

GENETIC SYSTEMS ASSESSMENT

HEARTLAND GENETICS SERVICES COLLABORATIVE
ANNUAL CONFERENCE

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The Project

- The Heartland Collaborative undertook the effort to develop The Genetic System Assessment (GSA) tool to comprehensively describe the state of quality services in genetics.
- A list of metrics has been developed, pilot-tested, and implemented in the Heartland region. The tool was further refined based on the implementation results and feedback from the process evaluation.
- The GSA tool has been disseminated in two other regions during the current fiscal year.

The GSA Tool

The GSA tool has 16 metrics, most of which have multiple elements, that are classified into 5 domains:

- Service capacity- 5
- Access to genetic services- 3
- Clinical processes and quality improvement- 2
- Performance reporting- 4
- Workforce-2

Nature/Type of Measures

The measures can be characterized as:

- (1) Basic blocks, where every quality genetics program should have;
- (2) Aspiring measures, where the program would like to be in the near future.

Element 3: Quality improvement**8 points**

Genetic services, including those contracted by the State, are integrated into public health programs or health systems through the life course

Yes

No

1. Laboratories associated with the genetic unit participate in proficiency testing.

2. State ensures adherence to licensing requirements, published guidelines, standards, and regulations.

3. State specifies performance outcome measures for the following populations, in addition to Title V measures:

a. Prenatal

b. Newborn

c. Children with special needs

d. Adult

Scoring

100%	75%	50%	25%	0%
The State responds YES to 1, 2, 3a-d	The State earns 6 points	The State earns 4 points	The State earns 2 points	The State responds NO to all

Explanation

Factor 1. Laboratories associated with the genetic unit participate in proficiency testing such as CLIA and CAP certification. A "Yes" response earns 2 points.

Factor 2: The State has a mechanism to ensure adherence of licensing requirements of appropriate providers (e.g., physicians and genetic counselors), guidelines, standards, and regulations. A "Yes" response earns 2 points.

Factor 3: The State has outcome measures for prenatal, newborn, children with special needs, and adults in addition to Title V measures, such as Health People 2020 indicators. Title V measures are those covering newborn screening and follow up services. The guidelines can be found on the following document: http://www.idph.State.ia.us/hpcdp/common/pdf/titlev/FY13_national_measures.pdf

A "Yes" response earns 1 point each for a-d.

Documentation

For all factors, the State provides written policies or procedures to support achievement of the items.

Version 2.0

- To recap: the GSA tool was completed in the Heartland Region and the tool was refined. “Must pass” metrics were identified.
- Informed by the implementation results, scoring scheme, definitions for each factor of the measure, and documentation supporting achievement of the measures have been completed.
- Additional changes based on update from the literature: Adding critical congenital heart disease (CCHD) screening for a couple of measures.

Early Screening & Diagnosis: Prenatal Services (4)

State _____ the following program:	
Maternal serum alpha-fetoprotein & associate marker screening	Same
Maternal infections	Noninvasive prenatal testing
Maternal disease	Maternal fetal medicine
Carrier status	Carrier status screening
Fetal ultrasonography	Family health history
AMA	Genetic counseling by trained professionals
Family history	

IDENTIFYING HIGH IMPACT MEASURES

Process

- During a progress update call with HRSA, the Heartland group was tasked with identifying a number of “high impact” measures from the GSA tool.
- After an internal discussion, 4 measures were proposed for initial consideration. We then consulted with the Heartland leadership and select members of the GSA Expert Panel for recommendations.
- After another round of discussion, we propose 3 measures as “high impact” and 3 additional measures as those with potential for high impact. These measures, if achieved, should provide a “landscape” of current quality improvement efforts in genetic services among states.

“High Impact” Measures

- The leadership and experts agreed that **Measures 4, 6, and 8**, if achieved, may be high impact:
 - 4=Early screening and diagnosis
 - 6=State’s provision of access to genetic services
 - 8=Accessibility/availability to genetic services
- They may lend insights into the type of services that are available within a state and the extent of access to genetic services.

Potentially “High Impact” Measures

- 3 additional measures were recommended for further consideration, taking into account feasibility of data collection and probability for achieving those elements since resources vary significantly across regions and states.
 - Looking from a traditional Title V public health perspective, **Measure 2B-information referral and coordination supported by the state**, may demonstrate great impact if public health can institutionalize provision of information and coordination for families with or at risk for genetic disorders.

Potentially “High Impact” Measures

- Similarly, **Measure 5**, institutionalizing family input in developing and implementing policies, procedures, services, assessments, etc., could have a great impact. This is especially true when families help support legislation and funding for the public health programs to expand genetic services and activities.
- **Measure 7-Access to Genetic Professionals** was proposed to be included as a complement to Measure 8. **Measure 7** asks about access to genetics professionals; **Measure 8** goes much beyond, asking if states actually have policies about scheduling appointments, same day visits, etc. **Measure 8** may be fairly intrusive in states that do not provide much or any financial support for services for some conditions, while **Measure 7** would provide a measure of access to professionals that all states should have.

COMPARISON OF IMPLEMENTATION FINDINGS

Dissemination in Other Regions

- Collaboratives from two regions were selected based on some of their similarities to those of Heartland: Mountain States and NYMAC
- They were approached and agreed to participate.
- Data collection has taken about three months, with more data coming in.
- Response rate for one region is estimated to be 63% while the other is projected to be 100%.

Information Referral & Coordination (2b)

The State supports the following:	Count	
Educational and other special services for individuals or families with special needs	7/8	9/10
Management of genetic disorders/coordination of care with medical home	6/8	7/10
Referral of families to support groups; or facilitation of contact with similarly affected families	6/8	8/10
System for direct referral from clinical genetics to early intervention services for infants < 3 years of age	4/8	9/10

Early Screening & Diagnosis: Prenatal Services (4)

State _____ the following program:	Count			
	Has		Supports	
Maternal serum alpha-fetoprotein & associate marker screening	8/8	10/10	1/8	2/10

Early Screening & Diagnosis: Newborn screening

	Count			
State _____ the following program:	Has		Supports	
Newborn blood spot screening				
% of infants screened for all state-mandated screenings	8/8	9/10	7/8	7/10
% of screened positive infants who receive timely follow-up to definitive diagnostic & clinical management for conditions mandated by state sponsored newborn screening programs	8/8	10/10	6/8	8/10
Newborn hearing screening				
% of infants screened by one month of age	7/8	9/10	6/8	9/10
% of infants who do not pass the final hearing screening and receive an audiologic evaluation by 3 months of age	8/8	9/10	6/8	8/10

Early Screening & Diagnosis: Child screening

	Count			
State _____ the following program:	Has		Supports	
Developmental disabilities	8/8	9/10	7/8	7/10
Sensory deficits	7/8	6/10	6/8	3/10
Other disorders	7/8	1/10	5/8	0/10

Early Screening & Diagnosis: Adulthood screening

State _____ the following program:	Count			
	Has		Supports	
Pre-symptomatic testing	6/8	8/10	3/8	3/10
Carrier testing/screening for neurodegenerative diseases in selected populations	5/8	9/10	2/8	2/10
Cancer susceptibility	6/8	8/10	3/8	1/10
Aging-related diseases	6/8	8/10	2/8	1/10

Access (6)

State	Count	
Provides access or has mechanisms in place to facilitate access to genetic services.	8/8	7/10
Provides the following to ensure access:		
• Funding	6/8	7/10
• Personnel	5/8	4/10
• Information	7/8	5/10
• Procedure/policies	6/8	5/10

Access to Staffing (7)

	Count	
State employs or has access to the following professionals:		
MD/PhD Clinical geneticist	8/8	9/10
Sub-specialists relevant to conditions screened	8/8	9/10
Genetic counselor/nurse	8/8	9/10
Cytogeneticist	5/8	7/10
Clinical biochemical geneticist	6/8	8/10
Clinical molecular geneticist	5/8	7/10
Advance practice nurse in genetics	6/8	5/10
Perinatologist/obstetrician boarded in genetics	4/8	5/10
Dietician/nutritionist	8/8	9/10
Audiologist	8/8	9/10

Accessibility/Availability of Services (8)

	Count	
Practice has established processes, standards, policies on	NBS	
Triaging how soon a patient needs to be seen	5/6	7/10
Scheduling appointments	6/6	4/10
Providing same day visits or urgent care	5/6	4/10
Standardizing and tracking maximum time to appointment	5/6	5/10
Providing telephone advice to families or other physicians	6/6	9/10
Consulting via secure e-mail with physician	5/6	4/10
Providing consultation via telemedicine to families	5/6	3/10
Providing outreach clinics/services	5/6	6/10
Providing follow-up services for patients and families	6/6	6/10
Coordinating consultation/visits and referrals with other providers	6/6	6/10

Accessibility/Availability of Services (8)

	Count	
Practice has established processes, standards, policies on	Contracted	
Triaging how soon a patient needs to be seen	5/6	4/10
Scheduling appointments	6/6	4/10
Providing same day visits or urgent care	4/6	3/10
Standardizing and tracking maximum time to appointment	4/6	2/10
Providing telephone advice to families or other physicians	6/6	5/10
Consulting via secure e-mail with physician	4/6	2/10
Providing consultation via telemedicine to families	4/6	3/10
Providing outreach clinics/services	5/6	5/10
Providing follow-up services for patients and families	4/6	4/10
Coordinating consultation/visits and referrals with other providers	4/6	6/10

Summary of Findings

- Majority of the states met the standards in GSA- all 3 regions are similar in their achievement of the metrics.
- Differences were found in primarily in state support of screening and diagnosis across various areas (e.g., prenatal, childhood, adult) and accessibility/availability to genetic services- with Heartland reporting higher performance in these areas.

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THANK YOU

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