RUSP Roundtable examines perspectives and potential changes for newborn screening

Rare Disease Day 2016 – NIH, Bethesda MD

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Introduction

The inaugural RUSP Roundtable, hosted by MLD Foundation, took place August 26, 2015, in Rockville, Maryland. The Roundtable convened newborn screening (NBS) key opinion leaders in a unique, independent forum to speak with candor about challenges, concerns, and developments impacting their daily work with an eye toward the future of NBS and improving quality of life for the newborn and their family.

Purpose

The RUSP Roundtable creates a permanent space for NBS key opinion leaders to share perspectives and insights, expand the common knowledge base, and identify opportunities for both coalition building and loose collaboration across sectors to innovate and accelerate solutions for a more robust and equitable NBS system. Where possible, specific issues and opportunities will be collaboratively addressed with much greater detail.

RUSP Roundtable History

Dean Suhr, President and co-founder of MLD Foundation has been active in the Rare Disease community on behalf of all rare diseases for nearly a decade. Recognizing that aside from the chemistry for each NBS screen, nearly every rare disease shares the same overall NBS policy and implementation issues, he started to engage and better understand dynamics, policies, and influences on developing and implementing a new NBS.

He found a system with many competent hard-working knowledgeable people that was the lacking broad cross-functional communications and collaborations, and from an outsider's view somewhat dysfunctional – not in what was being done, rather how the system adapted to change including new screens, patient and family centricity, genome sequencing as a NBS tool, and an inherent public/private public health/research conflict.

The RUSP Roundtable is very non-traditional as far as government and public health systems go, has no formal authority, no formal charter or host agency, and no official role in the NBS community ... yet.

NBS History

1961 – Universal NBS started when Robert Guthrie developed an assay to screen newborns for phenylketonuria (PKU), a rare and dangerous heritable disorder that can be well managed through a restricted diet. With support of the Kennedy Administration and state chapters of the National Association for Retarded Children, Guthrie lobbied states to require PKU screening. That set the precedent for state-mandated public health NBS.

It should be noted the RUSP Roundtable, while an initiative of MLD Foundation, is not about metachromatic leukodystrophy – rather it is about improving quality of life for newborns, children, and their families.

2004 – The US department of Health and Human Services HHS the American College of Medical Genetics and Genomics (ACMG) to identify the conditions for uniform NBS.

2005 – ACMG released a report recommending a core panel of 29 conditions, and a secondary panel of 25 conditions incidental to the core panel.

2006 – the HHS Secretary's Advisory Committee on Heritable Disease in Newborns and Children (SACHDNC) endorsed the ACMG recommendation for their initial Recommended Uniform Screening Panel (RUSP).

NBS is a State Public Health Program

Newborn screening consists of state public health programs run autonomously by the states so they are free to ignore, follow or exceed the federal RUSP. By 2009, most states had adopted the RUSP as a primary guide for their state panels.

NBS addresses merely a handful of the over 6,000 heritable diseases, most of which are rare diseases that primarily affect children. Thirty-four* conditions are on the RUSP core panel and twenty-five are on a secondary panel. Prior to 2006, some states tested for as few as four disorders and others for as many as fifty, raising concerns about social equity. A newborn could live or die depending upon which side of a state line the child was born.

** X-ALD and MPS-I were added to the RUSP in February 2016*

RUSP Criteria

Although the ACMG report initially informed the RUSP process and ACHDNC outlook, over time the Committee narrowed its focus to evidence-based review of clinical utility to the exclusion of factors that impact families and quality of life.

1a) condition is well understood

1b) has a significant health impact on the infant

1c) would likely not be promptly identified without NBS

2) effective and inexpensive screening test is available

3) existing viable therapy to treat the condition

4) cost-effective, accurate diagnostics to confirm a potential positive screen result

5) prospective population-based pilot study to validate the screen

Attendees

The attendees represented a very wide swath of perspectives on NBS and were put in an environment that fostered open sharing, educating, inquiry, postulation and discussion. The attendees included:

- **academia**
- **state and federal public health**
- **governmental agencies**
- **ACHDNC**
- **clinical and social research**
- **private industry**
- **patient advocacy**
- **clinicians**
- **geneticists**
- **ethics**

Though invited, healthcare payors declined to participate. It is desirable to have the payer perspective at future meetings as well as representatives from some of the key medical societies and the ACMG.

After about 2 hours of discussions the guards came down, the "cones of silence" and the discussion proceeded in earnest with sincere open conversation.

All of attendees expressed they learned something new from the other perspectives that will help them as they work on their respective aspects of NBS.

Highlight of Discussion Topics

Some of the topics discussed:

- What is the meeting of benefit?
- Who is the target of benefit? Just the child, or should benefit include the family and society?
- RUSP nomination process ... challenges and obstacles
- Obstacles to rare disease research and biotherapeutics development
- Trends and issues in informed consent for screening and subsequent dried blood spot (DBS) usage
- Next-generation genomic sequencing (NGS) in relation to current NBS technologies
- Other emerging technologies and trends in the private sector
- Health record data accountability and protection
- Scope and purview of the ACHDNC
- Public health is not a research platform
- Growing impact of advocacy groups and the patient-centered care movement
- Secondary and parallel paths for additional screening
- Equal access to NBS
- Resource constraints and potential solutions ... appropriations, equipment, staff, space, follow up resources,
- Public awareness and perceptions about newborn screening
- Is the viable therapy requirment still valid? Is just knowing your child has a particular disease a benefit?
- Potential policy changes at the state and federal level
- Possible alternatives and solutions not excluding policy change
- Impact of state legislated screening for conditions not on the RUSP

Follow Up & Next Meeting

The Roundtable reconvened in February 2016, and will meet again immediately adjacent to the next Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) quarterly meeting (currently scheduled for July 2016).

For more information

Please visit http://NewbornScreening.us or contact Dean Suhr at 503-656-4808 or email dean@MLDfoundation.org