The Current State of Newborn Screening (NBS)

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Focus and Mission

Public Relations

New and Emerging Screening Disorders

50-Year Anniversary Celebration

Rapidly Advancing Technology

Ethics / Privacy

Fiscal Limitations

Standardization and Harmonization

Emergency Preparedness
Celebrating 50 Years of Success in Public Health

When is your State’s 50th NBS Anniversary?
50-Year Celebration Campaign

• Key Messages:
  o Newborn Screening saves and improves the lives of more than 12,000 babies in the U.S. each year.
  o Newborn Screening is a quick and safe way to help protect your child against certain diseases and medical conditions.
  o Newborn screening is one of the most sophisticated tools in the arsenal to protect public health.
  o Public health laboratories are ever-vigilant, protecting our families’ health and saving lives every day.
Advises the Secretary of Health and Human Services regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk for, heritable disorders.
Newest Screening Disorders

• Severe Combined Immunodeficiency (SCID)
  ◦ Made the nationally Recommended Uniform Screening Panel (RUSP) in May 2010.
  ◦ The first NBS disorder that can be cured with a 97% success rate if detected and treated soon after birth.
  ◦ The first molecular primary NBS screening test.
  ◦ Has already been adopted by 12 states (50% of U.S. babies being screened).
April 2013 Implementation Status for SCID

Slide Courtesy of Newborn Screening Translational Research Network
Newest Screening Disorders

• Critical Congenital Heart Disease (CCHD):
  - Recommended by the SACHDNC and adopted into the RUSP by HHS in September 2011.
  - CCHD is a group of heart-related conditions present from birth that cause nearly 3% of infant mortality during the first year of life.
  - Early detection utilizing pulse oximetry screening at the birthing centers before discharge of newborns allows for immediate intervention to prevent brain damage and death.
  - Several states have implemented or are in the process of implementing CCHD screening in their state.
2013 Vital Statistics

16 states have passed legislation

18 States with Active Legislation

Sept 2011: 0.03% screened

Sept 2012: 10% screened

Q2 2013: 20% screened

source: www.cchdscreeningmap.org
Newest Screening Disorders

• Lysosomal Storage Disorders (LSD):
  
  o Pompe has been recommended to the Secretary of HHS for addition to the RUSP by the SACHDNC in May 2013.
  
  o Pompe is an inherited disorder whereby stored glycogen is unable to be broken down and causes severe cell damage throughout many parts of the body, especially in the muscles and heart.
  
  o Early treatment with enzyme replacement therapy (ERT) can stop the progression of cell damage and prevent crippling disabilities from occurring.
States Screening or Planning to Screen for LSDs - 2013

- Krabbe screening through NY.
- Pompe, Gaucher, Fabry, and Hurler in statewide pilot.
- Legislated but not yet implemented
- LSD methods comparison, Mayo/MN partnership
- Pilot study for Pompe, Gaucher, Fabry and Nieman-Pick
Rapidly Advancing Technology

Digital Microfluidics
National Standardization and Harmonization

• Great strides have been made with the help of SACHDNC and many other NBS partners and stakeholders.
  o Which disorders should be screened/reported
  o What do we call them; what acronyms do we use.
  o Recommendations on sample collection, testing and patient follow-up
  o Recommendations on residual sample policies
  o Definition of NBS terms such as positive, false positive, positive predictive value, and even the term “screening”.

2013 APHL ANNUAL MEETING & 7TH GOVERNMENT ENVIRONMENTAL LABORATORY CONFERENCE
How do we count what we do?
Components of NBS (It takes a village)

• Education (throughout the process)
• Screening, including specimen collection, transport, testing and reporting
• Follow-up and tracking
• Confirmation and diagnosis
• Management
• Program evaluation and CQI
Residual NBS Sample Policies

- A wide range of policies and practices are in place among states:
  - Mandated destruction of residual samples upon NBS completion.
  - Storage of samples for a large spectrum of time periods. Some states allow research.
  - Many states have opt-in or opt-out alternatives.
  - Wide range of storage scenarios (Bio-Banks to Back Rooms).
Missouri is an opt-out state. Parents may write a letter requesting the leftover NBS sample be destroyed, sent back to them or stored but not used. Otherwise it will be stored for 5 years and be available for anonymous research.
Jefferson City, Missouri, flood of 1993
Missouri State Health Laboratory, Flood of 1993
NBS Growth Preparedness

Jessy and Dustin (parents) and son Brady Cunningham with Bob Evanosky
NBS Growth Preparedness

- Public Health Laboratories and NBS Programs must be in-the-know about the what’s and how’s of all things on the NBS horizon.
- Know where you stand on what your State can and wants to screen.
- Collect all the facts and be proactive with your Genetics Advisory Board and Departmental Administration.
- Utilize the help of all NBS partners, especially APHL.
Thank You