



Newborn Screening Translational Research Network (NBSTRN) and LSD Pilot Studies



Michael Watson, MS, PhD, FACMG
ACHDNC
May 14, 2010

- ◆ **Supporting pilot studies of new conditions in newborn screening including diagnostic, management and follow-up protocols and associated IT, informatics and databases**
 - **SCID**
 - **LSDs**
 - ◆ **Supporting the development of new technologies for use in newborn screening**
-

◆ **Pompe needs**

- **Pilot studies in diverse US population**
- **Pilot testing using technologies likely to be used in US (e.g., MS/MS, bead-array technologies, microfluidics)**

◆ **Krabbe**

- **Not recommended for addition to NBS at this time**

◆ **Niemann Pick**

- **Not sent for evidence review due to incompleteness of treatment clinical trials**

◆ **Gaucher and Fabry**

- **Hang on to your hats**
-

◆ **New York**

- Screening for Krabbe for two years
- Legislation proposed to expand to 4 additional LSDs

◆ **Illinois**

- Mandates addition of 5 LSDs (Krabbe, Pompe, Niemann Pick, Gaucher, Fabry)
 - To be initiated in October, 2010

◆ **Missouri**

- Mandates addition of 5 LSDs above plus any others that become amenable to NBS

◆ **Washington**

- Limited NICHD pilot study testing new MS/MS technologies

◆ **Perkin Elmer supplemental screening**

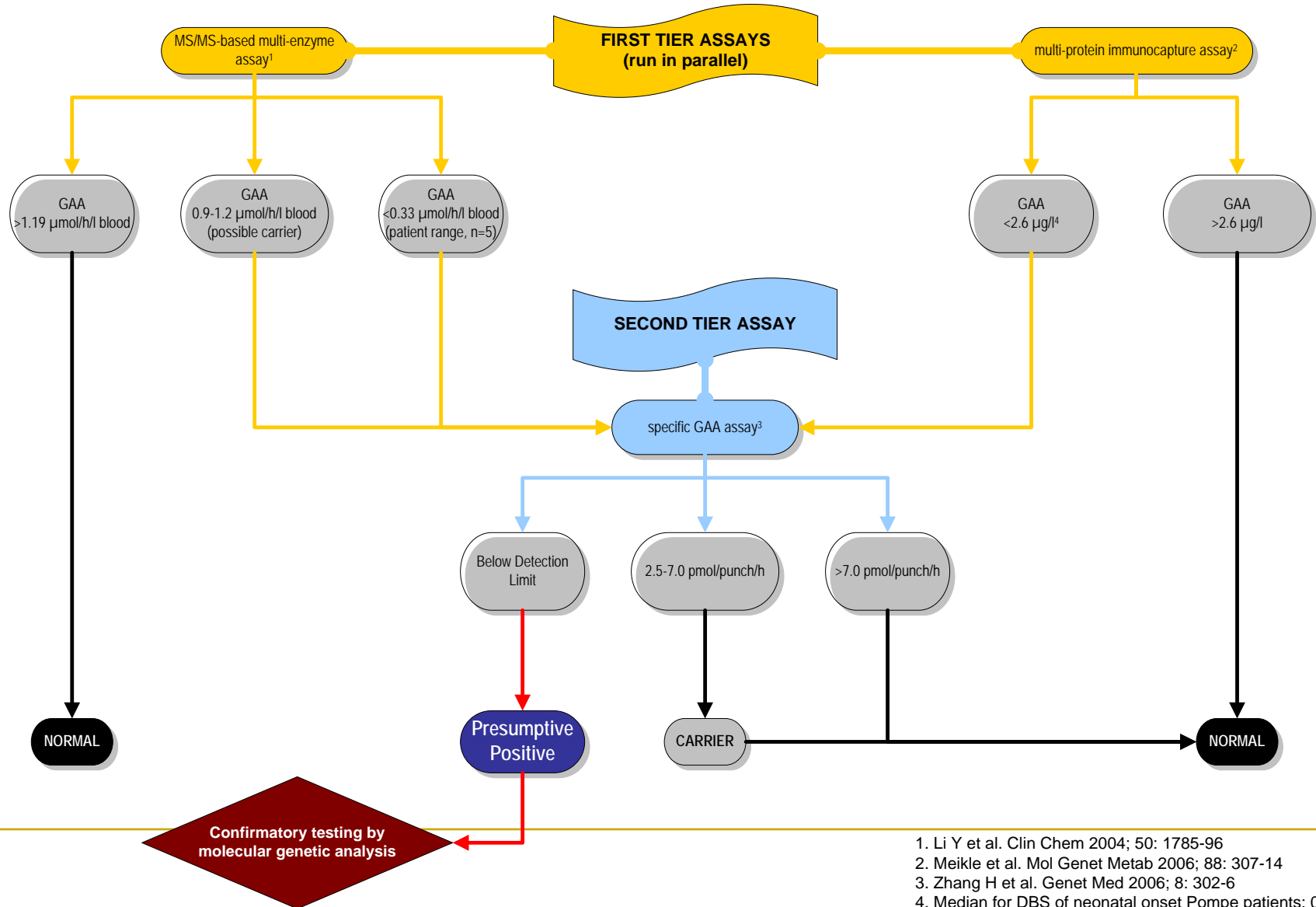
- ◆ **Primary care provider guidance**
 - **Provider networks**
 - **ACT Sheets**
 - **Guideline on Diagnosis and Management of the Asymptomatic LSD Patient**
 - **Diagnostic algorithms**
 - ◆ **Specialist and sub-specialist guidance**
 - **Establishing preferred technologies for NBS**
 - **Developing diagnosis and management protocols**
-

- ◆ **Mayo studies organized by Dieter Matern**
 - **MS/MS**
 - **Bead-based immunoassays**
 - ◆ **Adding microfluidics (Advanced Liquid Logic) to comparison**
 - ◆ **Normalizing to enzyme activity**
-

- ◆ **Organizing LSD experts for protocol development and associated language standardization**
 - **First substantive meeting in June 2010**
 - **Building off of LTFU data sets already developed to include LSD disease specificity**
 - **Developing databases to support collaborative pilot testing**
-

Screening Strategy

Example: Pompe Disease




1. Li Y et al. Clin Chem 2004; 50: 1785-96
 2. Meikle et al. Mol Genet Metab 2006; 88: 307-14
 3. Zhang H et al. Genet Med 2006; 8: 302-6
 4. Median for DBS of neonatal onset Pompe patients: 0.2 μg/L (n=3)

HOME | LINKS | SITEMAP Search: GO


Welcome

GC C T A G A C G A T T G G G T A C G C F T A G A C G T A T T G C F T A

ABOUT | CALENDAR | PARENTS | CONTACT




NBSTRN
NEWBORN
SCREENING
TRANSLATIONAL
RESEARCH
NETWORK



The Role of NBSTRN

The NBSTRN is a research resource for investigators engaged in newborn screening-related research. This may include new technology development, the clinical history of heritable disorders, and new therapy development.



Resources

Education

Services

The NBSTRN is funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development National Institutes of Health

[Home](#) | [Contact Us](#) | [Webmaster](#) | [Site Map](#)
 7220 Wisconsin Avenue, Suite 300 | Bethesda, MD 20814
 Telephone: 301-718-9603 | Fax: 301-718-9604
 E-mail: nbstrn@nbstrn.org

Hot Topics

[Save the Date!](#)

[Blood Feud: Controversy Arises Over NBS Program](#)

[Disputes over Research with Residual NBS Blood Specimens](#)

[The Test That Saved My Baby's Life](#)

[Living Proof](#)

THANK YOU!

NBSTRN is funded by contract HHSN27520080001C from the *Eunice Kennedy Shriver* National Institute for Child Health and Human Development, National Institutes of Health, HHS

NNC/RCs are funded by cooperative agreement MCO3957 from the Maternal and Child Health Bureau of the Health Resources and Services Administration, HHS