

*Report of HRSA Priority 2
Projects*

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What is the mission?

*Improve outcomes and
access for affected
children identified by
newborn screening*

Regional Genetics Collaboratives in partnerships
National Coordination Center and NBSCTRN

Points for interaction

- ✦ Public health surveillance
- ✦ Point-of-care activity = clinical care
- ✦ Research agenda - PH + clinical
- ✦ Define parameters of acceptable lab performance for screening and confirmatory testing

*Three projects in progress
funded by HRSA - "Priority 2"*

- ✦ NEGC (Region 1): Building on experience in public health quality assurance in long term follow-up
- ✦ SEGC (Region 3): Building a business case for data collection in NBS
- ✦ Region 4: Building a platform for knowledge and research - the Inborn Errors of Metabolism Information System
- ✦ (plus collaborative project in creation of datasets: Mountain States)

**NEGC (Region 1)
Priority 2 Project**

QuickTime™ and a decompressor are needed to see this picture.

Building Upon the Foundation of Six New England States' Comprehensive Newborn Screening Programs for Sustainable Follow-Up

Building on Experience

- Long standing success of LTFU data collection in MA
- Programmatic integration of testing and follow-up
- Collaboration with other programs
- Priority: extend this successful strategy across the Region

Dual approach:

- Facilitate interstate data sharing practices; encourage authorization of LTFU
- Create regional workgroups for collaborative generation of data elements

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Focus on quality assurance

- NBS programs extending MA experiences in LTFU
- Data collection as a public health measure by Departments of Health
- No consent requested for data collection

UMASS MEDICAL SCHOOL | COMMONWEALTH MEDICINE

LTFU System Development in Partnership with PHII

- Development of requirements for an information system to track long-term patient and disease-oriented outcomes
- Builds on work recently completed in project jointly funded by HRSA/MCHB and Region 3.
- Project facilitated by PHII using the PHII Collaborative Requirements development methodology.

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PHII Methodology

- Phased approach to engage stakeholders in reaching consensus on common definitions, work flows and the essential functions that must be supported by a future LTFU information system.
- Work processes required to support care, follow-up, and research will be identified and mapped by a national committee composed of content experts.
- Upon completion of the collaborative analysis, requirements and specifications will be developed by the Region 3 Collaborative.

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Information Needs for LTFU

- For information systems to serve program needs, information needs must be identified
- Requires
 - Determining what the program does (business process analysis)
Simply: What is done by whom?
 - Determining what information is needed to accomplish the activities (requirements definition)

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Completion of four primary business processes

- Screening
- Confirmatory /diagnostic testing
- Transition to LTFU
- Intervention management(LTFU)

Next Steps

- Developing and publishing requirements for the business process to be used by other states
- Can be used for validating existing information systems



Long-term follow-up after NBS: the Inborn Errors of Metabolism Information System (IBEM-IS)

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For the
Region 4
Priority 2 Project
Workgroup



Condition registry as a research platform

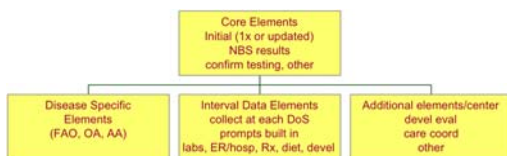
- Plan interventions that can be assessed with data in IBEM-IS
- Initial projects should examine
 - Natural history
 - Short term outcomes

We request at enrollment that registry subjects consider consent to allow continuing contact, anticipating engaging them as participants in future research trials

MCADD: How we initiated registry activity

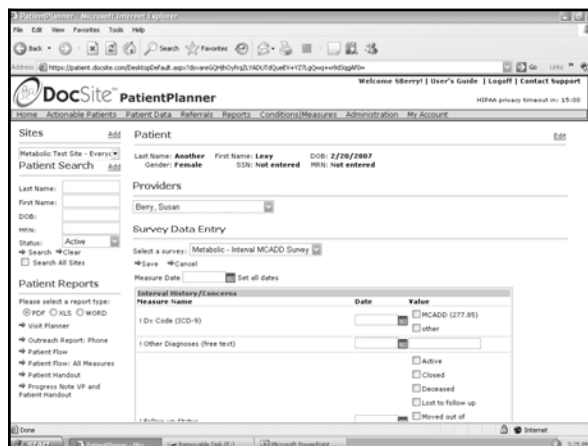
- Review of literature and existing plans for treatment to define disease-specific elements
 - Include disease-specific follow-up planning elements contributed by Mountain States Collaborative
 - Integrated elements in "Oregon database"
 - Identify elements that all agree are essential and that should be done uniformly
 - Identify elements that are anecdotal and could be subject to randomization
- ✓ *All states contributed treatment protocols so elements of difference could be characterized*


Plan: a relational database



Goal: add other IBEM, keeping core and interval elements, adding disease-specific elements, center-specific elements

The instrument: DocSite®






Building the data set: current progress in the R4 IBEM-IS 2/09

- Training for entry for 8 centers (two states have two centers)
- Only one state still pending IRB approval
- Entry proceeding for most FAOD, MSUD (enrollment and interval information)
- Data sets at <http://region4genetics.org/region4/dataelementssurveys.aspx>


Data entry:

MCADD	45 subjects
MSUD	7 subjects
LCFAOD	5 subjects
SCADD	3 subjects
TOTAL	60 subjects



What next??

- ◆ Defining the strategy/research agenda
 - Generally uniform f/u and reporting
 - Consensus on questions to ask?
- ◆ Add more disorders
- ◆ Collaboration with others
- ◆ Developing regional research projects
- ◆ *Planning integration of data with other data systems*



Mountain States Genetics Regional Collaborative Center

- ◆ Developing disease-specific datasets with quantifiable performance indicators and outcome parameters
- ◆ Plan systematic neuropsychometric evaluation
- ◆ Work collaboratively to use care plans in the entire Region
- ◆ Plan is to assure improved care, not dictate practice style
- ◆ Collect data using these defined datasets

Common attributes; differences

- ✦ Common:
 - ✦ Fostering collaboration in (and between) Regions
 - ✦ Using content experts to define datasets and workflows
- ✦ Differences
 - ✦ Products
 - ✦ Public health vs. clinically based
 - ✦ Use of informed consent