



IBEM-IS and National Data Collection

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For the

Region 4 Genetics Collaborative



NCC/RC goals

- ◆ Ensure that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home by:
 - Developing infrastructure and capacity
 - Strengthening communication and collaboration among public health, individuals, families, primary care providers, and genetic medicine and other subspecialty providers
 - Evaluating outcomes and using these data to enhance efforts



NCC - National Coordinating Center

- ◆ NCC - contract with HRSA
- ◆ Coordinate RGCs including R4 activities
- ◆ Includes LTFU data collection activity
 - R4
 - SE Region “Business case”
 - Mountain States “Minimal data sets” = AKA care plans
 - NYMAC - diagnostic criteria



Special Supplement to the NCC

- ◆ Supports LTFU planning for data collection
- ◆ Workgroup organized (SAB as chair)
- ◆ **Four components of LTFU identified:**
 - **Care coordination through a medical home**
 - **Evidence-based treatment**
 - **Continuous quality improvement**
 - **New knowledge discovery**



NCC LTFU supplement goals

- ◆ Accelerate and concentrate LTFU efforts.
- ◆ Determine information systems needs of state newborn screening programs to conduct LTFU of newborn screening identified patients.
- ◆ Incorporate findings into a public health evaluation tool that can be used to monitor and improve LTFU.



NBSCTRN

- ◆ Contract with NICHD
- ◆ Creates workgroups
 - Standing committee (Harvey Levy, chair)
 - LTFU
 - IT
 - Laboratories
 - Bioethics



NBS CTRN Initiatives

- ◆ Development national resources to support R&D related to newborn screening
- ◆ Long-term follow-up of NBS
- ◆ Developing evidence base for NBS candidate conditions
- ◆ Newborn screening laboratory network
- ◆ Clinical centers network
- ◆ Infrastructure development (informatics based)



Clinical Centers Networks

- ◆ **Diagnosis and long-term follow-up data**
- ◆ **Biospecimen repositories**
- ◆ **Funding opportunities**
- ◆ **Disease registries**



Joint activities

NCC/RC and NBSCTRN

- ◆ Establish initial clinical histories of disorders identified in NBS for use in public health and research initiatives
- ◆ Initial focus on metabolic diseases utilizing several LTFU data collection efforts as a baseline
- ◆ Expand focus to endocrinopathies, hemoglobinopathies, cystic fibrosis, hearing loss and other NBS identified disorders
- ◆ Communicate findings to groups developing health care information technologies standards for consideration
- ◆ Coordinate activities with GSB/MCHB/HRSA and NICHD



DHHS Secretary's Advisory Committee for Heritable and Congenital Disorders

- ◆ Prescribed by statute
- ◆ Work furthered by “NBS Saves Lives”
- ◆ Fostered the 29
- ◆ Serves as evaluative body for new disorders
- ◆ Brings together federal agencies
- ◆ Subcommittees



SACHCD

- ◆ LTFU Subcommittee
- ◆ Medical foods group - Kerry bill
- ◆ “Overarching Questions”
 - State and National PH
 - Specialty providers
 - Primary providers
 - Families and patients



Federal agencies all interested in LTFU

- ◆ HRSA
- ◆ CDC
- ◆ NIH
 - NBSCTR
 - RDCN



Where is R4P2 in this?

- ◆ NDCC
- ◆ Standing committee: CTRN
 - IT
 - Bioethics
 - LTFU
- ◆ SAC LTFU



National Dataset

- ◆ Used our dataset as template
- ◆ Major change suggested:
 - Include values
 - Disease specific info not yet defined



Up next?

- ◆ Uniform language for data sets
- ◆ Disease specific data sets
 - Workgroups
- ◆ IT planning
- ◆ Proposals (?)