

Region 4 Genetics Collaborative-EDHI WG

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Region 4 Genetics Collaborative

- Sally J. Hiner

Region 4 Genetics Collaborative
MPHI - Systems Reform



Region 4

Funded by HRSA/MCHB Cooperative Agreement
U22MC03963 *2004-

Region 4 Genetics Collaborative

Vision

All newborns will receive state-of-the-art newborn screening and follow-up; children and youth with heritable disorders will have access to genetic expertise and coordinated care in the context of a medical home.



Region 4 Genetics Collaborative

Mission

- 1) Increase access to information about newborn screening and genetic resources, services and family support systems
- 2) Facilitate data collection and analysis to guide decision-making regarding screening cut-offs, diagnosis and long term treatment of heritable disorders
- 3) Support state public health agencies in improving infrastructure for genetic service delivery to children with heritable disorders
- 4) Provide a forum for families, public health, and clinical providers to share best practices and models for improving newborn screening, follow-up and genetic care coordination
- 5) Link Region 4 states with regional and national initiatives for improving the quality of newborn screening and genetic service delivery
→ Several established workgroups

Region 4 Genetics Collaborative Base Funding Logic Model

Effective follow-up for children with heritable disorders provides “just in time information” to the child’s family and medical home. This includes children identified through newborn screening, **early hearing detection (EHDI)**, birth defect registries, CSHCN, and early intervention

Expand Follow-up Workgroup to include: **EHDI**, birth defects registry, early intervention

Develop and disseminate follow-up protocol intervention programs.

Goal 2

Improve follow-up practices for children with heritable disorders who are **identified through**: NBS, **EHDI**, birth defect registries, early intervention

Region 4 Logic Model

Assumption

Effective follow-up for children with heritable disorders provides “just in time information” to the child’s family and medical home. This includes children identified through newborn screening, early hearing detection, birth defect registries, CSHCN, and early intervention programs.

Goal 2

Develop & distribute follow-up protocols for population-based identification of children with heritable disorders

- Expand Follow-up Workgroup to include: EHDI, birth defects registry, early intervention
- Develop and disseminate follow-up protocols

Outcome

Improve follow-up practices for children with heritable disorders who are identified through: NBS, EHDI, birth defect registries, early intervention

Follow-up Workgroup

The Follow-Up Workgroup will focus on protocols and practices for providing information to primary care providers and families at the time of identification (positive screen or report) and improving communication practices between those identifying children with a heritable disorder and the primary care physician.

Workgroup History

- Partnership of stakeholders
- Existence for one year (March, 2008)
- Focus: **Genetics & Short-term follow-up**
- Several workgroup activities to
 - Identify issues
 - Identify promising practices
 - Develop Tools
 - Formulate recommendations
- Make workgroup infrastructure available for opportunities with regional implications

Members Region 4 EHDI

- Barb DeLuka, Illinois Dept of Health, NBS follow-up
- Gail Tanner, Illinois Dept. of Health, EHDI
- Ginger Mullin, Illinois Newborn Hearing Program
- Molly Pope Indiana State Dept of Health
Genomics and Newborn Screening
- Bob Bowman, Indiana State Dept of Health
- Kelly Daniel, Kentucky State Dept of Health
- Karen Mercer, Kentucky State Dept of Health

R4 EHDI WG Members (con't)

- Joan Ehrhardt, Michigan Dept Community Health
- Michelle Garcia, Michigan Dept of Community Health
- Yaoli Li State of MN Dept. of Health
- Allison La Pointe Minnesota Department of Health
Community & Family Health, Newborn and Child
Follow-up Unit, Data/Epi Unit
- Amy Gaviglio, MN Dept of Health, NBS Lab
- Barb Dalbec, MN Dept. of Health
- Nicole Brown, MN Dept of Health

R4 EHDI WG Members (con't)

- Kirsten Coverstone MDH, Public Health Laboratory
Newborn Screening Program
- Reena Kothari The Ohio Department of Health
Infant Hearing Program
- Anna Starr Ohio Department of Health
- Shelley Nottingham, Ohio Department of Health-Genetics
Program
- Ravi Shah Wisconsin Sound Beginnings
Health Services
- Gary Hoffman, Wisconsin State Lab
- Region 4 Coordinator – Sally J. Hiner

EHDI Follow Up Workgroup

Goal:

Develop and distribute best practice follow-up protocols/practice models for population-based identification of children with heritable disorders using the state supported and established EHDI program

EHDI STFU WG Outcome:

- Improve follow-up for children with heritable disorders who are identified through
 - NBS
 - UNHS, EHDI
 -birth defects registries, children with special health care needs, early intervention....

EHDI STFU WG Objectives:

- Determine needs in Region 4 and identify strategies to meet those needs
 - Collect and compare existing follow up protocols
 - Identify issues
 - Identify promising practices/strategies to address issues
 - Develop model practices and/or recommend promising practices for adaptation/use in Region 4

Accomplishments

- Review of data collection protocols for Region 4 states
- Collection and review of Region 4 State EHDI follow-up protocols/guidelines
- Review of existing state infrastructure – how does EHDI fit with NBS? Genetics?
- Survey of workgroup members to identify issues
- Categorization & Prioritization of EHDI issues

- Face to face meeting in Chicago for long term follow-up issues (opportunity)
- Identified areas of need for Region 4

Categorizing issues

- Identified 43 individual issues
- Categorized the issues into 10 categories
 - Data Collection (8 issues)
 - Data Accuracy (3 Issues)
 - Referrals (5 issues)
 - Later identified/onset (4 issues)
 - Education (6 issues)
 - Border babies (4 issues)
 - Family Support (1 issue)
 - Management (3 issues)
 - Follow up (6 issues)
 - Diagnosis (3 issues)

Prioritize the issues

- Work group polled
- Issues rated on a 5 point scale
 - 1 being lowest priority
 - 5 being highest priority
- Polling results
 - seven issues received the highest priority
 - Getting data back for early intervention/Part C
 - Getting babies into diagnostic testing
 - Border babies
 - Physician follow up for older children
 - Educating practitioners about repeat testing
 - Getting PCP to support and encourage re-screening

Prioritize the issues

- Polling results (Continued)
 - Second highest priority 14 issues
 - Third highest priority: 9 issues

Develop model practices and/or protocols

Priority Issues

- Each state provided their strategies for each issue
- Each issue has a “lead state”
- Action Planning worksheet
 - Identify and select strategies
 - Feasibility, impact, resources needed, effectiveness
 - Provide strategies or protocols in a standard format.

Accomplishments & Products

- Developed data points for a web based reporting system of long term follow-up for deaf and hard of hearing children
 - Final set of data elements for regional – or national – long term follow-up
- Reviewed risk factors collected by Region 4 states
 - Summary of risk factors collected by Region 4 states
- Developed Border Baby protocol for Region 4 (adapted from Genetic protocols)
 - Region 4 recommended and supported guidelines for Border Baby follow-up (February 23, 2009)

- Identified and prioritized EHDI issues in Region 4
 - List of issues, indicating for how many Region 4 states it is a priority
- Identified promising practices for impacting issue
 - Lists of strategies for each issue
- Assessed strategies for recommendation/implementation
 - Action planning summaries, list of strategies for consideration

Putting it all together

- Revisited all 7 state follow-up protocols
 - Review of protocols
 - processes for identifying children who might benefit from referral to genetic services
 - Steps for referring
 - Information provided by brief survey of workgroup members
 - Michigan Guidelines included detailed process
 - Other Region 4 states reporting did not have clear protocols for identifying and referring to genetic services

Pending EHDI STFU WG Activities

- Development of a Regional/State brochure for parent information kits
- Development of power-point presentation for professionals (audiologists, speech-language pathologists, early intervention providers and primary care physicians) discussing the need and importance of a genetic referral for infants and toddlers with diagnosed hearing loss
- Survey to assess knowledge of genetic referral needs when a child has a newly identified hearing loss
 - Are referrals being made?
 - By whom?
 - How helpful?

Pending EHDI STFU WG Objectives

- Determine how referrals to genetic services are made post hearing loss diagnosis, by EHDI partners
- Seek to educate parents and professional regarding
 - The importance of genetic referrals
 - Genetic services available
- Identify strategies for assisting in accessing genetic services
- Promote state supported genetic centers

The final picture

- Model guidelines for EHDI follow-up that include processes and strategies to improve EHDI program success in identifying and referring children and their families for genetic services.

Challenges

- There are a range of genetic services available in each state for children identified through the EHDI program
- Access to genetic services for children with hearing loss is unknown to the R4 WG
- Relationships between EHDI and other follow-up programs varies by state.
- Guidelines may need to include multiple strategies at different process steps to facilitate adaptability for all Region 4 states – a menu from which each state can choose their strategies.

Our Products & Tools

- www.region4genetics.org
- info@region4genetics.org