TuSalud

Cystic Fibrosis Is Underdiagnosed in Non-White Newborns

Researchers support equitable cystic fibrosis screenings for all racial groups.

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Newborn screenings for the genetic disorder cystic fibrosis (CF) are more likely to miss the detection of the condition among Latino, Black, Asian, American Indian and Alaska Native babies compared with white babies, according to a new study. This leaves children of color more vulnerable to irreversible lung damage and other harmful side effects.

In the study, which was published in Pediatric Pulmonology, Meghan McGarry, MD, associate professor of pediatrics at UC San Francisco (UCSF) Benioff Children's Hospitals and first author of the study, explained that while most states use genetic panels to screen for CF, these methods often test for genetic mutations in white populations and exclude mutations commonly found in non-white populations.

One of the most common genetic disorders, CF causes the body to overproduce mucus, sweat and digestive juices. These fluids become thick and sticky and can cause bronchitis and pneumonia and affect the lungs, pancreas and other vital organs.

"Newborn screenings are meant to be a public health measure that is equal across populations, but in practice, we are actually creating disparities because children of color are going undiagnosed for cystic fibrosis until an older age," McGarry said in a USCF news release. "That means they are treated later when symptoms occur, and their outcomes are often worse."

To detect CF, medical professionals typically screen for at least one variant of the cystic fibrosis transmembrane conductance regulator (CFTR). Using this method, study authors analyzed 46,729 people and found that Asian patients had the lowest detection rate (56% to 77%), followed by Black patients (73% to 86%), American Indian and Alaska Native patients (84% to 91%) and Latino patients (81% to 94%). White patients had a detection rate of 95% to 97%.

Study authors emphasize that without early diagnosis and treatment, CF can lead to serious lung damage that can be life-threatening.

"Most white kids with cystic fibrosis nowadays are never hospitalized; for them, it's an outpatient

disease and they will likely live a full life. If you are not white, however, you are more likely to be the one in the hospital all the time with a severe disease," McGarry said. "In order for newborn screenings to be equitable, they must include CFTR variant panels that reflect the racial and ethnic diversity of the population."

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