

Inborn Errors of Metabolism Information System (IBEM-IS) in Information Technology (IT) Planning

Presented by:

Anne M. Jurek, M.S., Ph.D.

University of Minnesota Department of Pediatrics

November 20, 2009



Region4
Genetics Collaborative

Outline

- Goals of national database planning
- Meetings attended and presentations
- Describe the purpose of the IT workgroup
- National infrastructure for research data collection

National Database Planning Goals

- Integrating genetics into electronic health systems
- Creating a national database and repository for research information

Genetics in Electronic Health Systems Meeting

- February 2008
- Purpose
 - Describe the current genetics content
 - Identify gaps
 - Explore solutions for integration and adoption
- IBEM-IS presentation

Newborn Screening Translational Research Network (NBSTRN) Planning Meeting

- April 2009
- Purpose
 - Facilitate planning for the systems and structures underlying the NBSTRN
- Sue presented IBEM-IS
- Broke up into workgroups
 - Newborn Screening programs/Laboratories
 - IT
 - Clinical Centers

Purpose of IT Workgroup

- Make recommendations regarding the preferred technologies for a widely distributed network of participants dealing with rare disease patients

NBSTRN IT Workgroup Meeting

- October 2009
- Purpose
 - Begin discussions and recommendations on platforms and vendors
- IBEM-IS presentation

NBSTRN Infrastructure to Meet Needs

- Informatics system to support
 - Patient registry development
 - Protocol development
 - Standardized protocol and data languages
 - Data warehousing
 - Minimal duplication of work and expense
- Patient data
 - Identifiable data available for provider
 - De-identified data for public health
 - Appropriate to needs in long-term follow-up as an evaluation tool
- Biospecimen repositories

IT Infrastructure Future Directions

- Converting NCI's Cancer Biomedical Informatics Grid (caBIG) as appropriate or developing compatible tools
- Developing tools to link specimen repositories with registries and long-term follow-up clinical information