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

Don Bailey, Ph.D.

Advisory Committee on Heritable Disorders in Newborns and Children

November 10, 2021

Early Check

Expanded health screening for your baby

Disclosures (current and recent)

Early Check and Screen Plus have much in common

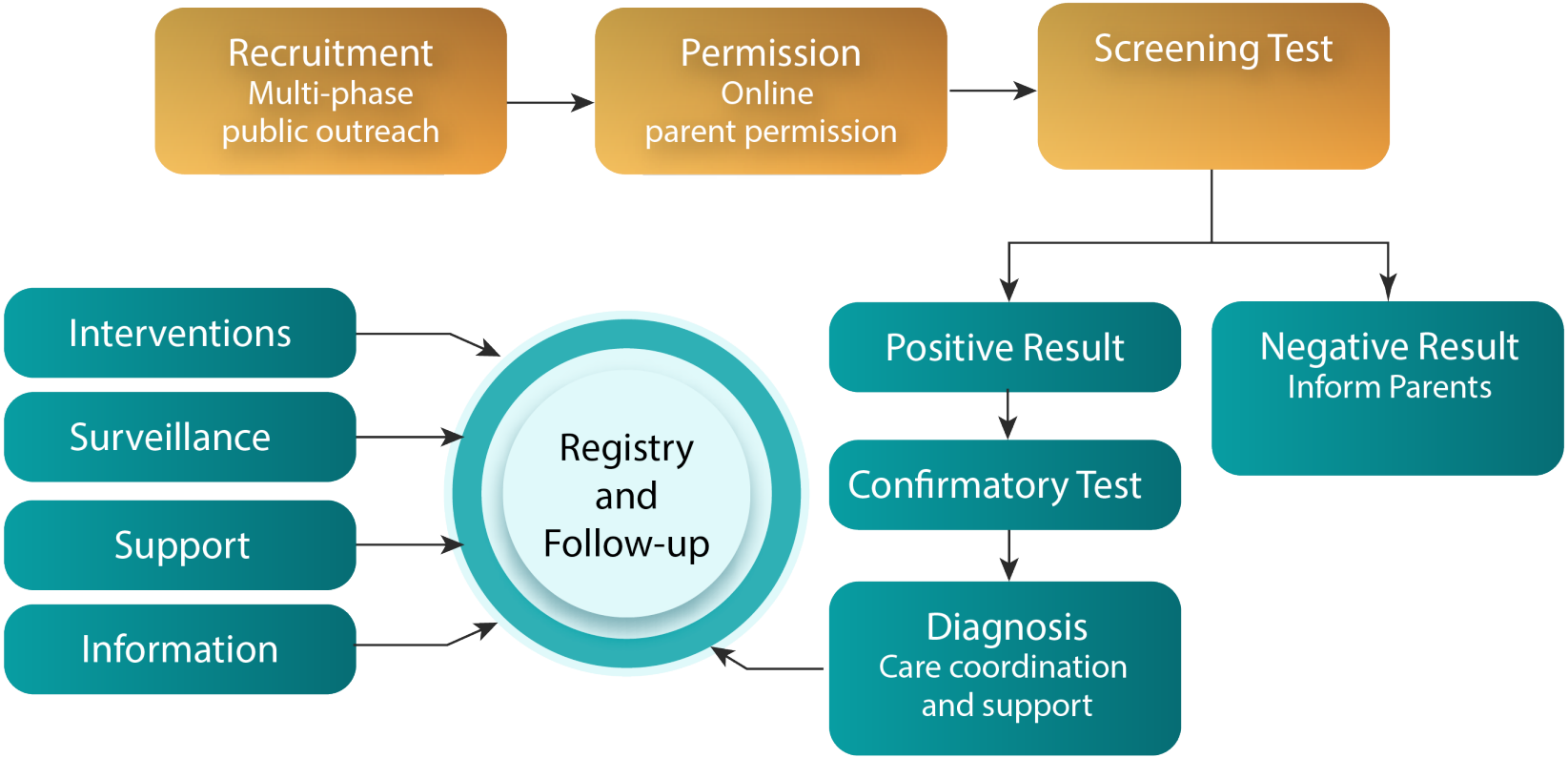
- Investigator-initiated projects
- Designed to advance NBS policy and practice
- Combine research with implementation studies, seen through a lens of public health ethics and respect for families
- Fill a gap in national capacity to gather policy-relevant data
- Multi-condition studies of disorders not yet on the RUSP
- Designed to be long-term, disease agnostic infrastructure resourse
- Funded by many different sources

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

What is Early Check?

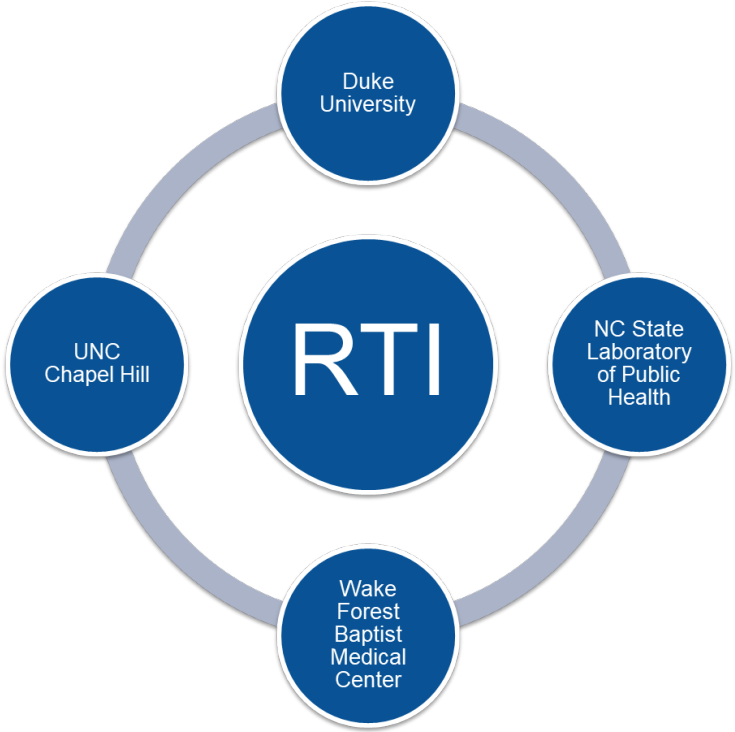
- An *Innovation Award* from NCATS, with additional support from NICHD, The John Merck Fund, Asuragen, Cure SMA, MDA, Sarepta
- A *research study* designed to
  - Develop and evaluate methods to offer free, voluntary screening to 120,000 parents/year for conditions not currently part of newborn screening (NBS)
  - SMA and FXS as initial prototypes, added DMD/CKMM screening in 2019
  - Acquire data to inform policy
- The *foundation* for
  - A long-term research resource to which new conditions can be added when ready
  - An envisioned future in which states offer a voluntary panel of “non-RUSP” conditions

Early Check Flow



Some unique features of Early Check

- Multi-institutional partnership integrated with public health and NBS
- Systematic formative work
- Use and evaluate virtual strategies for multiple system components
- Two-tiered consent for carrier results
- Screen using methods other than MS/MS
- Publish about laboratory methods
- Systems for tracking and evaluating everything from consent to follow-up
- Evaluation of early intervention



Lots of formative work

Contents lists available at ScienceDirect

Social Science & Medicine

journal homepage: www.elsevier.com/locate/socscimed

Parental intentions to enroll children in a voluntary expanded newborn screening program

Ryan S. Paquin [a, *], Holly L. Peay [b], Lisa M. Gehtland [b], Megan A. Lewis [a], Donald B. Bailey Jr. [c]

Parental preferences toward genomic sequencing for non-medically actionable conditions in children: a discrete-choice experiment

Megan A. Lewis, PhD[1], Alex Stine, BA[1], Ryan S. Paquin, PhD[1], Carol Mansfield, PhD[1], Dallas Wood, PhD[1], Christine Rini, PhD[2], Myra I. Roche, MS, CGC[3,4], Cynthia M. Powell, MD[3,4], Jonathan S. Berg, MD, PhD[4] and Donald B. Bailey Jr, PhD[1]

Genetics inMedicine

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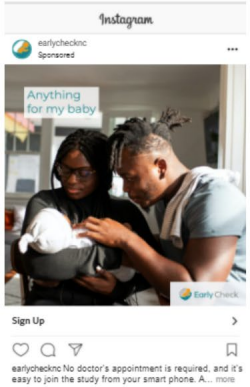
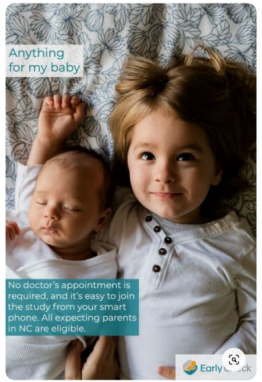


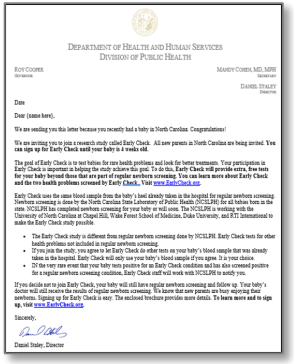


Use and evaluate virtual strategies for multiple system components

- Virtual recruitment
- E-consent
- Telegenetic counseling
- Family friendly web-based educational materials
- Virtual assessment
- Virtual intervention
- **As a result of these virtual strategies, we have been able to continue the project during the COVID pandemic**

Virtual recruitment methods

Postnatal letter/email



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 rare 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.

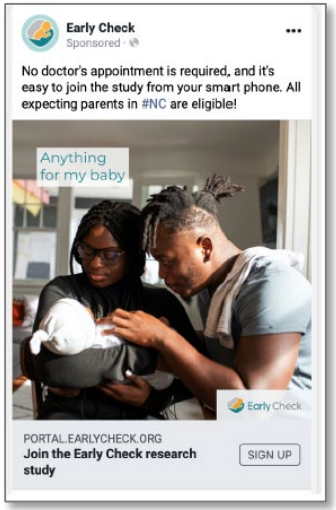
- The Early Check study is different from regular newborn screening done by NCSLPH. Early Check tests for other health problems not included in regular newborn screening.
- If you join the study, you agree to let Early Check do other tests on your baby's blood sample that was already taken in the hospital. Early Check will only use your baby's blood sample if you agree. It is your choice.
- It's 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

Social media ads



Information in health care settings

WIC NORTH CAROLINA

Patient portal invitations

myUNCChart™

Duke MyChart

myWakeHealth

In-person recruitment at Duke and University of North Carolina, started 2021

Evaluate and publish about virtual recruitment methods

Outreach to new mothers through direct mail and email: recruitment in the Early Check research study

Ryan S. Paquin[1] | Megan A. Lewis[1] | Blake A. Harper[1] | Rebecca R. Moultrie[1] | Angela Gwaltney[1] | Lisa M. Gehtland[1] | Holly L. Peay[1] | Martin Duparc[1] | Melissa Raspa[1] | Anne C. Wheeler[1] | Cynthia M. Powell[2] | Nancy M. P. King[3] | Scott M. Shone[4] | Donald B. Bailey Jr[1]

CTS Clinical and Translational Science
Open Access

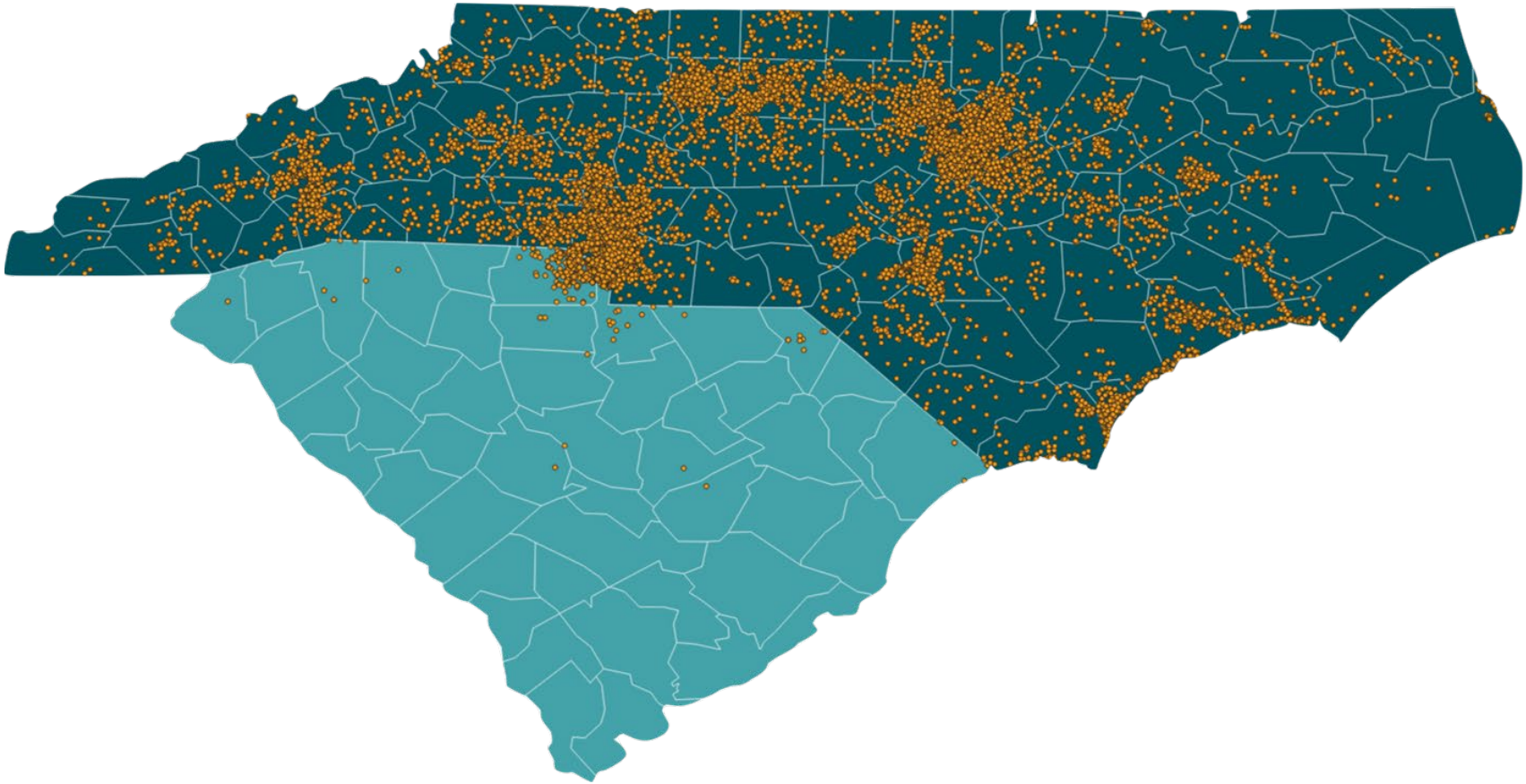
Using Social Media to Conduct Outreach and Recruitment for Expanded Newborn Screening

Jamie Guillory[1], Alyssa Jordan[2], Ryan S. Paquin[2], Jessica Pikowski[3], Stephanie McInnis[2], Amarachi Anakaraonye[2], Holly L. Peay[4] and Megan A. Lewis[2]*

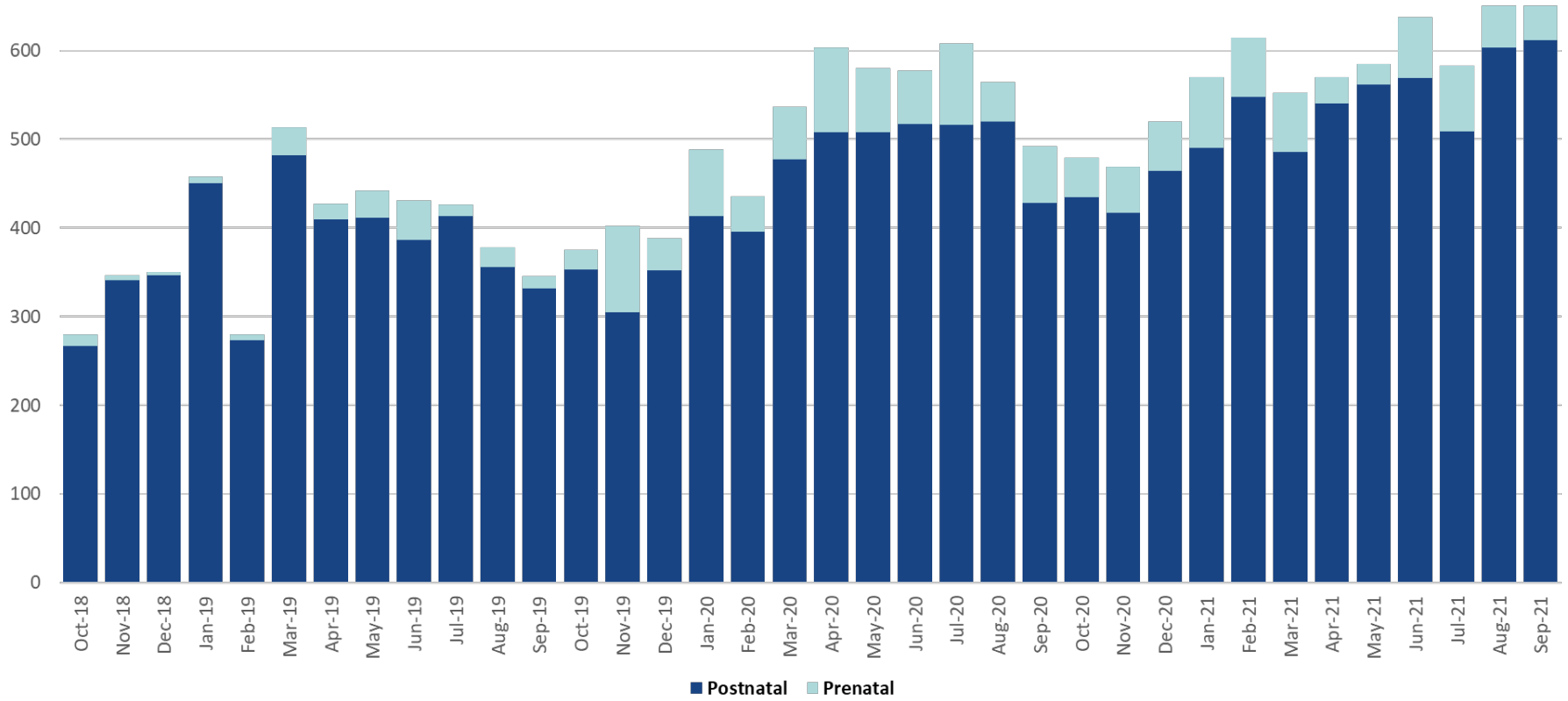
frontiers in Communication

- My Chart recruitment paper (*minor revisions submitted*)
- Expanded social media paper (*in progress*)
- Phone (and maybe text) reminder study (*begins soon*)

>18,000 enrollees from 100% of birthing hospitals and 99% of counties



Early Check consents by month

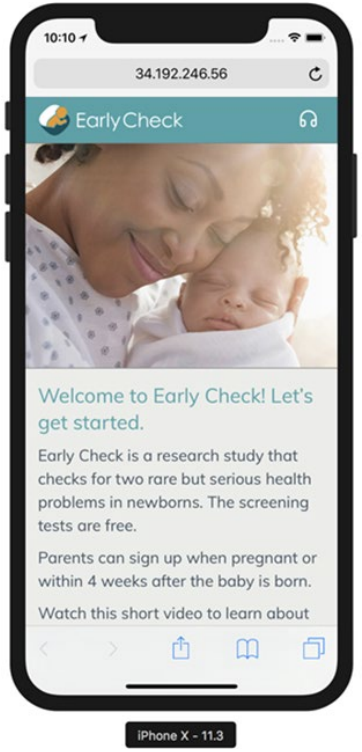


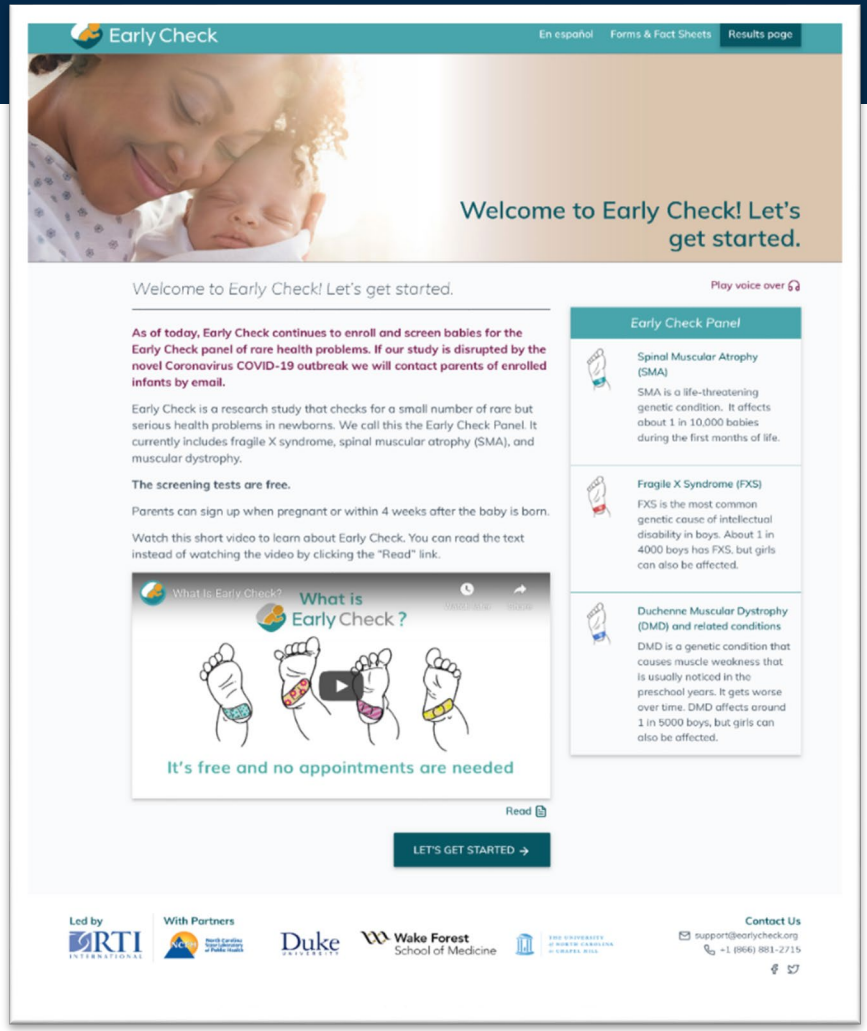
Self-reported race/ethnicity compared with NC population

Self-reported race/ethnicity	Study Percentage	NC 2020 Census
Not answered	7	-
White, non-Hispanic	57	63
Hispanic only	9	10
African American only	6	22
Asian only	8	3
Other or mixed (African American plus something else)	12 (6)	4

Electronic consent

Consent is obtained through an online permission portal





Telegenetic Counseling for Return of Screening Results

Technology

- HIPAA-compliant
- Multiple users can join on multiple devices (e.g., partner, interpreter)
- Screen sharing, multi-party document signing, A/V recording, provider note storage
- Convenient, user-friendly, easy self-service scheduling with automated reminders for all parties
- Parents appear to be at ease using online meeting platform from home while newborn sleeps nearby.

Educational Web Content

Studying and publishing about laboratory methods

THE JOURNAL OF PEDIATRICS • www.jpeds.com ORIGINAL ARTICLES

The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening

Jennifer L. Taylor, PhD[1], Kristin Clinard, MS, CGC[2], Cynthia M. Powell, MD[2], Catherine Rehder, PhD[3], Sarah P. Young, PhD[3], Deeksha Bali, PhD[3], Sara E. Beckloff, PhD[4], Lisa M. Gehtland, MD[1], Alex R. Kemper, MD, MPH, MS[5], Stacey Lee, PhD[1], David Millington, PhD[3], Hari S. Patel, MS[4], Scott M. Shone, PhD[1], Carol Woodell, BSPH[1], Scott J. Zimmerman, DrPH[4], Donald B. Bailey, Jr, PhD[1], and Joseph Muenzer, MD, PhD[2]

JAMA Network Open

Original Investigation | Pediatrics

Evaluation of X-Linked Adrenoleukodystrophy Newborn Screening in North Carolina

Stacey Lee, PhD; Kristin Clinard, MS, CGC; Sarah P. Young, PhD; Catherine W. Rehder, PhD; Zheng Fan, MD; Ali S. Calikoglu, MD; Deeksha S. Bali, PhD; Donald B. Bailey Jr, PhD; Lisa M. Gehtland, MD; David S. Millington, PhD; Hari S. Patel, MS; Sara E. Beckloff, PhD; Scott J. Zimmerman, DrPH; Cynthia M. Powell, MD; Jennifer L. Taylor, PhD

Validation of Fragile X Screening in the Newborn Population Using a Fit-for-Purpose *FMR1* PCR Assay System

Stacey Lee,* Jennifer L. Taylor,* Charles Redmond,† Andrew G. Hadd,† Jon A. Kemppainen,† Brian C. Haynes,† Scott Shone,* Donald B. Bailey, Jr,* and Gary J. Latham†

the Journal of Molecular Diagnostics

jmd.amjpathol.org

International Journal of *Neonatal Screening*

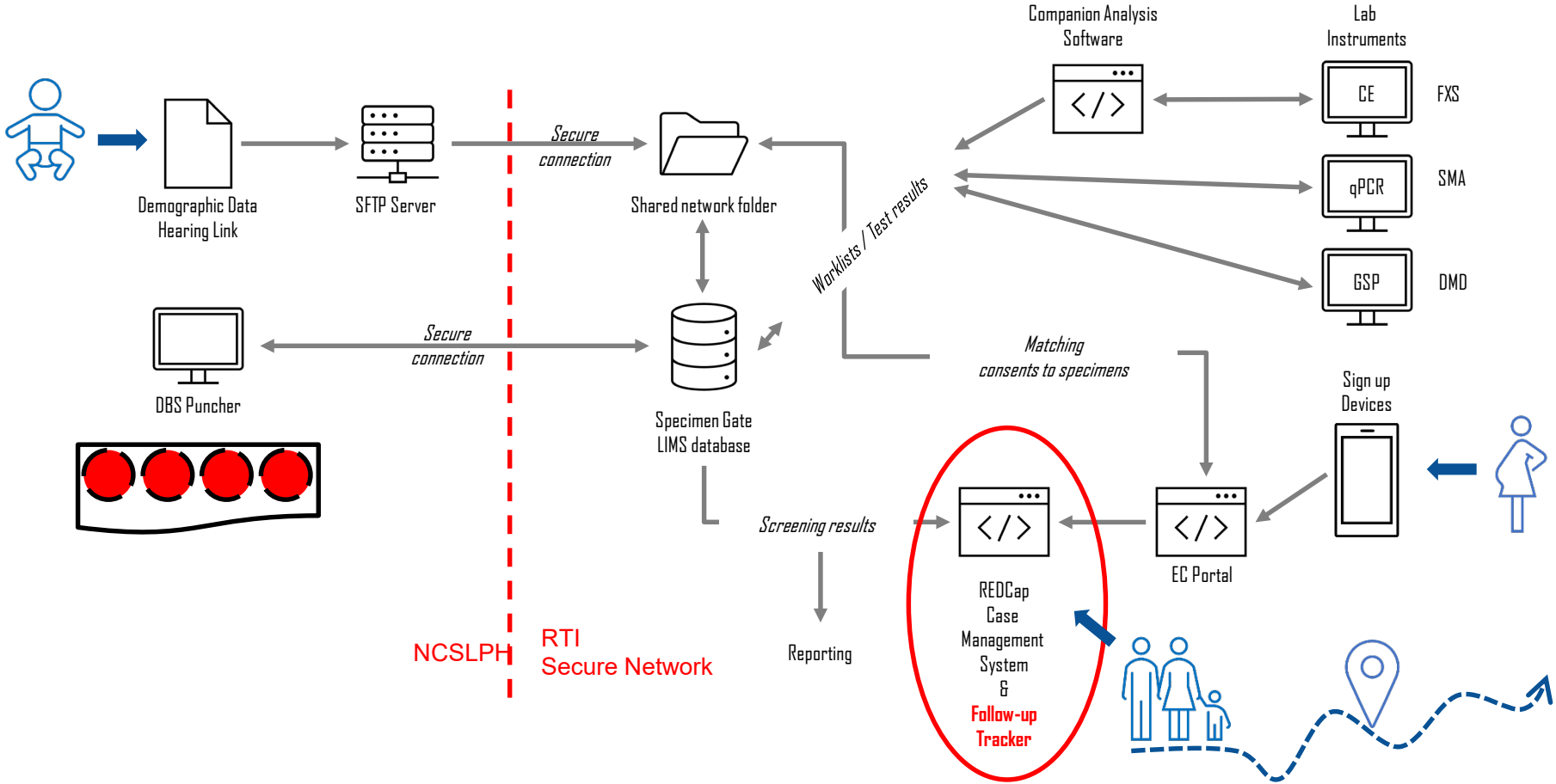
MDI

Article

A Voluntary Statewide Newborn Screening Pilot for Spinal Muscular Atrophy: Results from Early Check

Katerina S. Kucera [1,*], Jennifer L. Taylor [2], Veronica R. Robles [1], Kristin Clinard [3], Brooke Migliore [1], Beth Lincoln Boyea [1], Katherine C. Okoniewski [1], Martin Duparc [1], Catherine W. Rehder [4], Scott M. Shone [5], Zheng Fan [6], Melissa Raspa [1], Holly L. Peay [1], Anne C. Wheeler [1], Cynthia M. Powell [7], Donald B. Bailey, Jr. [1] and Lisa M. Gehtland [1]

Comprehensive data systems



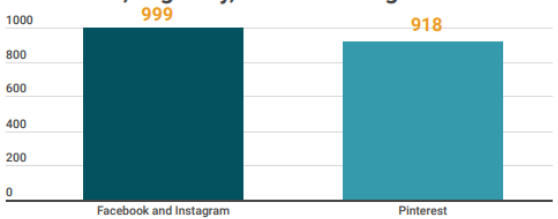
Monthly Social Media reports

Early Check - Social Media Paid Advertising

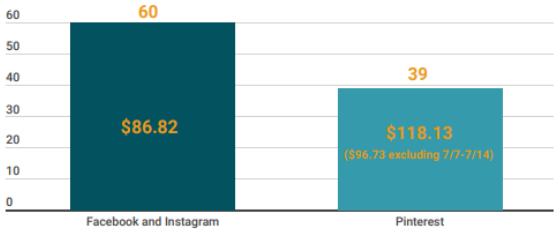
Pinterest: 7/7 - 8/4; Facebook and Instagram: 7/21-8/4

Facebook & Instagram Spend	Pinterest Spend	Facebook & Instagram Eligible	Pinterest Eligible	Facebook & Instagram Sign Ups	Pinterest Sign Ups
$5,209.28	$4,607.16	60	39	26	28

Link Clicks, Eligibility, & Cost Per Eligible



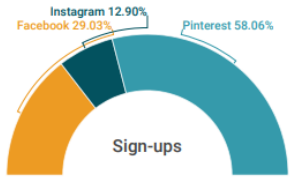
Eligibility & Cost Per Eligible



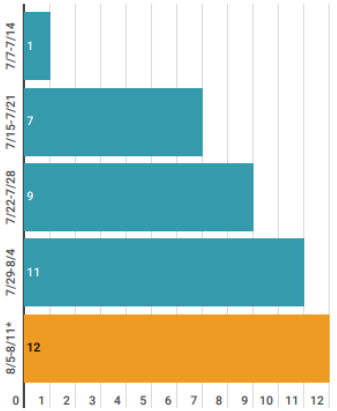
Performance by Platform

	Facebook	Instagram	Pinterest
$$ Spent	$4,223.97	$985.31	$4,607.16
Reach	58,336	20,808	172,904
Impressions	242,016	43,657	348,523
Clicks	886	113	918
Eligible	47	13	39
Complete	18	8	28*
Completion Rate	39.1%	61.5%	71.8%

Sign-ups by Platform



Pinterest Performance by Week



*We were not running ads for the week of 8/5-8/11

Early Check - Social Media Paid Advertising

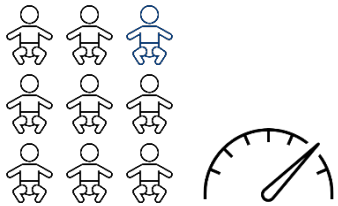
Pinterest: 7/7 - 8/4; Facebook and Instagram: 7/21-8/4

EC Follow-up Tracker and EC Dashboard

EC Follow-up Tracker

Visual interface with functionality to import and input data and track individual follow-up progress

- Automatic data import from multiple sources
- Daily use to track and document participant status



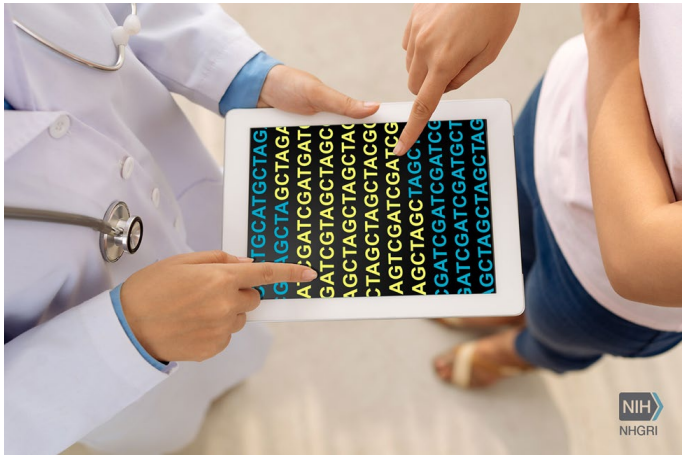
EC Dashboard

Visualization of current EC status - aggregated data

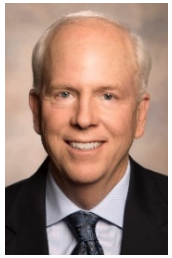
- Consent counts, screening counts for SMA, FX, DMD

Future directions

- Move from one disorder at a time to multiplexing a larger number of disorders
- Chromosome 15 disorders
  - Angelman syndrome
  - Prader-Willi syndrome
  - Dup 15q syndrome
- Large targeted sequencing panel
- Flexible systems that can respond quickly to new transformative therapies



The RTI Early Check team


Don Bailey


Melissa Raspa


Lisa Gehtland

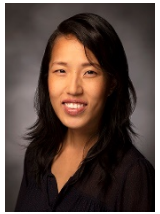

Holly Peay


Anne Wheeler


Kate Kucera


Barbara Biesecker


Casey Okoniewski


Angela Gwaltney


Anne Edwards

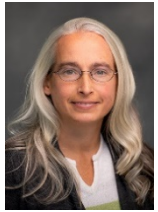

Sara Andrews


Kathi Porter


Martin Duparc


Beth Boyea


Veronica Robles


Brooke Migliore


Manisha Dass

Early Check partners

Cindy Powell

Michael Cotten

Eddie Smith

Nancy King

Scott Shone

Contact information

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for global good

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