

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

Subcommittee on Follow up and Treatment



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Subcommittee members

- Coleen Boyle
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- Denise Dougherty
- Carol Greene
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- Brad Therrell
- Jill Fisch
- James Newton
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- Joseph Telfair
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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
 - 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
- Coordination of Care/Services
- 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 framework:
 - Chronic Care Model: common diseases/disorders
 - Disease specific models – CF; Children's Oncology Network



Clinical Care

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



Coordination of Care/Services (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
- *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



Model for LTFU

- *Hybrid model* – Chronic care model + public health function
 - Clinical Component -- comprehensive focus (developmental, medical, mental health, education/support)
 - Public Health Component – surveillance and tracking
 - Health Information Exchange – tool to link components and help coordinate



Family/Individual Issues

- Comprehensive focus – developmental, medical, education, emotional/social
- Family education, empowerment, and self management
- Providers need to be trained how to partner with families



Personal Health Record

- Web-based, interoperable, personal health data
- Maintenance and access issues
- Role of Regional Health Information Organizations (HHS)
- Standards (existing federal standards and need for data standards)



Next Steps

- Complete position paper
- Roles and Responsibilities:
 - Family
 - Health Sector – PCP and specialty care
 - Public Health





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