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Newborn screening program aims to help transform treatments for genetic diseases detected at birth

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Within the first days of life, screening tests are performed on all newborns born in the U.S. to identify rare and often life-threatening medical conditions that are not apparent at birth. These newborn screening programs have been operating for decades, permitting doctors to intervene early and improve outcomes in potentially devastating conditions.

Now a pediatric research project plans to strategically expand the data collection linked to newborn screening results, aiming to open up broad opportunities to develop new ways to screen for and treat childhood diseases.

Many existing therapies for rare childhood diseases are seriously limited, while screening tests and treatments remain to be discovered for many other congenital disorders not currently included in newborn screening. At The Children's Hospital of Philadelphia, a research group is developing tools to store long-term clinical data on children with conditions picked up in the screening tests. The goal is to harness the power of numbers—using clinical data from many patients over years of their lives as a resource for researchers seeking new and better tests and treatments.

As biomedical knowledge and screening technology advance, more disorders have been added to those included in newborn screening, and

the list will continue to grow.

"Currently, newborn screening programs are primarily limited to a short-term focus," said project leader Peter S. White, M.D., director of the Center for Biomedical Informatics (CBMi) at The Children's Hospital of Philadelphia. "The programs screen for disorders in which early intervention is possible. If we can broaden the data capture to follow up children over a longer term, we can tap the potential to develop new medical tests and interventions for diseases that are not currently detectable or treatable."

Awarded this past October, the CBMi's project, the Long-Term Follow-Up Data Collection Tool, is part of an ongoing five-year award from the Newborn Screening Translational Research Network (NBSTRN) to the American College of Medical Genetics. The NBSTRN, in turn, is funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, part of the National Institutes of Health.

The subcontract award to Children's Hospital totals $1.8 million over three years.

A classic example of a disease detected in newborn screening is phenylketonuria (PKU), in which a mutated gene disables a patient's ability to process the amino acid phenylananine. Untreated, the excess amino acid causes severe mental retardation. But major diet restrictions, beginning in the first few weeks of life, allow near-normal development. Newborn screening programs have sharply reduced PKU-related mental retardation over the past four decades.

Over the years, more than 50 additional diseases have been added to the newborn screening list, including sickle cell disease and cystic fibrosis. If the initial screening flags a suspected disorder, healthcare providers order further tests to confirm or rule out the first result.

The data repository planned in this project will store long-term clinical records of patients who test positive for a disorder in the confirmatory test. Secure, centralized records will collect results of follow-up tests, disease complications, medications and treatment records over the years. To protect confidentiality, the clinical information will not include patient identities--but will provide invaluable clues to authorized researchers seeking to improve disease outcomes.

"Genetic diseases don't go away; a big question is what happens after the newborn period," said Michael J. Bennett, Ph.D., director of the Metabolic Disease Laboratory at Children's Hospital, who chaired a group in the National Academy of Clinical Biochemistry that developed national guidelines for follow-up testing of metabolic diseases detected in newborn screening. "Collecting data will allow researchers to monitor long-term outcomes and develop better strategies to diagnose and treat diseases."

A centralized database will allow scientists to detect patterns in the data that typically would not be discernible among the small number of cases followed in any single center. "Researchers will be better able to determine which treatments may yield better outcomes," Bennett said, adding that knowledge of genetic profiles may also allow investigators to modify treatment based on genetic background. Such treatments may include new gene therapies, enzyme replacement strategies, or developing drugs to act on targeted metabolic pathways.

White said that longer-term follow-up information in the repository may provide the potential to develop interventions for currently untreatable diseases, for example, spinal muscular atrophy, a complex and devastating neurological disorder that can be detected at birth, though not currently on the list of diseases included in newborn screening. "We envision that this data collective could become a transformative resource for biomedical research," he added.

Provided by Children's Hospital of Philadelphia