



Early Check: A Partnership to Advance the Science and Practice of Newborn Screening

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2019 Public Health Leaders' Conference

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Early Check

Expanded health screening for your baby



RTI Center for Newborn Screening, Ethics, and Disability Studies



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RTI Center for Newborn Screening, Ethics, and Disability

Finding solutions for those affected by special health care needs



The newest statistics show that approximately 10.2 million children in the U.S.—15 percent of all people under the age of 18—have special health care needs, and more than a fifth of U.S. households with children have at least one child with special needs.

FOCUS AREAS



Our interdisciplinary team

- Psychology
- Medicine
- Chemistry
- Bioethics
- Genetic counseling
- Public health
- Early childhood special education
- Molecular microbiology
- Family studies
- Neuroscience
- Nutrition
- Social Work



Don
Bailey



Melissa
Raspa



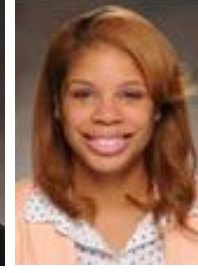
Lisa
Gektland



Holly
Peay



Anne
Wheeler



Jennifer
Taylor



Scott
Shone



Barbara
Biesecker



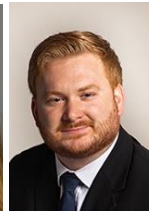
Anne
Edwards



Sara
Andrews



Stacey
Lee



Blake
Harper



Casey
Okoniewski



Kathi
Porter



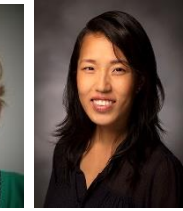
Martin
Duparc



Danielle
Toth



Beth
Boyea



Angela
Gwaltney

Newborn screening policy in the U.S.

- More than 4,000,000 babies each year
- 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
- 35 conditions on RUSP
- States are gradually implementing newly recommended conditions



Advisory Committee on Heritable Disorders in Newborns and Children

Many conditions will not make the RUSP in the near future

- Since the Committee was formed, 9 conditions have been reviewed but not initially recommended or sent to evidence review
 - SMA, Niemann-Pick, neonatal hyperbilirubinemia, Krabbe disease, hemoglobin H disease, Fabry disease, 22q11.2 deletion, GAMT, CTX
- Major reasons for not recommending
 - No accurate and cost-effective screening test
 - Implementation too complicated or expensive
 - No pilot study data
 - No data on pre-symptomatic treatment efficacy
- Frustrations are evident
 - Advocates think the pace of expansion is too slow
 - States think the pace of expansion is too fast

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

To help solve this problem, we designed 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 two prototypes:
 - Spinal muscular atrophy
 - Fragile X syndrome



Early Check

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Overarching goals for Early Check

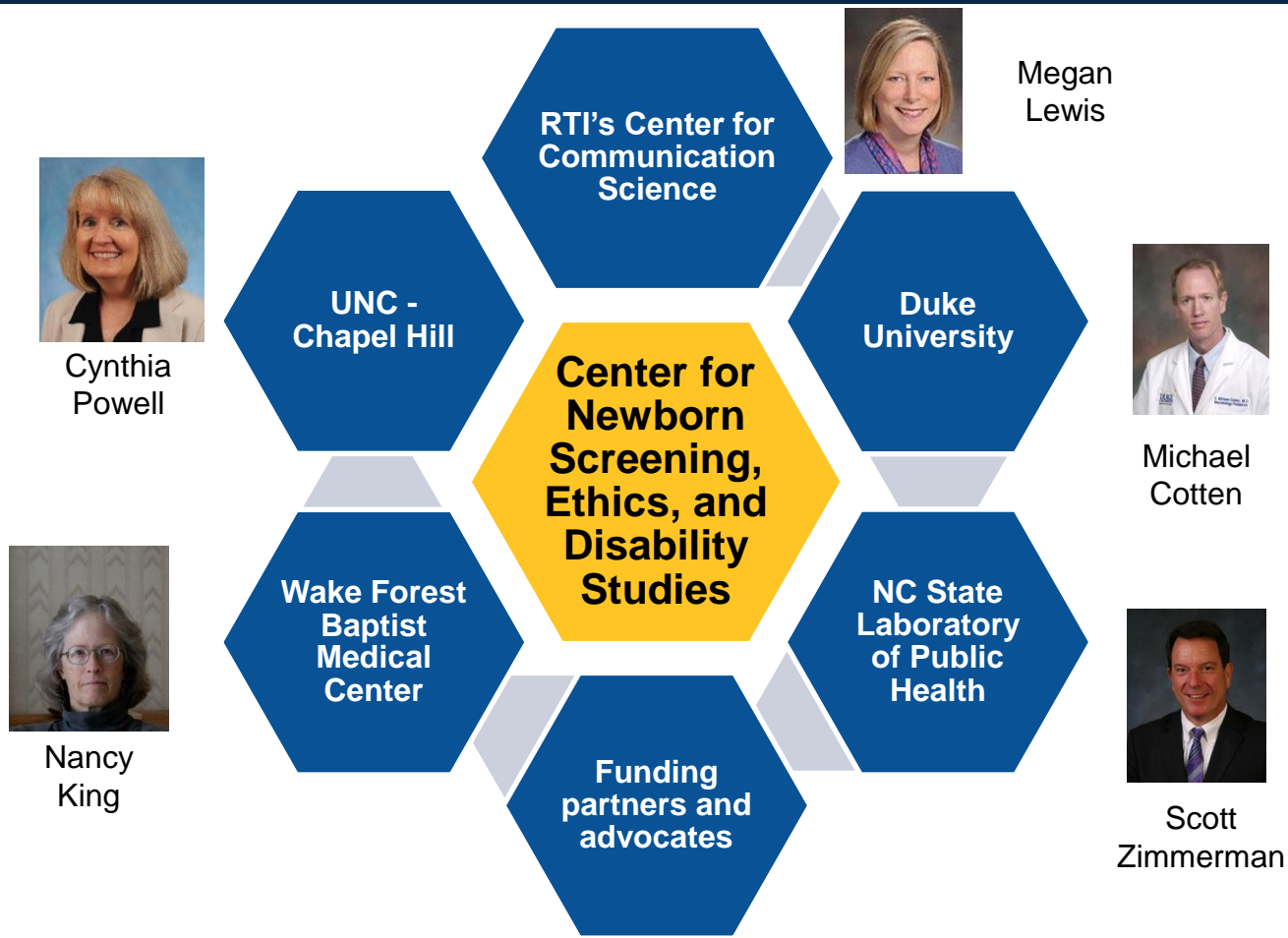
- Develop and evaluate methods to inform 120,000 parents/year about a research program and seek permission to participate in Early Check
- Accelerate the acquisition of data to support decisions about adding new conditions to the RUSP
 - Gauge parents' interests in screening for new conditions
 - Test potential for large-scale screening in partnership with 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



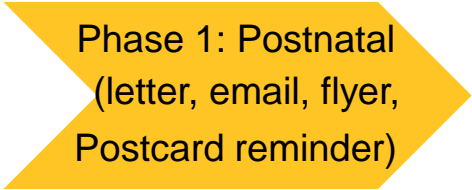
Unique features of Early Check

- Collaboration among multiple research partners
- Consortium of funders
- Statewide recruitment
- Systematic evaluation of recruitment methods
- E-consent
- Highly secure information management system
- Comprehensive program, including short and long-term follow-up, individually tailored to each disorder
- Telegenetic counseling
- Studying or supporting evidence of benefits and harms

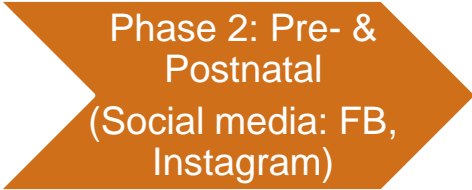
Center partners are critical to our success



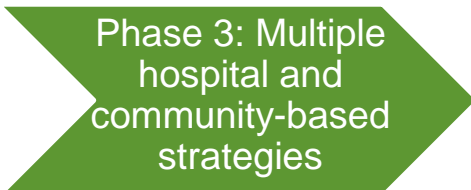
Phased studies of targeted outreach strategies



Phase 1: Postnatal
(letter, email, flyer,
Postcard reminder)



Phase 2: Pre- &
Postnatal
(Social media: FB,
Instagram)



Phase 3: Multiple
hospital and
community-based
strategies

300-400 infants are born each day in NC. Blood spots drawn, demographic data are entered, and cards are shipped to NCSLPH



STATE LABORATORY OF PUBLIC HEALTH
4312 DISTRICT DRIVE
1918 MAIL SERVICE CENTER
RALEIGH NC 27699-1918

DO NOT WRITE HERE

041180837 SN

NEWBORN'S MEDICAL RECORD # _____

FIRST TEST REPEAT UNSAT REPEAT OTHER KNOWN PKU

NEWBORN'S LAST NAME _____ FIRST NAME _____ MULTIPLE BIRTHS:
(if multiple, A, B, or C, etc.) _____

NEWBORN'S BIRTHDATE: MO DAY YEAR BIRTHTIME: 24 HR CLOCK RACE: SEX: TYPE OF FEEDING: BIRTHWEIGHT IN GRAMS

1. White 1. MALE 1. Breast 3. Non-Soy _____
 2. Black 2. FEMALE 2. Soy 4. Parenteral _____
 3. Am. Ind. 3. AMBIGUOUS NICU Y N _____
 4. Asian HISPANIC OR LATINO ORIGIN MECONIUM ILEUS Y N _____
 5. Native Hawaiian/ Other Pacific Is. 1. Yes 2. No U _____
 6. Unknown 3. Unknown

COLLECTION DATE: MO DAY YEAR COLLECTION TIME: 24 HR CLOCK GESTATIONAL AGE (WKS) CURRENT WEIGHT IN GRAMS

MOTHER'S LAST NAME _____ FIRST NAME _____ LATEST RBC TRANSFUSION DATE: MO DAY YEAR TRANSFUSION TIME: 24 HR

MOTHER'S MAIDEN NAME _____ AREA CODE _____ MOTHER'S PHONE _____

MOTHER'S EMAIL ADDRESS _____ AREA CODE _____ MOTHER'S CELL PHONE _____

MOTHER'S MAILING ADDRESS _____

CITY _____ ST _____ ZIP _____ COUNTY OF RESIDENCE / COUNTY CODE _____

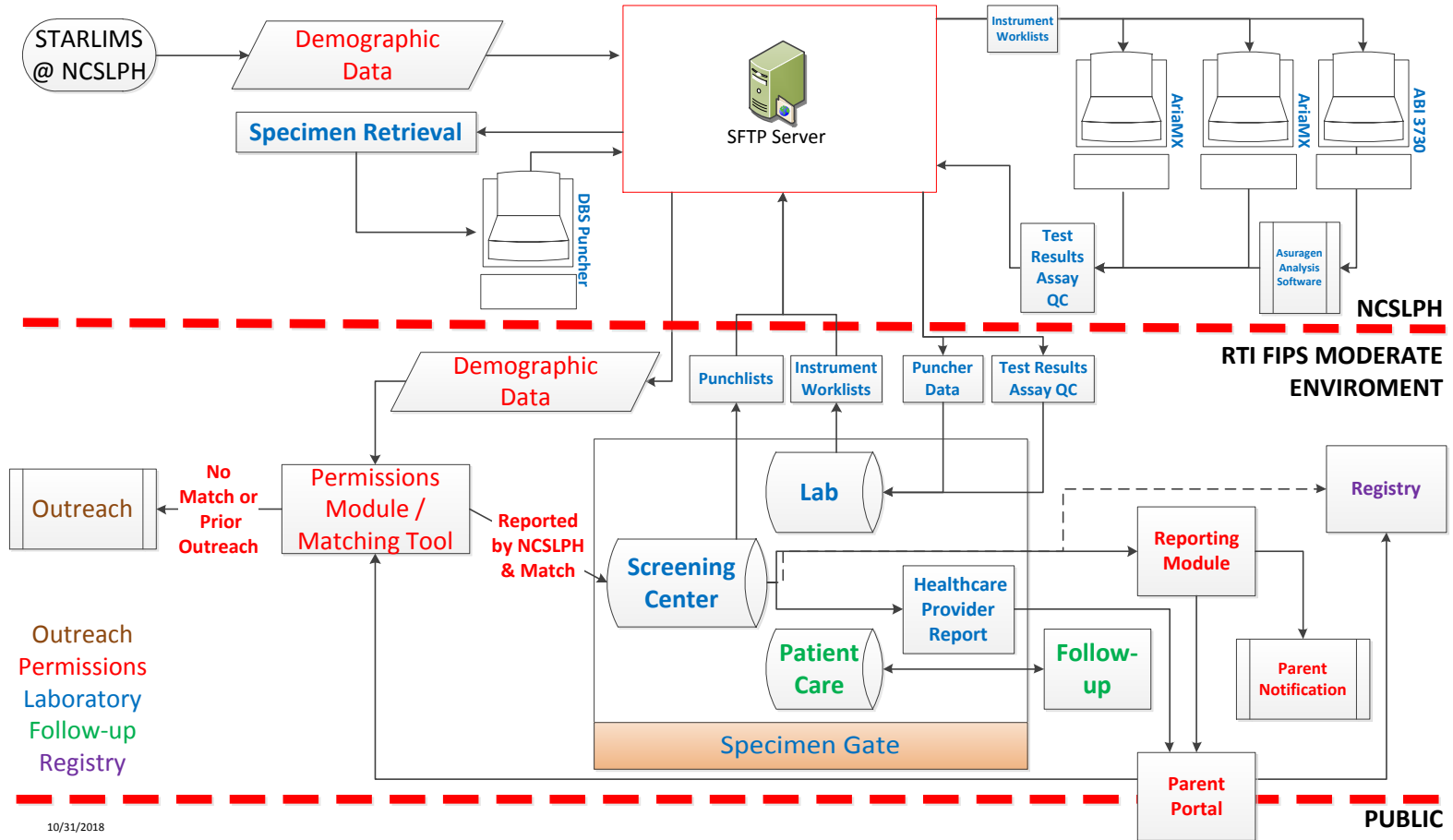
HOSPITAL/SPECIMEN SUBMITTER _____ FED. TAX ID _____

PHYSICIAN / PRACTICE NAME _____ FED. TAX ID _____

ORDERING PHYSICIAN LAST NAME _____ FIRST NAME _____ ORDERING PHYSICIAN NPI _____

DHHS 3105 (REVISED 10/17) VER 4.00, LABORATORY REVIEW 10/17 COLLECTOR'S INITIALS, PRINT LEGIBLY: _____

Informatics – Data Flow and Security



Postnatal letter and brochure are mailed within 5 days after birth



DEPARTMENT OF HEALTH AND HUMAN SERVICES
DIVISION OF PUBLIC HEALTH

ROY COOPER
GOVERNOR

MANDY COHEN, MD, MPH
SECRETARY

DANIEL STALEY
DIRECTOR

Date

Dear (name here),

We are sending you this letter because you recently had a baby in North Carolina. Congratulations!

We are inviting you to join a research study called Early Check. All new parents in North Carolina are being invited. You can sign up for Early Check until your baby is 4 weeks old.

The goal of Early Check is to test babies for rare health problems and look for better treatments. Your participation in Early Check is important in helping the study achieve this goal. To do this, Early Check will provide extra, free tests for your baby beyond those that are part of regular newborn screening. You can learn more about Early Check and the two health problems screened by Early Check. Visit www.EarlyCheck.org.

Early Check uses the same blood sample from the baby's heel already taken in the hospital for regular newborn screening. Newborn screening is done by the North Carolina State Laboratory of Public Health (NCSLPH) for all babies born in the state. NCSLPH has completed newborn screening for your baby or will soon. The NCSLPH is working with the University of North Carolina at Chapel Hill, Wake Forest School of Medicine, Duke University, and RTI International to make the Early Check study possible.

- The Early Check study is different from regular newborn screening done by NCSLPH. Early Check tests for other health problems not included in regular newborn screening.
- If you join the study, you agree to let Early Check do other tests on your baby's blood sample that was already taken in the hospital. Early Check will only use your baby's blood sample if you agree. It is your choice.
- IN the very rare event that your baby tests positive for an Early Check condition and has also screened positive for a regular newborn screening condition, Early Check staff will work with NCSLPH to notify you.

If you decide not to join Early Check, your baby will still have regular newborn screening and follow up. Your baby's doctor will still receive the results of regular newborn screening. We know that new parents are busy enjoying their newborns. Signing up for Early Check is easy. The enclosed brochure provides more details. To learn more and to sign up, visit www.EarlyCheck.org.

Sincerely,

Daniel Staley, Director



Early Check
Expanded health screening for your baby



What is Early Check?

- Early Check is a research study to check newborns for rare but serious health problems. You can learn about the two health problems Early Check tests for by visiting the link below.
- The free Early Check test allows us to find newborns with these health problems before symptoms start.
- The goal of Early Check is to learn about the health problems and look for better treatments to help babies that have them.

How is Early Check Done?

- Early Check does extra, free tests on the newborn screening blood sample that was already taken in the hospital using a heel prick.
- You do not need to take your baby anywhere to be a part of Early Check. There's nothing else Early Check needs from you or your baby to do the tests. However, you need to give permission for Early Check to do this testing.

Who can join Early Check?

- You can join Early Check if
 - ✓ You're more than 12 weeks pregnant, or
 - ✓ You have a baby less than 4 weeks old who was born in North Carolina.
- If you choose not to join Early Check, your baby will still have newborn screening and follow up just like other babies.

What happens after the test?

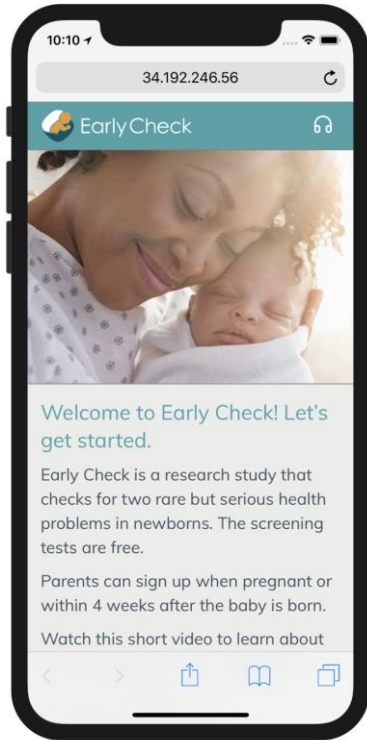
- Early Check will contact you with the results. Most babies will get a reassuring normal result.
- If we find a baby with one of the rare health problems, Early Check will give information, counseling, and recommendations for medical care.

You must sign up before your baby is 4 weeks old to be a part of Early Check.

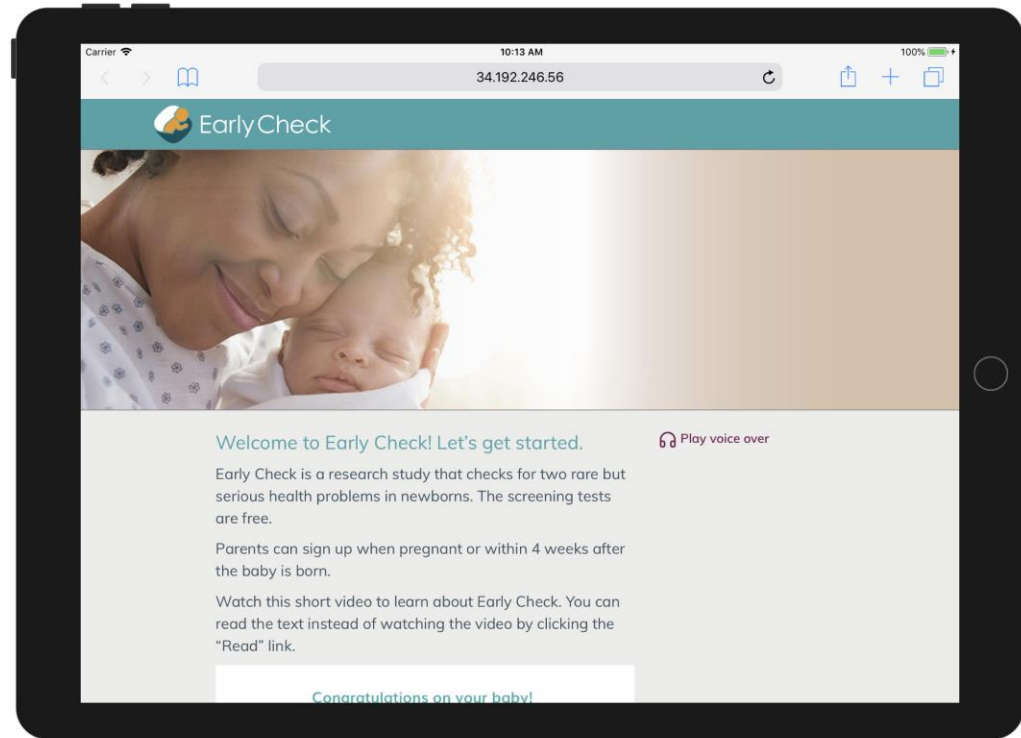
Visit portal.EarlyCheck.org to sign up and to learn more about Early Check.

Early Check is made possible by a partnership between The North Carolina State Division of Public Health, the University of North Carolina at Chapel Hill, Wake Forest School of Medicine, Duke University, and RTI International, a non-profit research institute.

The “enroll” button on the Early Check web site (www.earlycheck.org) takes parents to the permissions portal



iPhone X - 11.3



iPad Pro (10.5-inch) - 11.3

How is Early Check done?

 Play voice over

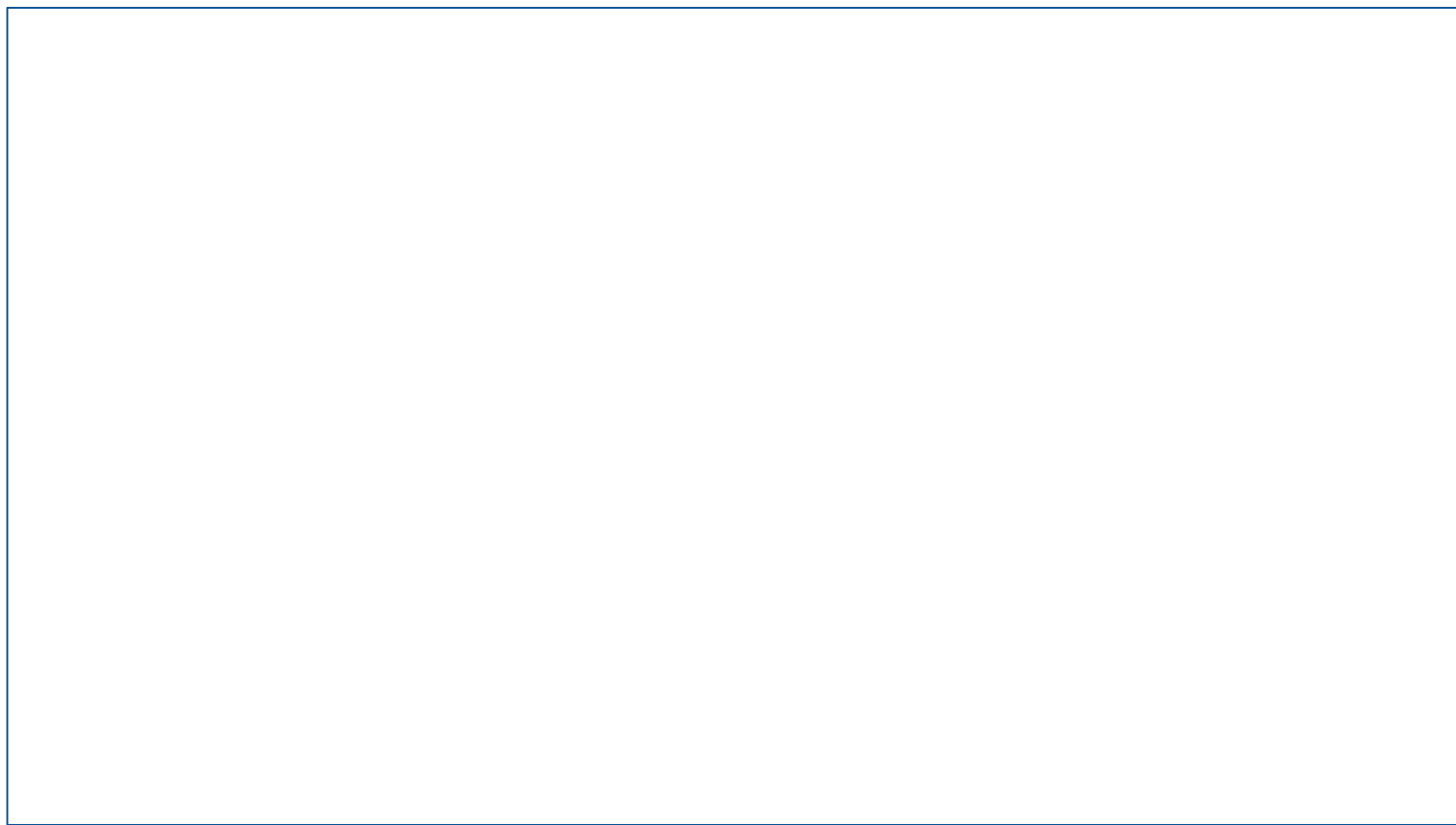
Watch this second short video about Early Check.

Click “play” to begin. You can read the text instead of watching the video by clicking the 'Read' icon.

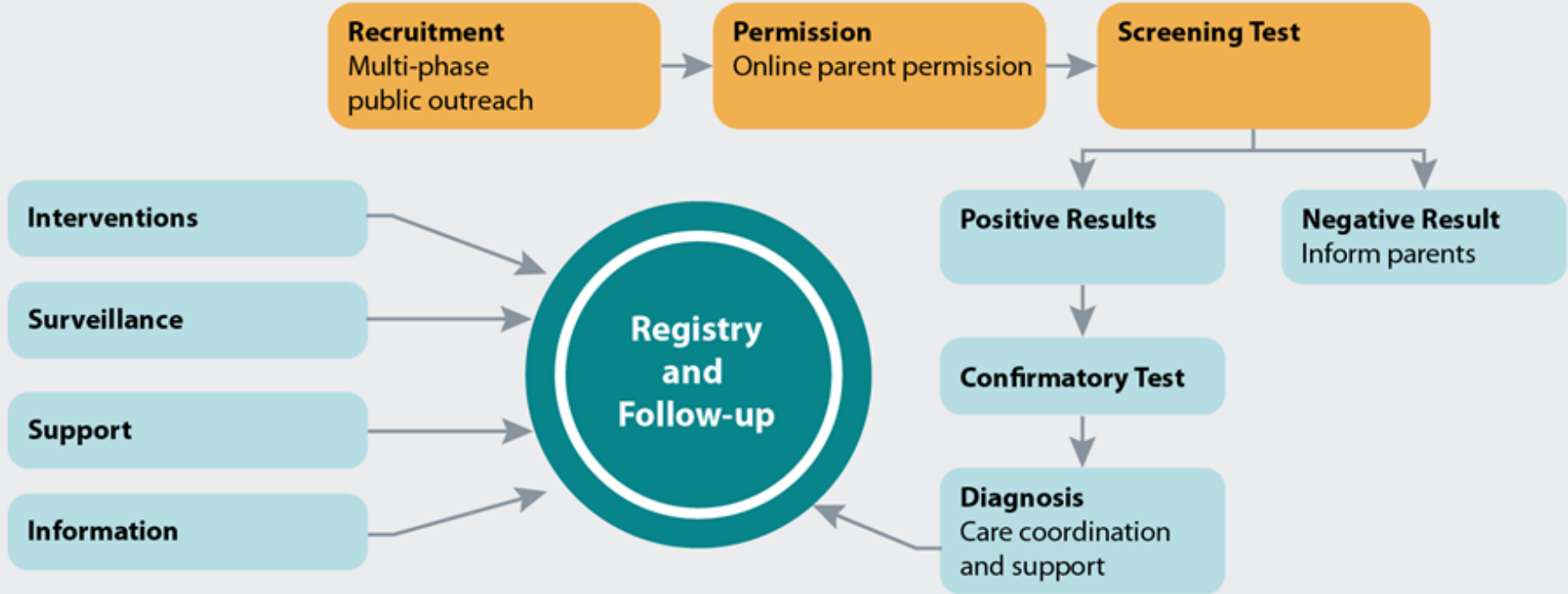
How is Early Check screening done?



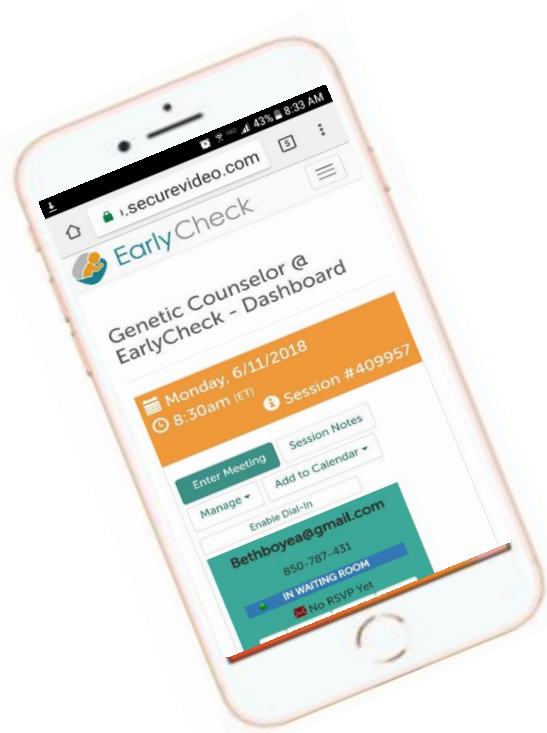
Read



Follow up plans for screen-positive babies



HIPAA-compliant videoconferencing for telegenetic counseling



- Uses top-rated Zoom platform
- Participants, their partners, and certified medical translator can join genetic counseling session on smartphones or computers from multiple locations.
- Counselor can share visual aids
- Genetic counseling content tracking tool will allow parents to drive the order of the genetic counseling session while standardizing content across all participants.
- Allows for audio recording and secure data storage.

zoom



North Carolina State Newborn Screening Laboratory Partnership

- RTI International and SLPH have a contractual relationship that includes a Business Associate Agreement to ensure HIPAA compliance
- Together we have received funding to implement pilot studies and acquire equipment to screen for SCID (CDC), MPS I (NICHD), X-ALD (NICHD), SMA (NICHD)
- With parent consent, we access the existing residual dried blood spots for expanded testing



Scott
Zimmerman



Dee
Pettit

State Lab Role in Early Check research project

1. Contribute expertise based on NBS experience
2. Assess and monitor risk to traditional NBS
3. Mechanism to respond when non-RUSP conditions are proposed
4. Platform for pilot work to inform state policy
5. Provides scientific evidence for decision-making

State Lab Role in Early Check research project – continued

6. Assess strategies for communicating with parents
7. Establishes a partnership that could be expanded to broader research-based initiatives to support lab policy and practice
8. Enhance within-state visibility
9. Opportunity to provide national leadership

Special issue of *NCMJ* on Newborn Screening



- Published by the North Carolina Institute of Medicine
- Co-edited by Bailey and Zimmerman
- 9 articles/sidebars about NBS nationally and in NC
- Co-editor introduction focuses on the future of newborn screening
- <http://www.ncmedicaljournal.com/>

The Future of Early Check

- Our long-term goal is a lasting research program to which new disorders can be added when they are ready and funding is available.
 - When is a disorder “ready” for Early Check?
 - What is our risk tolerance for uncertainty of results, treatment potential, complicated follow-up, very large or very small incidence?
 - How can we keep a basic infrastructure but individualize work based on condition-specific needs/research questions
 - When should a disorder be taken off of Early Check?
- Ultimately Early Check will be successful if:
 - We can recruit enough families
 - We have a sustainable business model
 - We demonstrate benefit and objectivity
 - We are seen as a trusted source of data to inform newborn screening policy

Contact information

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www.earlycheck.org