What Questions Should Newborn Screening Long-Term Follow-Up Be Able to Answer?:

A (proposed) statement of the United States Secretary for Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children

Presentation to the United States Secretary for Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children

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Purpose of the Paper

- **Presents broad questions and important issues for consideration when assessing if LTFU is meeting its goal:**
  - achieving the best possible outcomes for affected children and families

The Process

- **Built off previous work of LTFU and Treatment subcommittee**
  - Kemper et al., *Genetics in Medicine* 2008; 10:259-261
- **Four components of LTFU**
  - Care coordination, evidence-based practice, continuous quality improvement, and new knowledge discovery
- **Reviewed data collection materials from the Regional Collaboratives, ACMG and NBSTRN, CDC projects**
- **One-day workshop (participants listed Appendix)**
- **Shared with NBSTRN/RCC LTFU Workgroup, APHL roundtable**

The Matrix

Audiences	Components of LTFU
Families	Care Coordination through a Medical Home
Medical Home/Primary Care Provider /Specialists/Investigators	Evidence-based Treatment
Nation/State	Continuous Quality Improvement
	New knowledge discovery

Care Coordination through a Medical Home

Families	Medical Home/Primary Care Provider/ Specialists	State/Nation
Is my child receiving coordinated care through a medical home?	**Are children/adolescents receiving coordinated care through a medical home?**	**Do children/adolescents receive coordinated care through a medical home?**
Does the child have a family-centered medical home? Is the family/child knowledgeable about the specific diagnosis? What do families need to facilitate follow through with treatment and care plans? Does the family/child have the skills and tools to self-advocate? Is the family/child prepared for transition to adolescent or adult system of care? What percentage of families/individuals receives carrier identification and age-appropriate genetic counseling that also addresses psychosocial implications?	What percentage of children (combined & by specific disease) identified by the newborn screening program have an individual health/care coordination plan that is updated at regular intervals?	What percentage of children/adolescents has a family-centered medical home? What percentage of children/adolescents has a care coordination plan that is regularly reviewed? Are services available? Do families of children/adolescents have financial access to services? What percentage of youth has successfully transitioned from a pediatric to an adult system of care? How many children are lost to follow-up?

Evidence-based Treatment

How is my child doing clinically?	**How are the children/ adolescents doing clinically?**	**How are the children/ adolescents doing clinically?**
What percentage of families reports a good understanding of their child's treatment regimen, options, and other medical and non-medical needs and resources?	Are children identified through NBS and enrolled in care doing better than those identified clinically? Are best practices used appropriately in treatment? How are these best practices communicated to the family?	What are developmental, physical, and mental outcomes among affected children? Are service providers using best practices?

Continuous Quality Improvement

Is my child getting the best care and treatment? How can I improve my child's outcome?	Am I doing the best for my patients?	How do we assure ongoing QI?
Is up-to-date information on treatment made available to families? What percentage of families feels they have ongoing access to age-appropriate education? Do families have the opportunity to be in communication with a medical team for effective management of their child's care?	Is there an annual review of best practices and care plan for each child across all levels of the care continuum?	Is there a coordinated ongoing process for collecting and synthesizing information about effective treatments? Is there a coordinated mechanism for connecting affected individuals with the most effective treatments or clinical research trials if the appropriate management is uncertain? Is there ongoing evaluation of the effectiveness of various treatment protocols/regimens? Are there policies in place at the state/national level that facilitate collection and exchange of information among all components of the NBS system?

New Knowledge Discovery

Is my child able to enroll in clinical research related to his/her disorder?	**Do children in my care have the opportunity to enroll in clinical research?**	**What clinical and observational long-term follow-up research efforts are being performed at the state and national levels?**
What percentage of families are engaged in the development of disease-specific registries, standardization of best practices and research studies?	What percentage of children are enrolled in clinical research related to their disorder and does enrollment in research influence outcome? Is knowledge gained from longitudinal studies informing clinical care and treatment development for children with these conditions?	Are high quality NBS surveillance and tracking systems in place at the state and national level? Do states use national data standards to collect data and link systems? Are safeguards in place to protect the privacy of children and families enrolled in clinical research? Are the results of basic, clinical and translational research incorporated into best practices for the care of children?

Next Steps

- Guide current and future data projects to develop systems that incorporate measures to address these issues
- Develop specific and measurable indicators for LTFU care after newborn screening, e.g. NCQA
- Assure adequate resources to accomplish the goals of LTFU care after newborn screening, as well as ensure continuing resources for LTFU care in the future