Survey of US parents highlights need for more awareness about newborn screening, cystic fibrosis

May 5 2024

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A national survey led by Ann & Robert H. Lurie Children's Hospital of Chicago found that parents have insufficient knowledge of newborn

screening in general and of cystic fibrosis (CF) in particular. Researchers asked specific questions about CF based on studies showing that initial CF follow-up visits after a positive newborn screening often occur after 4 weeks of age, which is later than the recommended timeframe for best outcomes.

Later follow-up is associated with worse nutrition in childhood, a predictor of long-term health in CF. Parents reported difficulties in understanding abnormal newborn screening results and uncertainty as to what to do next.

An abstract of the findings was presented at the Pediatric Academic Societies (PAS) meeting in Toronto, Canada, on May 5, 2024, by Ashley Hayes, MPH, from Lurie Children's.

"Although all U.S. States screen for at least 33 metabolic and genetic disorders using blood spots collected in the first days of life, we found that parents don't know enough about newborn screening and need more support from clinicians if their infant has an abnormal test result," said Marie Heffernan, Ph.D., lead author on the abstract and Survey Science Lead at Mary Ann & J. Milburn Smith Child Health Outcomes, Research and Evaluation Center, Stanley Manne Children's Research Institute at Lurie Children's, and Assistant Professor of Pediatrics at Northwestern University Feinberg School of Medicine.

"Our main message to parents is to know your baby's newborn screening results and follow-up immediately if the test is abnormal. Not all babies with an abnormal screening test have the disease that is identified, but timely treatment can make a huge difference for the baby's health, development and survival. This is certainly true if newborn screening identifies cystic fibrosis."

CF is a progressive genetic disorder that damages the lungs and digestive

system. Pre-symptomatic treatment of CF leads to better long term health, and is the overarching goal of its inclusion in newborn screening. However, CF diagnosis is often delayed or even missed, especially in Black/African American, Hispanic/Latine, or Asian newborns.

Delays in evaluation and initiation of treatment are associated with more lung disease during the first year of life and poorer growth that persists through early childhood.

In the survey of nearly 1,600 parents, most respondents had heard of CF prior to participating (79%), but about half did not know it was included in newborn screening tests (52%). Overall, just over half of parents reported that they knew which conditions are included in newborn screening tests (51%), that false-positive results were possible (58%), and that false-negative results also were possible (54%).

Among parents who had a child with a positive newborn screening test, most parents (75%) reported difficulty understanding the test results and 34% of parents did not feel supported by their child's health care team.

"To improve outcomes of conditions like cystic fibrosis that are included in newborn screening tests, stronger partnerships are needed between clinicians and new parents," said Susanna McColley, MD, senior author on the abstract and Scientific Director for Interdisciplinary Research Partnerships at Manne Research Institute at Lurie Children's, and Professor of Pediatrics in Pulmonary and Sleep Medicine at Northwestern University Feinberg School of Medicine.

"Our results also indicate the need for greater public awareness of newborn screening. Specifically, we need more awareness among providers and the public that infants of all racial and ethnic backgrounds can have cystic fibrosis."

Provided by Ann & Robert H. Lurie Children's Hospital of Chicago