Early Check: Rationale and Progress Toward a Voluntary Newborn Screening Program

NBSTRN Network Meeting, September 2015

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RTI International

Early Check™

Expanded health screening for your baby

Disclosures

- Current funding to support this effort from The John Merck Fund and the National Center for Advancing Translational Science
- Current funding for other projects from NICHD, CDC, and Novartis Pharmaceuticals
- I am an appointed member of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, but am not speaking on behalf of the Committee.
- I am a parent of an adult daughter with FXS
- I think this is a good idea!

Four emerging "disrupters" are now posing significant challenges to the RUSP

- Slow, condition-by-condition review
- Frustration by patient advocates, leading to state-level advocacy
- Commercial options for screening outside of public health
- New screening methods on the horizon

VIEWPOINT

Newborn Screening
Evolving Challenges in an Era of Rapid Discovery

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Newborn screening is designed for presymptomatic identification of serious conditions for which there are effective treatments. Because newborn screening programs in the United States are operated by states, there has historically been considerable cross-state variability in screened conditions and thus a need for a mechanism to guide states.

The Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, which was ap- disease) and 2 (Pompe disease and mucopolysaccharidosis I) are still under consideration.

Eight conditions have been reviewed but not recommended (spinal muscular atrophy, Niemann-Pick disease, neonatal hyperbilirubinemia, Krabbe disease, hemoglobin H disease, Fabry disease, adrenoleukodystrophy, and 22q11.2 deletion syndrome). Even though a careful and deliberative process can be justified, a central question is whether this

JAMA The Journal of the American Medical Association

The ACHDNC is facing challenges in evidence review

U.S. Department of Health and Human Services

Advisory Committee on Heritable Disorders in Newborns and Children

The evidence base for newly nominated conditions **is weak**

Rare diseases are caught in a classic "**Catch 22**" situation – screening cannot be mandated without evidence but screening is needed in order to gather the evidence

We are developing *Early Check*, a voluntary screening program

Offers a choice for families who want to know about other conditions

Early Check™

Expanded health screening for your baby

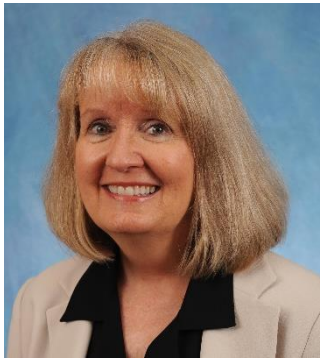
Provides first-ever opportunities to

- Determine population prevalence
- Detect early symptom onset
- Examine genotype-phenotype correlations
- Test pre-symptomatic treatments

Could reduce the length of time for conditions to be added to the RUSP

We are building an interdisciplinary team

Don Bailey



Cindy Powell

Lisa Gehtland

Melissa Raspa

Anne Wheeler

Megan Lewis

Ryan Paquin

Jennifer Taylor

Holly Peay

Doug Therique

We are building collaborative partnerships with two states

State Laboratory of Public Health

NCPH North Carolina Public Health

- Lab director, Secretary of HHS, and NBS Advisory group have approved participation in planning
- We are now an "administrative partner" with the lab and have led two partnership applications
  - New SCID pilot award from CDC
  - NICHD pilot award proposal under review

Wisconsin State Laboratory of Hygiene
UNIVERSITY OF WISCONSIN-MADISON

- Presenting to state NBS advisory board in December
- Joint RTI-UW application submitted for ELSI Center to study ethical issues evoked by *Early Check*, especially consenting an entire state
- Joint RTI-UW-UNC-Duke Innovation Award proposal submitted to NCATS to build infrastructure for pre-symptomatic clinical trials

We have completed a Parental Willingness Survey

- **Online survey of 1000 recent and expectant parents**
- **Several sections, including**
  - Newborn screening awareness & attitudes
  - Fragile X, Duchenne muscular dystrophy, & spinal muscular atrophy
  - Willingness to participate
  - Information seeking re: infant health or pregnancy
  - Health care system distrust
  - Relationship characteristics
- **Embedded mixed-level fractional factorial design**

Levels	Timing of Consent	Cost to Participate	Choice of Conditions	Consent Format
1	Birth	Free	Fixed (all or none)	Electronic
2	2-weeks	$10, with waiver	À la carte	Print
3	2-months	$10	–	–

Activities planned for 2015-2016

Continue planning and communication

Finalize strategic vision and logic model

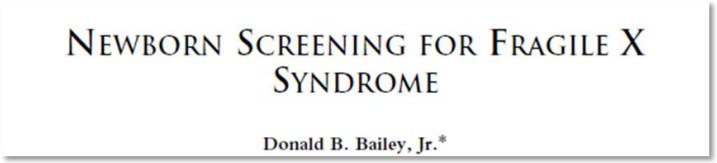
Build public web site
www.earlycheck.org

Form advisory group(s)

Develop plan for IT and informatics

Continue planning and communication

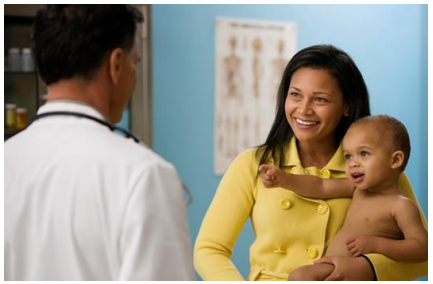
Develop a decision model for conditions to be included


NEWBORN SCREENING FOR FRAGILE X SYNDROME

Donald B. Bailey, Jr.*

Develop follow up and treatment protocols

Develop a plan for consent



Develop business model

- Federal
- Advocacy
- Commercial
- Private

General timeline for *Early Check*

YEAR	PRIMARY ACTIVITIES
2015	Initial planning, build stakeholder community, finalize state partners, determine likely conditions, initial data collection from parents and professionals; **submit applications for support**
2016	**Field test models** for awareness and consent, determine screening protocol for selected conditions, develop workflow, personnel and bioinformatics structure, develop framework for follow-up services, identify key research questions and measures, develop funding model
2017	Finalize print and web-based materials, complete logic model and workflow, purchase any needed equipment, hire and train lab technicians, **clear protocol for services and follow-up**, potential launch in Fall, 2017
2018-2020	Offer screening to **500,000 families**, identify screen positive children, implement follow-up services, surveillance and treatment studies