# Early Check

*Expanded health screening for your baby*

## The Early Check Team

Early Check is being developed by investigators in the Center for Newborn Screening, Ethics, and Disability Studies at RTI International (www.rti.org), a nonprofit research institute with headquarters in Research Triangle Park, North Carolina. We are a multidisciplinary team with expertise in medical genetics, pediatrics, laboratory testing, psychology, genetic counseling, health communication, informed decision-making, ethics, family adaptation, registry development, special education, and child assessment. Our partners include the University of North Carolina at Chapel Hill, Duke University, Wake Forest Baptist Medical Center, and the North Carolina State Laboratory of Public Health.

**www.earlycheck.org**

## The Problem

Newborn screening (NBS) is designed for pre-symptomatic identification of conditions for which there are effective treatments that must begin early. The Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) provides guidance to state health departments for conditions to include in NBS—the Recommended Uniform Screening Panel (RUSP)—with four primary considerations: (1) the condition is a significant public health problem; (2) there is a cost-effective and accurate laboratory test; (3) treatments exist with proven efficacy; and (4) states are capable of screening and follow-up. Currently, 34 conditions are on the RUSP. Many other conditions fall short of the RUSP criteria but are of great interest to families and health care providers, who argue that early identification could result in benefits for both children and families. And as technological advances continue to bring down the cost of screening for rare disorders, advocates are increasingly asking, "Why not screen for them?"

Because most conditions nominated for NBS are rare, researchers have difficulty identifying enough babies to test the benefits of pre-symptomatic identification and treatment. As a result, the ACHDNC often lacks the information needed to make evidence-based recommendations. We need a way to study the benefits of NBS for rare disorders so that policy makers and state NBS programs can make good policy and practice decisions.

## Our Proposed Solution

We are addressing this gap by designing and implementing Early Check, a voluntary program in which screening for a carefully selected panel of conditions will be offered under a research protocol with parent permission. Systematic research and a scaled-up implementation effort across North Carolina will determine the best way to offer the program; demonstrate costs and benefits; and address the wide range of ethical, legal, and logistical challenges. If successful, Early Check could have a game-changing impact on state and national policy by accomplishing five major goals:

- Provide a choice for parents who want to know about other conditions
- Study early development of infants with rare conditions before symptoms occur
- Inform families about ongoing clinical trials they might want to consider
- Determine the benefits and costs of early identification and treatment
- Provide evidence to inform policy.

## Our Accomplishments

With support from The John Merck Fund and the National Center for Advancing Translational Sciences (NCATS), we are preparing for the launch of Early Check in 2018. Some major accomplishments to date include the following:

- A formal partnership with the North Carolina State Laboratory of Public Health
- Awards to support screening in North Carolina for severe combined immunodeficiency, mucopolysaccharidosis Type I, and X-linked adrenoleukodystrophy
- A survey of 1,000 new or expectant parents to determine intent to participate if offered
- A survey of pediatricians, obstetricians, family physicians, and nurses to determine their perspectives on a voluntary screening program
- Two awards to support Early Check development and implementation
  - A 5-year Innovation Award from NCATS to support the Early Check infrastructure
  - A 6-year award from The John Merck Fund to support adding fragile X syndrome to the Early Check panel.
- A model of permission in which parents can choose to participate in Early Check either prenatally or postnatally up to 1 month after birth
- A plan to provide comprehensive follow-up for newborns identified through Early Check and their families, to include genetic counseling, confirmatory testing, clinical evaluation, and connection with supportive services
- A consortium to sustain newborn screening research and activities through engagement with diagnostic and pharmaceutical companies, patient advocacy groups, and government agencies.

## Our Plan

Our goal is to offer Early Check to all birthing parents in North Carolina, approximately 120,000 babies each year. To reach this goal, we have completed initial planning and are currently building the infrastructure, with Early Check launch planned in 2018.

| Year | Primary Activities |
|---|---|
| 2015–2016 | Built stakeholder community, finalized partners, surveyed parents and professionals; determined screening protocols; developed plans for workflow, personnel, and informatics; developed framework for follow-up services; identified key research questions |
| 2017 | Prepare print and web-based materials, complete logic model and workflow plans; establish communication plan; finalize permission and electronic consent module; purchase equipment; finalize protocol for services and follow-up |
| 2018 | Launch program in North Carolina; identify screen-positive children; implement follow-up services, surveillance, and treatment studies |
| 2019 | Continue screening and follow-up in North Carolina; modify protocol based on lessons learned from Year 1 |
| 2020 | Continue screening program; refine as necessary; develop sustainability plans; consider addition of new conditions |
| 2020 and beyond | Sustain newborn screening partnership and collaboration to continue funding new innovative approaches to impacting newborn continuum of care |

## More Information

Visit www.earlycheck.org to learn more. To find out about other newborn screening activities and capabilities at RTI and our partner institutions, visit www.rti.org or contact the Center Director:

**Don Bailey, PhD, Distinguished Fellow**
Director, RTI Center for Newborn Screening, Ethics, and Disability Studies
RTI International
919.541.6488
dbailey@rti.org

RTI 9778 R2 0817

RTI INTERNATIONAL