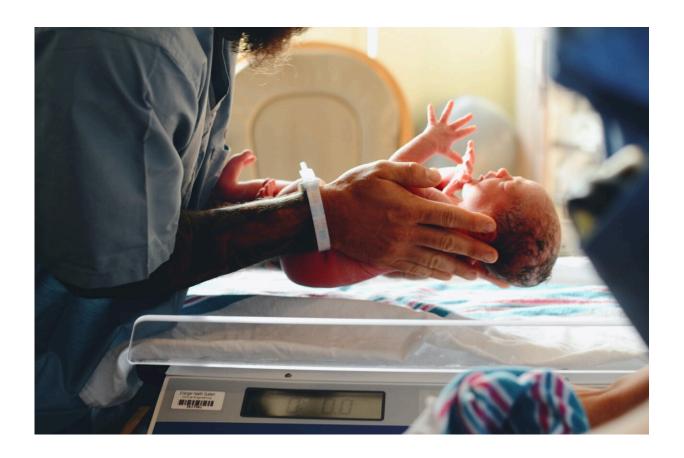


Study underscores importance of timely newborn screenings in early care for cystic fibrosis

July 31 2023



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Newborn screening for cystic fibrosis (CF) was fully implemented in all 50 states in the U.S. by 2010, but delays in timeliness of evaluation for



infants with positive newborn screen tests persist. Through evaluation of national patient registry data, Dr. Martiniano of Children's Hospital Colorado and her team determined that later initiation of CF care is associated with poorer long-term nutritional outcomes.

Specifically, a delay in initial care at a CF center from just 10 days of life to 47 days of life was associated with reduced weight-for-age through 1 year of age and reduced height-for-age through 5 years of age. The article, "Late Diagnosis in the Era of Universal Newborn Screening Negatively Effects Short- and Long-Term Growth and Health Outcomes in Infants with Cystic Fibrosis," has been published in *The Journal of Pediatrics*.

These are clinically important findings because early life weight-for-age and height-for-age are associated with later pulmonary function and survival.

"It is imperative that not only do we not miss diagnosing babies with CF, but that we also emphasize urgent referral for newborn screen follow-up for babies with an out-of-range screen, including sweat testing and referral for care at an accredited CF care center," said Martiniano.

Infants in the later cohort most commonly had a sweat test as their first CF evaluation and were more likely to have only one CF gene variant detected on commercially available panels used by most newborn screening programs. This may have caused a diminished sense of urgency to get the infants evaluated possible leading to delays in treatment based on a false assumption that two variants would be detected if the infant had CF.

"This will allow all babies with CF to be equitably started on treatments, with a goal before 1 month of age, to have improved long-term health outcomes," Martiniano said. "This is especially important today due to



emergence of ground-breaking, disease-modifying treatment available for infants and young children with CF."

CF is an inherited disorder that affects the lungs, digestive system and other organs in the body. The disorder disrupts the normal function of epithelial cells, which line passageways in the <u>respiratory tract</u>, digestive system, <u>sweat glands</u> and reproductive system. Because <u>cystic fibrosis</u> impacts so many organs, it can disrupt many of the body's essential functions.

More information: Stacey L. Martiniano et al, Late Diagnosis in the Era of Universal Newborn Screening Negatively Effects Short- and Long-Term Growth and Health Outcomes in Infants with Cystic Fibrosis, *The Journal of Pediatrics* (2023). DOI: 10.1016/j.jpeds.2023.113595

Provided by Children's Hospital Colorado

Citation: Study underscores importance of timely newborn screenings in early care for cystic fibrosis (2023, July 31) retrieved 28 April 2024 from https://medicalxpress.com/news/2023-07-underscores-importance-newborn-screenings-early.html

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