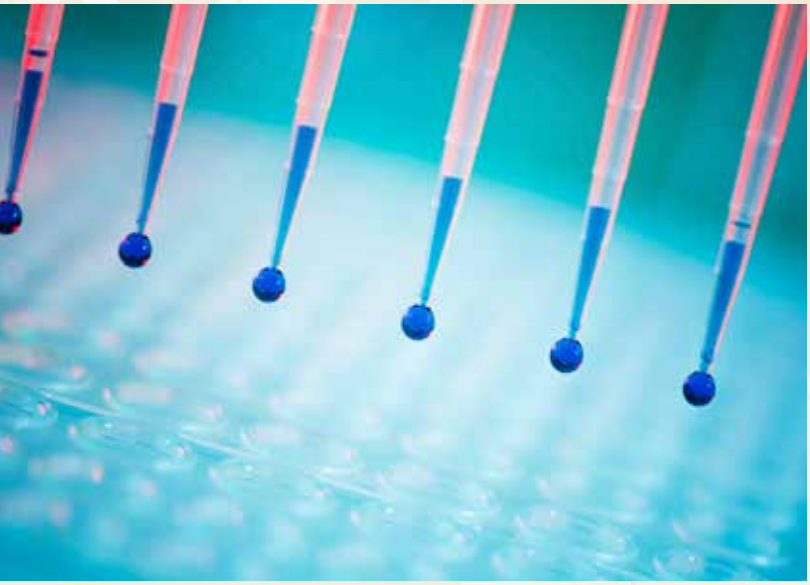
RTI Center for Newborn Screening, Ethics, and Disability Studies



The Center for Newborn Screening, Ethics, and Disability Studies at RTI International promotes early identification of adolescent special needs by conducting systematic research and evaluating program policies and practices. Our interdisciplinary team of investigators uses a wide range of methods to study the nature and impact of disabilities, develops solutions to complex ethical problems, understands the needs of families, and advances the science and practice of newborn screening.

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Overview

When children miss developmental milestones or begin to show health problems—such as seizures or muscle weakness—parents and health care professionals work together to find the cause. Sometimes a diagnosis is easy, but for many disorders—especially rare ones—diagnoses can take months or even years. Challenges associated with newborn health problems and diagnoses are the focus of the RTI Center for Newborn Screening, Ethics, and Disability Studies.

We appreciate the challenges faced by researchers, commercial companies, states, and policy makers who seek to understand and improve newborn screening as well as the lives of newborns and children diagnosed with disabilities. To address these needs, we have built an interdisciplinary team to advance newborn screening, ethics, and disability science and practice through research, implementation, and evaluation.

Capabilities

Our expertise spans the breadth of newborn screening, and our team includes experts in rare diseases, health communication, bioethics, family adaptation, newborn screening policy, psychology, public health, infant assessment, genetic counseling, laboratory testing, and biomedical informatics. This expertise allows us to

- Study family adaptation to a new or an unexpected diagnosis
- Investigate the ethical challenges associated with newborn screening and parent permission, facilitate meaningful decision-making, and create solutions for large-scale informed consent
- Develop novel, validated health communication tools via traditional, electronic, and social media channels, including electronic decision aids
- Implement statewide newborn screening pilot studies and assist laboratories with implementation of new screening technologies
- Develop rare disease registries and conduct surveys of patients and families to better understand the real-life consequences of disease
- Perform longitudinal studies of infants to better understand the natural history of a disorder
- Conduct formative and summative evaluations of projects focused on genetic conditions identified through newborn screening.

Experience

Issues, Policies, and Solutions

We identify potential benefits and challenges in newborn screening and suggest strategies to enhance or solve them. Working with partners in North Carolina, we are developing Early Check—a voluntary screening panel that would allow parents to choose additional screening for their baby beyond the standard newborn screening panel. Early Check will

- 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.

Registry Development

We design, implement, and maintain large-scale research registries. For example

- *Our Fragile X World*. We created an online survey registry of U.S. families with a child or children with fragile X syndrome. Over 1,700 families have participated in surveys since 2007.
- *DuchenneConnect*. In collaboration with Parent Project Muscular Dystrophy, we participate in research activities using a self-report registry that includes over 3,000 participants with Duchenne and Becker muscular dystrophy.

Survey Research

We have wide-ranging capabilities in survey research and a history of developing innovative technological solutions to modern survey challenges, including data management systems and the use of mobile survey technology in low-resource settings. Examples include

- An online survey of 1,000 recent and expectant parents to understand parental attitudes, preferences, and other factors impacting willingness to participate in a voluntary newborn screening program
- A discrete choice experiment to quantify how parental preferences for learning a child's genome-scale test results change in relation to health-condition characteristics
- A stated preference experiment exploring health care provider preferences for traditional and expanded newborn screening.

Family Adaptation, Parent Experiences, and Perceptions

We study the psychosocial consequences of health problems and disabilities for families, develop better methods to assess those consequences, examine caregiver opinions about screening, and examine the "diagnostic odyssey" experienced by many families. We also create tools to measure quality of life and family outcomes in rare diseases. One tool, *The Family Outcomes Survey*, has been translated into more than a dozen languages.

Pilot Studies and Laboratory Testing

We conduct newborn screening pilot studies on a large scale and have expertise in laboratory method development and implementation. Our projects include

- A recently completed study that offered newborn screening for fragile X syndrome to 28,000 families in three birthing hospitals
- Three current pilot studies to assist North Carolina in implementing screening for severe combined immunodeficiency, X-linked adrenoleukodystrophy, and mucopolysaccharidosis Type 1.

Ethics, Consent, and Informed Decision-Making

Our research includes studies of clinical trial decision-making, expectations and understanding of informed consent processes, and the ethical issues of genetic sequencing for newborn screening. Our work also includes the creation of innovative decision aids such as

- An electronic aid to support parents making decisions about the use of genomic sequencing in newborn screening
- A decision aid to help parents decide whether to participate in a study offering neonatal screening for fragile X syndrome.

Evaluation

We have extensive experience conducting evaluations of public health programs and projects that focus on genetic conditions identified through newborn screening. We also serve as an evaluator for the New York–Mid-Atlantic Consortium for Genetic and Newborn Screening Services, a regional collaborative that provides training and technical assistance to seven states and the District of Columbia.

Early Intervention

We study early intervention programs and the pathways by which early intervention supports positive family adaptation and child development. Our research includes modeling the impact of formal and informal supports for young children with disabilities and their families and as well as understanding the outcomes for families of children with disabilities participating in early intervention.

Health and Economic Consequences

We survey the impact of disability or chronic health conditions on parent health and financial burden, as well as the implications for extended family members.

Natural History Studies of Early Development

We examine the early development of children so that we have a good understanding of natural history, providing important baseline data for evaluating the effects of earlier identification. Our clinical psychology, early child development, and rare disease expertise allows us to study developmental trajectories and the implications of early identification for child development.

Informatics Solutions

We collaborate with experts and community stakeholders to gain consensus around newborn screening data standards, promoting the availability and use of newborn screening data in laboratory information management systems, electronic health records, and patient portals.

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More Information

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