: What Providers and Parents Need to Know", to address these needs.

Findings:

- 91 (84 nurses and physicians and 7 parents) participated in the conference.
- 90% of the participants rated the conference content as informative and interesting.
- 75% of the participants rated the conference content as useful to them to complete the work of their current position.
- Based on written comments, participants reported that they had a better understanding of newborn screening and specific disorders, an awareness of organizations and support groups available, and knew where to access available national resources.
- Lessons learned:
 - Need to continue to find strategies to recruit physicians and family members to participate in training.
 - Need to have distinct educational tracks for health providers and family members.

Infrastructure Support

Goal 3: Promote quality public health programs by establishing an infrastructure to facilitate networking, education, information sharing, assessment, policy activities, program development and evaluation within the region for the public health genetics programs.

Genetics Systems Assessment (GSA)

The GSA project was initiated to provide an assessment framework for states to review their delivery system for NBS and genetics services. The GSA will include indicators of quality genetics services including measures. During 2008-2009, GSA assessment tool was drafted and reviewed by stakeholders. A literature review was submitted to *Am J of Med Genet Part C*. . In 2009-2010, the measures will be piloted and preliminary evaluation data will be collected.

Newborn Screening Exchange

Five states (North Dakota, Kansas, South Dakota, Missouri and Arkansas) volunteered to participate in an exchange to enhance their NBS processes in their state. During 2008-2009, three states completed their exchange visits. The purpose of the exchange was to provide information to states to improve their short-term follow-up (STFU) and long-term follow-up (LTFU) programs. Evaluation of this exchange included completion of a subset of the Program Evaluation and Assessment Scheme (PEAS) (completion by two states) pre- and post-visit and information gathered during a focus group of participating state representatives.

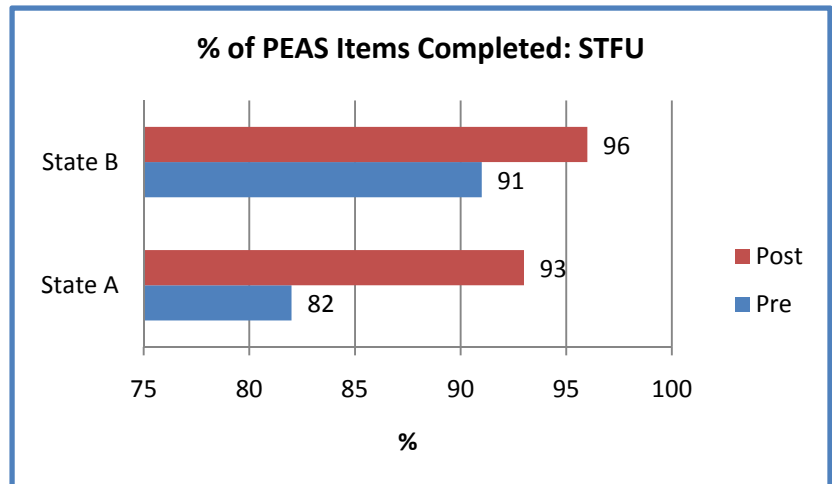
Findings:

Results of the PEAS- Subsection of STFU:

- The results of the pre-post PEAS assessments from two states found an overall increase in STFU procedural steps (based on the pre-post assessments using the PEAS).
- On average, states made an 8% increase in procedures as a result of the exchange.

Results of Focus Group:

- Information on follow-up procedures and materials helped states make modifications to their processes (e.g., developed web-site with resources for providers and families).
- Opportunities to network were reported as very valuable.
- Benefits were noted for both the sending and receiving states as they exchanged materials and ideas.



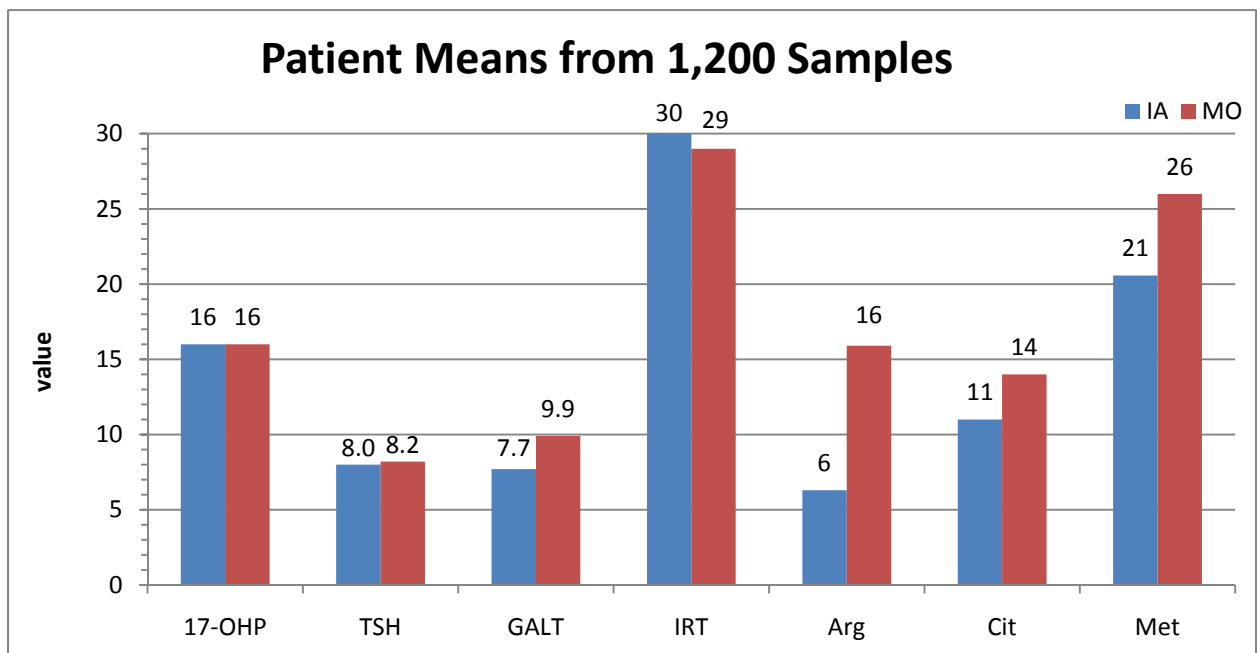
NBS Back-Up Testing and Quality Assurance Project

The Heartland NBS Back-up Testing and Quality Assurance Project, a collaborative project of University of Iowa Hygienic Laboratory and Missouri Public Health Laboratory, was established in response to a regional and national need for emergency preparedness for NBS services. The nature of the disorders and the requirement for time critical interventions make it essential that adequate and timely contingency backup services be available to prevent any interruption of routine NBS services.

The goal for the first year of this ongoing project (2007-2008) was to plan, establish and verify back-up NBS testing capabilities between the states of Iowa and Missouri by conducting two separate one-day emergency drills utilizing the federally authorized Emergency Management Assistance Compact (EMAC). The EMAC process imparts authorization and liability protections to serve as both the structure for providing emergency backup NBS services as well as when conducting functional emergency preparedness drills between the two state NBS programs. The project goal for 2008-2009 was to challenge the system by extending the duration of the drills to three consecutive days to more closely replicate a real case scenario. The evaluation of the project included information collected during interviews of the project directors.

Findings:

- The project successfully demonstrated the ability to provide NBS emergency backup services for the collaborating laboratory.
- EMAC process was successfully utilized to provide a simple, reliable and legal structure under which to conduct the drills and to ultimately count on for addressing a true emergency.
- The drills provided the opportunity to strengthen emergency procedures within the partnering laboratory agencies and resulted in updating and fine tuning some departmental procedures.
- The back-up drill provided an opportunity to compare pre-analytical, analytical and post analytical procedures and findings. The results found that MO and IA's testing results have compared amazingly well in both last year's and this year's drills.



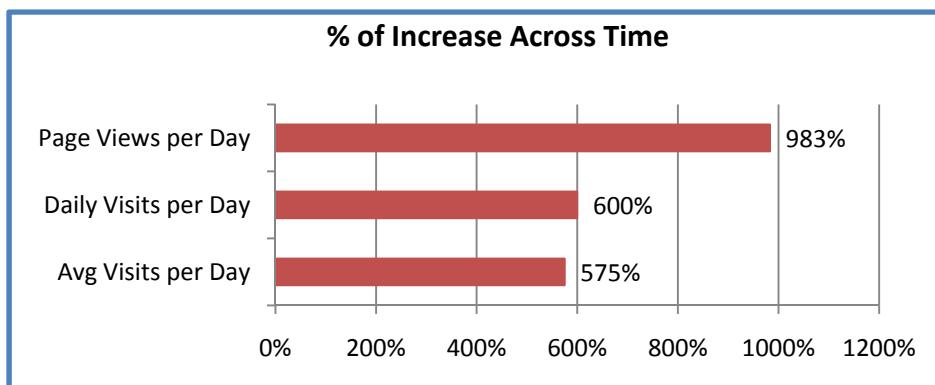
- Completing the backup plan drill on an annual basis provided the opportunity for new staff to gain experience in the process and build confidence in their ability to effectively carry out the additional responsibilities.
- Emergency testing by another laboratory also presented the challenge of ensuring that the babies born and tested during the emergency were able to be brought back into the state’s information system so there is not a “black hole” in the state NBS system following the emergency.
- Lessons Learned:
 - Need to find resources to complete an annual backup drill.
 - Prolonged backup would require additional staff.
 - It is recommended that backup labs be matched based on comparable size.

Sarah Lawrence Scholarship Program

Each year, one scholarship is given to support an individual in the Heartland region to participate in the Sarah Lawrence Public Health Genomics program. This strategy meets the Heartland Collaborative goal to build a cadre of leaders/decision-makers who are educated in public health genomics and who are positioned to use their new knowledge to effect change in their states. To date two individuals have completed the course and a third is in the process of completing her project. Each provider completes a Capstone project[e.g. in 2008 the capstone project was a collaborative work with the Centers for Disease Control and Prevention (CDC) on mortality trends in a single gene disorder, tuberous sclerosis complex (TSC)]. The impact of participation in the program will be evaluated in the 2009-2010.

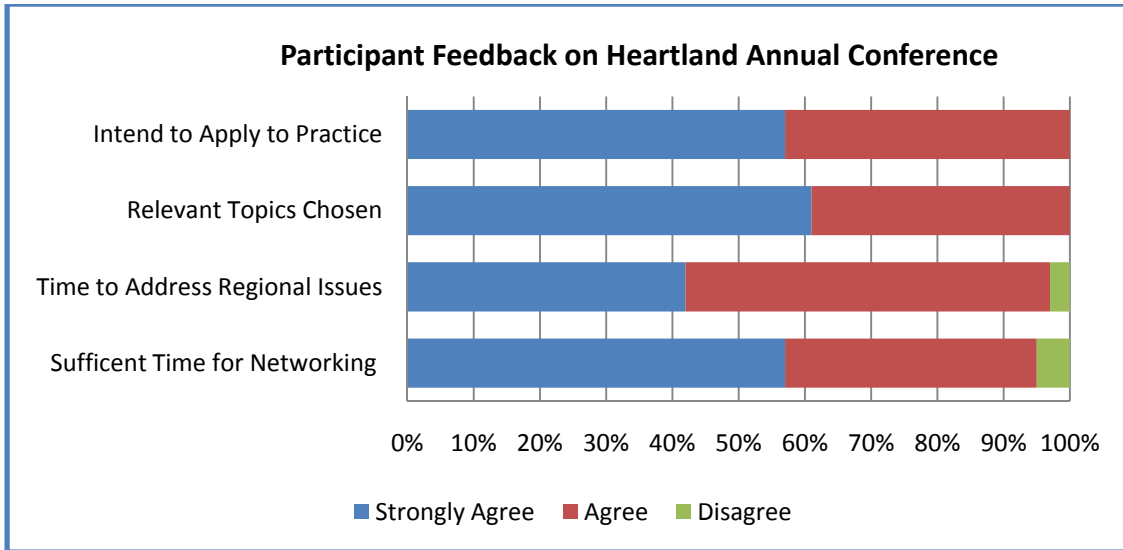
Dissemination of Information and Resources

The Heartland Collaborative used a variety of strategies to disseminate information including an E-newsletter, list serve and web-site. The Heartland Collaborative completed a major overhaul of its website. The goal was to increase usage of the two sites (www.heartlandcollaborative.org and www.heartlandfamilyhistory.org). An annual conference is sponsored each year to allow for educational opportunities and networking for participants. In addition, Heartland partners presented at national meetings.



Findings:

- Tracking of web-based usage over a two year period found that there was a significant increase of usage once the new sites were launched.
- Three presentations and three posters were presented at three national meetings.
- The Heartland Regional Conference was rated positively. Comments suggested that the networking was valuable and participants appreciated different learning formats, e.g., posters, sessions, work group meetings. Suggested improvements including extending the time of the conference and the having CEUs available for participants.



Leadership Strategies

Goal 4: Provide leadership in the region and facilitate activities to promote access to quality clinical genetic services, genetic testing, collaborative research, and public health programs for the citizens of the states in the Heartland Collaborative.

The Heartland Regional Coordinating Center (RCC) provides the leadership for the Heartland Collaborative. They have set up a structure and implemented a number of strategies to maximize meaningful participation from all stakeholders. The following activities were implemented as part of the leadership activities.

- The Heartland Regional Strategic Plan was reviewed and modified based on stakeholder feedback in 2008-2009.
- As outlined under Goal 3, the RCC has implemented multiple strategies to communicate with Heartland Collaborative stakeholders, e.g., website, work group meetings, annual conference and e-newsletters.
- Evaluation on the extent of Advisory Board collaboration is planned for in the fall of 2009.

Summary of National Performance Measures: Comparison with National Data

The national evaluation was designed to (1) measure the progress being made by the RCs toward the major goals of the initiative and (2) identify areas in which collaboration among and technical assistance from RCs, the National Collaborative Center and HRSA could be helpful in moving the goals of the initiative forward. The following summary represents ten (10) common outcome measures across seven evaluation domains. Data presented below covers the period of December 12, 2007 through November 30, 2008. The following compares the results of Region 5 data on the performance measures compared to the national average across RCs.

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED CARE COORDINATION		
Measure A1: Increase % of states with collaborations facilitated by Regional Collaborative (RC) between PCPs and specialty providers to improve care coordination.	100 % of states	48% of states and territories

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED ACCESS		
Measure B1: Increase # of genetic services visits and NBS follow-up specialty visits through distance strategies.	238 visits	502 visits

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED EMERGENCY PREPAREDNESS/CONTINGENCY PLANNING		
Measure C1: Increase % that have received current materials or assistance from the RC on emergency preparedness/contingency planning for NBS and genetic services.	100% of states	48% of states and territories

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED FOLLOW-UP OF CHILDREN IDENTIFIED WITH HERITABLE DISORDERS THROUGH NBS		
Measure D1: Increase % of states that made recommendations on implementing the ACHDGDNC recommended NBS panel.	100%	98% of states and territories
Measure E1*: Increase % of states with systems in place to track entry into clinical management for newborns who are diagnosed with conditions mandated by their State-sponsored newborn blood spot screening programs.	100%	93%
Measure E2: Increase % of states with systems in place to track entry into clinical management for newborns who are diagnosed with hearing loss.	100%	65% of state and territories
Measure E3*: Increase of system to track receipt of clinical services and/or health outcomes for children who are diagnosed with condition(s) by State-sponsored newborn blood spot screening program.	25%	15% of state and territories

*Performance Measures Reported Nationally by State

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED EDUCATION OF PCPS ABOUT TREATMENT OF PEOPLE WITH HERITABLE DISORDERS		
Measure F1*: Increase the NBS programs that disseminate “just-in-time/point-of care” information on specific heritable disorders to primary care providers.	87.5%	88% of the states and territories

Outcome Measure	Region 5 Results	National Results
DOMAIN: IMPROVED REGIONAL PLANNING AROUND DELIVERY OF GENETIC SERVICES WITH HERITABLE DISORDERS		
Measure G1: Increase the % of Regional Collaboratives (RCs) that have completed a regional genetic services plan.	100%	86% of RCs
Measure G2: Increase in the % of RCs that have reviewed and/or updated their regional genetics services plan at least every two years.	100%	57% of RCs

*Performance Measures Reported Nationally by State

Summary

The Heartland Genetics and Newborn Screening Collaborative actively engaged partners to increase access to and improve the quality of genetic services in the region. The work was completed under the leadership of the RCC and the Heartland Advisory Board. The Heartland Collaborative invested in training activities and technical assistance to build the capacity of health care providers, public health professionals and family members. Research continued to be a high priority as states collaborated in regional and national research projects. System capacity building was addressed through a number of projects, ranging from piloting innovative practices to building a quality indicator assessment system. Overall, the results indicated that the Heartland Collaborative has implemented a wide array of activities that successfully addressed its overall project goals.

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APPENDIX A: Heartland Genetics and Newborn Screening Collaborative Evaluation Plan
2008-2009

Goals	Strategies	Implementation Evaluation Measures	Outcome Evaluation Questions	Outcome Evaluation Measures	Data Collection Timeline
Clinical Genetic Services 1) Facilitate access to quality clinical genetic services	<ul style="list-style-type: none"> Collaborate with Region 4 on Tandem Mass Spectrometry (MS)research project (Lab Performance Project) 	<ul style="list-style-type: none"> Document # of states who contribute data to the project 			<ul style="list-style-type: none"> Ongoing
	<ul style="list-style-type: none"> Collaborate with Region 4 on research on clinical long-term follow-up of children with inborn errors of metabolism Promote use of telehealth strategies for clinical genetics 	<ul style="list-style-type: none"> Document participation in the study and # of data entries reimbursed Document states use of telehealth clinical genetics. 			<ul style="list-style-type: none"> Ongoing
Genetics Education 2)Promote genetics education	<ul style="list-style-type: none"> Partner with Utah Medical to strengthen medical home through education of primary care providers through web-based modules Establish CME and assessment requirements and identify demographic data for future evaluation analyses 	<ul style="list-style-type: none"> Document completion of 7 diagnosis modules Document # of providers that complete modules Describe characteristics of providers that completed the modules 	<ul style="list-style-type: none"> To what extent do the providers find the content and format of the modules helpful? What percentage of providers who complete the knowledge assessment meet the set criteria? 	<ul style="list-style-type: none"> Survey tool completed by providers who complete the modules. Completion of module knowledge assessments. 	<ul style="list-style-type: none"> Beginning Year 4 Beginning Year 4
	<ul style="list-style-type: none"> Identify strategies to promote 6 regional LEND program’s awareness of Heartland & NBSC Partner with LENDs to evaluate the potential use of Diagnosis Modules as a training option for Lend Trainees Investigate potential of presenting at Midwest LEND meeting in Fall 2009 	<ul style="list-style-type: none"> Document awareness strategies to promote HGNBSC with LEND network Document faculty and students who participated in collaborative project to determine usefulness of modules 	<ul style="list-style-type: none"> To what extent is the partnership between HGNBSC and Lend programs strengthened? To what extent are the diagnosis modules a useful educational tool for Lend students? 	<ul style="list-style-type: none"> Interviews of key stakeholders from both programs Completion of module ratings by LEND faculty and students 	<ul style="list-style-type: none"> Year 3 Year 3
	<ul style="list-style-type: none"> Pilot electronic child profile with PCP 	<ul style="list-style-type: none"> Document # of PCPs who participate in the pilot 	<ul style="list-style-type: none"> To what extent do PCPs find the electronic profile useful alternative to standard documentation strategies? 	<ul style="list-style-type: none"> Interviews of PCPs 	<ul style="list-style-type: none"> Year 3 & 4
Infrastructure Support 3) Infrastructure to facilitate networking	<ul style="list-style-type: none"> Genetics System Assessment (GSA) developed and piloted in two states 	<ul style="list-style-type: none"> Expert panel meets & modifications made to GSA GSA is established based on scientific approach 	<ul style="list-style-type: none"> To what extent was the GSA Report Card a helpful approach to assessing a quality system? To what extent do states in the 	<ul style="list-style-type: none"> Survey tool completed by pilot sites 	<ul style="list-style-type: none"> Year 3

Goals	Strategies	Implementation Evaluation Measures	Outcome Evaluation Questions	Outcome Evaluation Measures	Data Collection Timeline
Infrastructure Support Infrastructure to facilitate networking	<ul style="list-style-type: none"> GSA implemented in 8 states to determine the quality of their genetics service system 	<ul style="list-style-type: none"> Genetics Report is modified based on feedback from pilot 8 states complete GSA 	Heartland Collaborative have quality systems of care?	<ul style="list-style-type: none"> Completion of Genetics Report Card in 8 states 	<ul style="list-style-type: none"> Years 4 and 5
	<ul style="list-style-type: none"> Public health agencies identify a partner for a NBS program exchange and implement a site visit 	<ul style="list-style-type: none"> Agenda for the site visit is submitted and approved by Heartland Site visit is completed 	<ul style="list-style-type: none"> To what extent did the site visit contribute to adoption of or modification of a clinical practice or system procedure? 	<ul style="list-style-type: none"> Pre-post analysis of PEAS (post test at follow-up) 	<ul style="list-style-type: none"> Year 2 & 3
	<ul style="list-style-type: none"> EMAC project complete drills with 2-3 states, implementing back-up system Implement one NBS workshop per year related to EMAC process with network Establish links with FEMA and HSEEP regarding EMAC 	<ul style="list-style-type: none"> Document completion of EMAC drill Document protocol and process of the EMAC procedures in a manuscript Document the # of states that could implement EMAC protocol Document contacts with FEMA and HSEEP 	<ul style="list-style-type: none"> How effective was the implementation of the EMAC back-up system? 	<ul style="list-style-type: none"> Data from the EMAC sites with comparative analysis of results from two sites. 	<ul style="list-style-type: none"> Year 3
	<ul style="list-style-type: none"> Heartland provides resources on website related to youth transition Heartland sponsors speakers to increase network knowledge on youth transition Heartland links state participants who are interested in addressing youth transition issues 	<ul style="list-style-type: none"> Document # of posted resources on website Document # of awareness activities, including # of states and participants impacted 			<ul style="list-style-type: none"> Year 3, 4, and 5
	<ul style="list-style-type: none"> Sarah Lawrence Scholarship is granted for one student per year to obtain a Public Health Genomics Certificate 	<ul style="list-style-type: none"> Document selection of scholarship recipient and completion of course Document presentation of Capstone project at annual conference 	<ul style="list-style-type: none"> To what extent did completion of the certificate help the participant contribute to the field? 	Interview with participants at course completion and one year post course completion	<ul style="list-style-type: none"> Year 3, 4, and 5
Leadership Strategies 4) Provide leadership to facilitate activities to promote access to quality clinical genetic programs.	<ul style="list-style-type: none"> Revise and submit strategic plan Disseminate educational resources using multiple strategies (i.e., website, e-newsletter, list serves, meetings, conferences) Link projects in network based on individual requests for TA Contact PCA/PCO in Heartland Region to determine interest in being added to newsletter mailing list 	<ul style="list-style-type: none"> Document submission of plan Document type and # of participants per strategy Post minutes of meetings Document conference schedules 	<ul style="list-style-type: none"> To what extent do the leadership activities reflect the priorities and needs of the Heartland Collaborative? What are the contributing and inhibitory factors the influence the success of the Heartland Collaborative? To what extent have the Heartland Core Advisory group formed a collaborative partnership? 	<ul style="list-style-type: none"> Focus group Satisfaction survey on group meetings Measure of collaborations (i.e., Working Together, Tent exercise) 	<ul style="list-style-type: none"> Year 3,4,and 5

Domain	Outcome	Data Measure	Related Heartland Goal
A. Improved care coordination for people with heritable disorders.	Increase in the percentage of states in the region with collaborations facilitated by the regional collaborative between PCPs and specialty providers to improve care coordination for people with heritable disorders.	<ul style="list-style-type: none"> # of States in the region # of states with collaborations facilitated by regional collaborative between PCPs and specialty 	<ul style="list-style-type: none"> Goal 1: Clinical Service
B. Improved access to genetic services for people with heritable disorders.	Increase in the # of genetic services visits provided to people with or at risk for heritable disorders through distance strategies implemented by the regional collaborative.	<ul style="list-style-type: none"> # of genetic service visits provided to people with or at risk for heritable disorders Types of distance strategies employed 	<ul style="list-style-type: none"> Goal 1: Clinical Service
C. Development of regional emergency backup systems for NBS and genetic services.	Increase the % of states in the region that have received current materials or other assistance from the RFC on developing back-up systems for NBS and genetic services.	<ul style="list-style-type: none"> # of state in region that received current materials or other assistance from the RC on developing back-up systems for NBS and genetic services List materials or assistance provided 	<ul style="list-style-type: none"> Goal 3: Infrastructure
D. Implementation of expanded NBS.	Increase in the % of states in the region that have evaluated and made recommendations on implementing the ACHDGNC (Advisory Committee on Heritable Disorders on Newborns and Children).	<ul style="list-style-type: none"> # of states that evaluated and made recommendation on implementing the ACHDNC recommended NBS panel List of states that implemented recommendations 	<ul style="list-style-type: none"> Goal 3: Infrastructure
E. Improved follow-up of children identified with heritable disorders through NBS.	Increased in % of state with systems in place to track entry into clinical management for newborns who are diagnosed with <u>conditions mandated</u> by their State sponsored NBS.	<ul style="list-style-type: none"> # of states with systems in place to track newborns who are diagnosed with conditions mandated by their State 	<ul style="list-style-type: none"> Goal 3: Infrastructure
E2.	Increase the % children who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening program and/or hearing loss through their State-sponsored newborn hearing screening programs.	<ul style="list-style-type: none"> # of states with systems in place to track receipt of clinical services and outcomes. 	<ul style="list-style-type: none"> Goal 1: Clinical Service
E3.	Increase of states with a system to track receipt of clinical services and/or health outcomes for children who are diagnosed with condition(s) by State-sponsored newborn blood spot screening program.	<ul style="list-style-type: none"> # of states with system in place to track clinical services and outcomes 	<ul style="list-style-type: none"> Goal 1: Clinical Service
F1. Improved/expanded education of PCPs about treatment of people with heritable disorders and about clinical genetic resources in region	Increase in the % of states in region whose NBS programs disseminate "just-in-time/point-of-care" information on specific heritable disorders to PCPS	<ul style="list-style-type: none"> # of states whose NBS disseminate information to PCPs 	<ul style="list-style-type: none"> Goal 2: Promote genetics education
G1. Improved regional planning around delivery of genetic services to people with heritable disorders.	Completion of regional genetic services plan	<ul style="list-style-type: none"> Document if regional genetic services plan is completed 	<ul style="list-style-type: none"> Goal 4: Leadership strategies