

**Advisory Committee on Heritable Disorders  
and Genetic Disease in Newborns and  
Children**

**Subcommittee on Follow up and Treatment**



**SAFER • HEALTHIER • PEOPLE™**

# Subcommittee members

- Coleen Boyle
- Christopher Kus
- Denise Dougherty
- Carol Greene
- Javier Aceves
- Brad Therrell
- Jill Fisch
- James Newton
- Alan Hinman
- Joseph Telfair
- Julie Miller
- Jill Shuger



# Focus on Long Term Follow up (LTFU) (...including Treatment)

- Guidelines available for short term follow – Corn, 1992; CLSI, 2006 and PEAS
- Little guidance for LTFU
- Varying definitions and goals of LTFU



## Existing Definitions – LTFU

“LTFU allows for the evaluation of the benefits resulting from newborn screening throughout the life of an individual. These benefits may impact the individual, the family, and/or society.....LTFU may include facilitation of care coordination services to ensure that the needs of the affected newborn/individual and family are met.” CLSI, 2006



# Existing Definitions -- LTFU

- “LTFU extends the period of follow-up substantially to monitor continuously the medical management and care coordination of those affected who require such services. LTFU also allows assessment of efficacy, sustainability, and safety of early treatment intervention, and can uncover new disease/treatment outcomes, and is valuable for demonstrating utility or limitation of screening.” Watson et al (2006)

# Activities of Subcommittee in LTFU

- Develop a position paper:
  - Working definition
  - Goal(s) and major components of LTFU and treatment
  - Short, medium and long term outcomes
  - Major participants/systems
- Meeting of stakeholders to provide input into the development of the position paper (4/18/07)
  - Major perspectives/systems impacted – individual/family, primary care, specialty care, public health, financial and regulatory, health information systems



# Core Components of LTFU

- Clinical Care/Treatment
- Care Coordination
- Quality evaluation/Surveillance ( as a public health mandate)
- Research Platform (clinical/interventional trials)



# LTFU – The Basics

- Goal – to achieve the best possible outcome for children and their families
- Life span approach with emphasis on transitions – focus to age 18/21
- Possible models:
  - Chronic Care Model: common diseases/disorders
  - Disease specific models – CF; Children's Oncology Network



# Clinical Care

- Access/manpower issues regarding speciality care
- More emphasis on collating and distributing available “best practices” and existing evidence



# Care Coordination (CC)

- Multiple facets of CC including public health component and clinical component
- “Medical home” as point of coordination
- CC may be disease specific
- Single “point of contact” for families for CC



# Evaluation/Surveillance

- Long term tracking of *natural history/treatment history*
- Federal mandate – as a *public health* function
  - Need to address privacy and genetic discrimination issues
- *Coordinated efforts* among federal agencies for collection and analysis of data



# Platform for Research

- *Care improvement* is an integral part of LTFU
- *Infrastructure* of clinical research
  - Clinical trials and observational studies
- *Translations* of research back into treatment, care and improvement



# Model for LTFU

- Hybrid model – Chronic care model + public health function
  - Clinical/Medical Home -- comprehensive focus (developmental, medical, mental health, education/support)
  - Public Health Component – surveillance and tracking
  - Health Information Exchange – tool to link components and help coordinate
    - ◆ Data elements standards for NBS conditions
    - ◆ Other federal initiatives addressing privacy/confidentiality



# Family/Individual Issues

- Family education, empowerment, and self management need to be integrated into the clinical care model
  - Include financial support
- Providers need to be trained how to partner with families
- Families need a point of contact for care coordination
  - Sustainable funding



# Personal Health Record

- Web-based, interoperable, personal health data
- Who maintains, who has access
- Role of Regional Health Information Organizations
- Standards (existing federal standards and need for data standards)
  - Family/individual control of access



# Next Steps

- Compete position paper
- Roles and Responsibilities:
  - Family
  - Health Sector – PCP and speciality care
  - Public Health





**SAFER • HEALTHIER • PEOPLE™**