RTI International

- Non-profit research institute, with headquarters in RTP, NC
- More than 5000 employees, offices throughout the U.S. and around the world
- Entirely supported by grants and contracts
- Four major groups:
  - Social, Statistical, and Environmental Sciences
  - Discovery Sciences and Technology
  - International Development
  - Health Solutions
RTI is involved in many different collaborations and partnerships, e.g.:
NC Precision Health Collaborative

**Vision:**
To advance transformative precision health through innovation and partnership

**Mission:**
To accelerate collaborative initiatives that foster research, enable providers, engage industry and empower citizens to improve health outcomes and optimize resource utilization in North Carolina

Center for Newborn Screening, Ethics and Disability Studies

Advancing the science and practice of newborn screening

Developing evidence-based solutions to social and ethical challenges

Understanding the nature and impact of disability for children and families

Families
Strategy

- Form an interdisciplinary team of investigators focused on the broad mission of the center
- Align the center’s work with other groups at RTI, drawing on key strengths across the institute to support the center’s mission
- Solidify partnerships with North Carolina universities, the North Carolina State Laboratory of Public Health, and advocacy groups
- Establish NIH funding and peer-reviewed publications as our primary reputational identity
- Build and grow a diversified portfolio of projects to support our mission
- Establish at least one signature program engaging the entire center
- Be a trusted and objective source of evidence to inform policy and practice
Goals for this presentation

- Describe the current context of newborn screening in the U.S.
- Discuss challenges in gathering the data needed to inform newborn screening policy
- Describe a proposed solution – Early Check – a research infrastructure designed to enable research that informs newborn screening policy for rare conditions
Two different pathways to identify a health problem

Screen when an individual begins to show symptoms

Screen the population before symptoms appear
Two different pathways to identify a health problem

Screen when an individual begins to show symptoms

Screen the population before symptoms appear

Use if timing of treatment is not critical, or screening is not practical
Two different pathways to identify a health problem

Screen when an individual begins to show symptoms

Screen the population before symptoms appear

Use if treatment works best when delivered before symptoms are obvious
Newborn screening policy in the U.S.

- More than 4,000,000 babies each year, screened in state-based programs
- Historically considerable cross-state variability
- Now a well-established process for reviewing NBS nominations
- Rigorous review and recommendation by a national committee of experts
- DHHS Secretary determines whether a condition should be on the RUSP (Recommended Uniform Screening Panel)
- States decide
- Successful state expansion and national harmonization
- 34 conditions on RUSP
- States are gradually implementing newly recommended conditions
Many conditions will not make the RUSP in the near future

- Since the Committee was formed, 8 conditions have been reviewed but not recommended
  - Spinal muscular atrophy, Niemann-Pick, neonatal hyperbilirubinemia, Krabbe disease, hemoglobin H disease, Fabry disease, 22q11.2 deletion syndrome, GAMT
  - SMA was recently recommended but the DHHS Secretary has not yet approved

- Major reasons for not recommending
  - No accurate and cost-effective screening test
  - Implementation would be too complicated or expensive
  - No pilot study data
  - No data on pre-symptomatic treatment efficacy

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
No randomized controlled studies comparing a screened population versus a non-screened population

OBJECTIVES: To assess whether there is evidence that newborn screening for galactosaemia prevents or reduces mortality and morbidity and improves clinical outcomes in affected neonates and the quality of life in older children.

SEARCH METHODS: We searched the Cochrane Cystic Fibrosis and Genetic Disorders Group Trials Register comprising references identified from electronic database searches, handsearches of relevant journals and conference abstract books. We also searched online trials registries and the reference lists of relevant articles and reviews. Date of the most recent search of Cochrane Cystic Fibrosis Group's Trials Register: 18 December 2017. Date of the most recent search of additional resources: 11 October 2017.

SELECTION CRITERIA: Randomised controlled studies and controlled clinical studies, published or unpublished comparing the use of any newborn screening test to diagnose infants with galactosaemia and presenting a comparison between a screened population versus a non-screened population.

DATA COLLECTION AND ANALYSIS: No studies of newborn screening for galactosaemia were found.

MAIN RESULTS: No studies were identified for inclusion in the review.

AUTHORS' CONCLUSIONS: We were unable to identify any eligible studies for inclusion in this review and hence it is not possible to draw any conclusions based on randomised controlled studies. However, we are aware of uncontrolled studies which support the efficacy of newborn screening for galactosaemia. There are a number of reviews and economic analyses of non-trial literature suggesting that screening is appropriate.
To help solve this problem, we are designing and preparing to launch Early Check

- Innovation Award from NCATS, with additional support from NICHD, The John Merck Fund, Asuragen, and CureSMA
- Building a statewide research infrastructure
- Studying the benefits of pre-symptomatic screening and treatment for rare disorders, beginning with:
  - Spinal muscular atrophy
  - Fragile X syndrome
Overarching goals for Early Check

- Develop and evaluate methods to inform 120,000 parents/year about a research program and seek permission to participate in Early Check
- Accelerate the acquisition of data to support decisions about adding new conditions to the Recommended Uniform Screening Panel (RUSP)
  - Gauge parents’ interests in screening for new conditions
  - Test potential for large-scale screening in a state public health lab
  - Understand population prevalence and early natural history of “screen positive” infants
  - Identify infants who could participate in pre-symptomatic treatment trials
- Provide the foundation for an envisioned future in which states offer screening for a voluntary panel of “non-RUSP” conditions
Center partners

UNC-CH is Central IRB

RTI Center for Communication Science
Duke University
NC State Laboratory of Public Health
Funding partners and advocates

UNC - Chapel Hill
Wake Forest Baptist Medical Center

Megan Lewis
Ryan Paquin
Linda Squiers
Loren Pena
Sara Beckloff
Scott Zimmerman

Cynthia Powell
Nancy King
Sara Beckloff
Scott Zimmerman

Loren Pena
North Carolina state newborn screening lab partnership

- RTI is an administrative partner with the lab, and we have signed a Business Associate Agreement
- Together we have received funding to implement pilot studies to screen for
  - SCID (CDC)
  - MPS I (NICHD)
  - X-ALD (NICHD)
  - SMA (NICHD)
- We have received other awards to
  - support timeliness (APHL)
  - purchase sequencing equipment (CDC)
  - purchase two real-time PCR instruments (Cure SMA)
How it works – high-level overview of Early Check

- **Informational materials**
  - Prenatal clinical settings

- **Recruitment**
  - Birth hospital and 1 week after birth

- **Consent**
  - Online parent permission

- **Screening test**
  - Positive result
  - Confirmatory test
  - Diagnosis
    - Information, referral to treatment, psychosocial support, care coordination

- **Registry**

- **Clinical trials**

- **Longitudinal assessments**
  - Child development
  - Family adaptation

- **Family support**
  - Ongoing psychosocial support and family engagement

- **Health services research**

- **Negative result**
  - Inform parents
**Problem:** Telling 120,000 parents per year about Early Check…

**Solution:** Phased studies of targeted outreach strategies

- **Phase 1 (~6 months)** will determine enrollment rates as a function of three key strategies:
  - Awareness information to professional organizations and providers
  - A postnatal letter and flyer
  - Email reminders

- **Phase 2:** Pre- & Postnatal (Social media: FB, Instagram)
- **Phase 3:** Pre- & Postnatal (Materials in Clinician offices)
- **Phase 4:** Pre- & Postnatal (Paid targeted ads via Google)
Awareness to providers and professional organizations

- Primary goals:
  - Build awareness about Early Check
  - Reinforce consistent messaging – *if parents ask about EC, encourage them to visit the web site to learn more and enroll if they want to*

- Major strategies
  - Letter to providers from NCLSPH
  - 1-page FAQ and 1-minute YouTube video
  - Outreach to professional associations and state conferences
    - Ideally we will secure “endorsement” from key organizations
Outreach to providers – need some champions!
300-400 infants are born each day in NC. Blood spots drawn, demographic data are entered, and cards are shipped to NCSLPH.
NCSLPH will export demographic data daily to RTI’s secure LIMS.

Demographic Entry in NC NBS LIMS

Demo Export from StarLIMS

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Postnatal letter and brochure will be mailed within 5 days after birth.
Interested parents go to the Early Check web site to learn more

- www.earlycheck.org
The "enroll" button takes parents to the permissions portal.
Welcome to Early Check! Let’s get started.

Early Check is a research study that checks for two rare but serious health problems in newborns. The screening tests are free.

Parents can sign up when pregnant or within 4 weeks after the baby is born.

Watch this short video to learn about Early Check. You can read the text instead of watching the video by clicking the “Read” link.

Congratulations on your baby!
Ready to find out if your baby can join Early Check?

Find out if you're eligible for Early Check

Was your baby born in North Carolina?

NO  YES
Ready to find out if your baby can join Early Check?

Find out if you're eligible for Early Check

Where does your baby live?

SOUTH CAROLINA   NORTH CAROLINA

ANOTHER STATE
Ready to find out if your baby can join Early Check?

Find out if you're eligible for Early Check

It looks like your baby is eligible for Early Check!

Choose "next" to start the sign-up process.

Next

← Back
How is Early Check done?

Watch this second short video about Early Check.
Click “play” to begin. You can read the text instead of watching the video by clicking the ‘Read’ icon.
Why might you say yes to Early Check? And why might you say no?

It's OK to decide yes or no to Early Check. All research has benefits and risks. You should make the best decision for your family.

Watch this last short video about Early Check. Click "play" to begin. You can read the text instead of watching the video by clicking the 'Read' icon.

Here is a checklist to help you decide:

- Would you want to know if your newborn has one of the health problems screened in Early Check?
- Are you okay knowing that right now there is no cure for the health problems?
- Do you have the information you need to make the decision?
- Do you feel ready to learn the answer of the screening tests?
- If your baby has a screening test that is not normal are you okay with your baby having a second test to tell for sure?

If you checked most of these questions, maybe you are ready to sign up your baby. If you did not check most of these questions, maybe you are not ready or it is not the right decision for you.

Parents can contact the Early Check team to answer questions.

Read
Let's Review

It's our job to explain Early Check to you. Let's check in and see how we are doing.

Which babies can be screened in Early Check?

a. Babies who have regular newborn screening in North Carolina

b. Babies at higher risk

c. Babies with a family history of rare health problems.

The correct answer is a. Babies who have regular newborn screening in North Carolina and who live in North or South Carolina are invited to join Early Check.

Answers b and c are also correct because we ask mothers of all babies to join Early Check. We don't know anything about the health of any baby or about any baby’s family history when we invite the mother to join.
Which is **false** (not true) about Early Check?

Early Check:

   a. is a research study
   
   b. checks newborns for rare health problems 
   
   c. screens for health problems that can be cured 
   
   d. uses the same blood spot used for newborn screening

**Answer: Item c is false.** Early Check screens newborns for rare health problems that currently **cannot** be cured, though there may be treatments that can help.

**Items a, b, and d are true.** Early Check is a research study and you can choose whether or not to join Early Check. Early Check uses the same blood spot used for newborn screening.
Let's Review

Which is **true** about the Early Check screening test?

a. It may not find every single baby with the health problems.

b. It will check for every kind of health problems in newborns.

c. If the screening result is not normal, the baby definitely has the health problem

**Answer:** Item a is **true.** Screening tests are not perfect and may miss a small number of babies with the health problems.

**Item b is not true.** Early Check screening will only check for a few specific health problems.

**Item c is not true.** The screening tests are not perfect. We will use a second test for babies with a screening test that is not normal. A small number of those babies will have a normal second test because they do **not** have the health problem.
Would you like to join Early Check?

- Yes, I am ready to sign up for Early Check
- I would like time to think it over
- I have questions for the Early Check team first
- No, I do not want to sign up

The Early Check study has been approved by the University of North Carolina at Chapel Hill Institutional Review Board (IRB). An IRB is a committee that reviews research to protect the rights and welfare of participants. If you have questions about your rights as a research participant, you may contact the IRB at 919.966.3113.
Yes, I am ready to sign-up for Early Check
I have questions for the Early Check Team

If you have any questions, please contact us.

Email us  Call us  Chat with us
I would like time to think it over

Need a reminder?

If you would like time to think about Early Check, please enter your email address and we will send you a reminder in a couple of days.

Email

Submit
No, I would not like to sign up for Early Check

Would you like to join Early Check?

We can learn and improve Early Check by understanding why some parents choose no. Are you willing to tell us why?

[Survey with a couple of options and an open text]

You may be able to change your mind and decide to have Early Check screening later. Early Check screening is offered until your baby is 4 weeks old. You can come back to this Early Check research portal at any time.
You have a choice to get an extra Early Check screening result on your baby.

You recently signed up for the Early Check research study. You gave permission for your baby to be screened for two rare health problems: spinal muscular atrophy and fragile X syndrome.

The Early Check screening test for fragile X syndrome also gives extra results about a milder form called fragile X premutation. Early Check will know if a baby has fragile X premutation through the same screening test used for fragile X syndrome. You can now choose whether to get this extra screening result for your baby. Every parent in Early Check is given this choice.

To get the extra result, you need to read the following 6 screens and decide whether you want this extra information on your baby. It is up to you to decide.

To get the extra result you have to sign up at the end.
Example ad concepts that are being tested to use in Phases 2-4

You Can Make a Difference

You can help your family and other families in North Carolina by joining Early Check, a newborn screening research study. By joining, you’ll help advance science, help doctors find treatments for rare disorders, and improve the lives of infants affected by rare disorders.

Early Check
Screening health awareness for your baby

EarlyCheck
Screening health awareness for your baby

Anything for My Baby

I want to make sure that my baby has the opportunity to live a healthy and happy life. The Early Check research study uses blood already collected from my baby to test for rare conditions. By caching it early, babies with these conditions can be treated sooner, giving them a better opportunity for a healthier life.

A Few Tiny Drops Can Make a Difference

A few tiny drops of blood are already gathered from your baby through a heel prick at birth. When you agree to have a drop of your baby’s blood screened for extra conditions, you open up a world of possibilities for other babies and their families.
Follow up plans for screen-positive babies
2018 – 2019 activities

- Launch in June, 2018, test and improve Phase I recruitment strategies
- Provide confirmatory testing & counseling for screen positive babies
- Immediately refer babies with SMA to clinical care and partner with national SMA registry efforts
- Implement a telegenetic counseling program for families of infants with the *FMR1* premutation
- Implement an intensive early intervention program for infants with the *FMR1* full mutation
- Initiate long-term follow up for children and families
- Prepare to begin Phase II recruitment strategies after 6 months
- Continue to build partnerships with researchers and advocacy groups, likely preparing to add one or more conditions in 2019
Contact information

delivering the promise of science for global good

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www.earlycheck.org