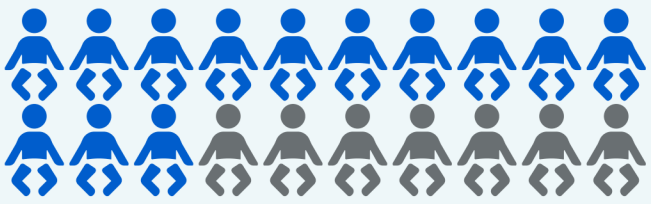
CYSTIC FIBROSIS NEWBORN SCREENING

IMPORTANT FINDINGS FOR PUBLIC HEALTH PROFESSIONALS

Cystic fibrosis (CF) newborn screening (NBS) has been performed throughout the United States since 2010. CF newborn screening has led to early diagnosis in most affected infants and improves health outcomes. A summary of evaluations on the first decade of universal NBS is below.

CF NEWBORN SCREENING LEADS TO EARLY DIAGNOSIS IN MANY AFFECTED INFANTS.

65% of infants with a positive NBS test had an age of first event* within the first 30 days, as recommended by guidelines, allowing for earlier diagnosis and intervention. [2, 4]



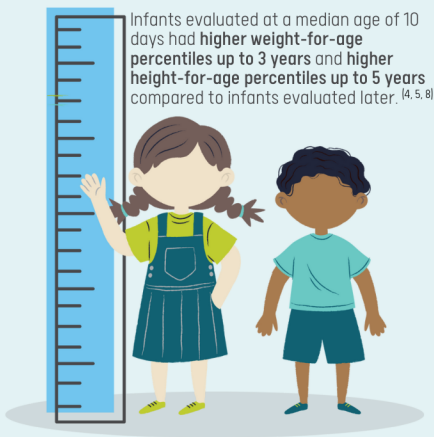
** surrogate measure for timeliness inclusive of sweat test, clinic visit, and/or hospitalization*

1 OF EVERY 8

infants with CF born between 2010-2018 were evaluated ***after two months.***

EARLIER EVALUATION FOR CF IS ASSOCIATED WITH BETTER EARLY LIFE NUTRITION.

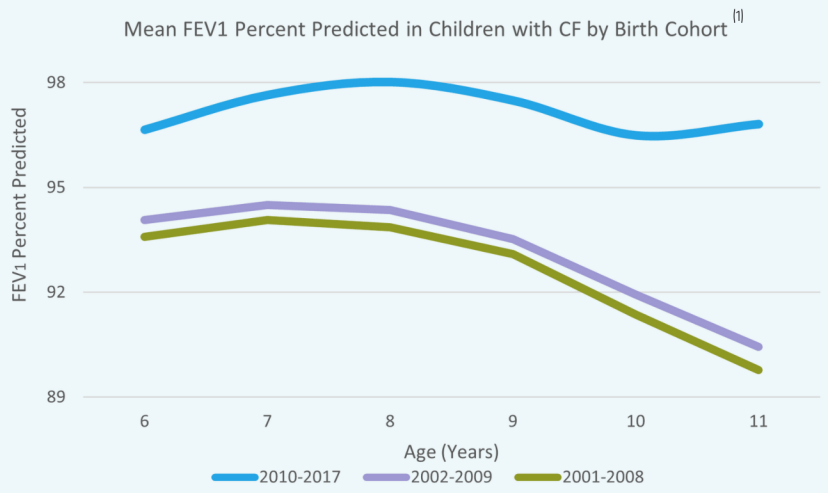
Infants evaluated at a median age of 10 days had **higher weight-for-age percentiles up to 3 years** and **higher height-for-age percentiles up to 5 years** compared to infants evaluated later. [4, 5, 8]



Why is this important?

Better early life nutrition is associated with improved lung function and survival. [3, 4, 5, 8, 9, 11]

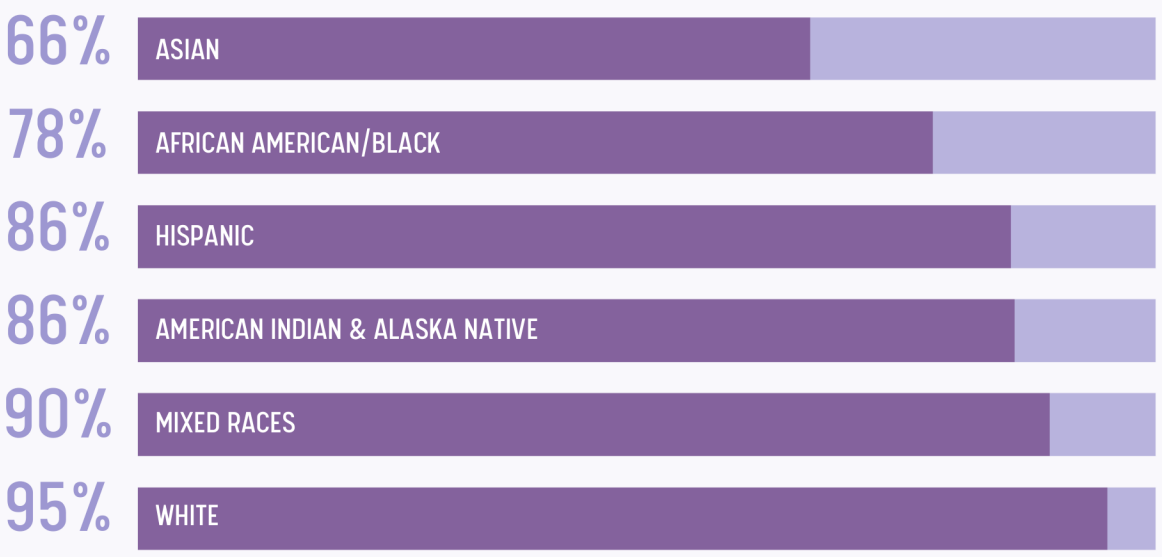
LUNG FUNCTION HAS IMPROVED SINCE NBS IMPLEMENTATION IN 2010.



DETECTION OF CFTR VARIANTS VARIES ACROSS RACE AND ETHNICITY

Since 2020, all U.S. Cystic Fibrosis NBS programs have used CFTR gene variant detection. Detection of **at least one** variant defines a positive test and requires follow-up. Some states detect more variants than others, but all miss more infants from non-white races and Hispanic ethnicity. [6,7]

AVERAGE DETECTION OF 1 CFTR VARIANT ACROSS 9 COMMON VARIANT PANELS BY RACE AND ETHNICITY



DELAYED DIAGNOSIS AND FALSE NEGATIVE NBS CONTINUE TO IMPACT PEOPLE WITH CF.

11% of people with CF are estimated to have experienced a delayed diagnosis (>180 days) or been diagnosed after a false-negative NBS. [1]

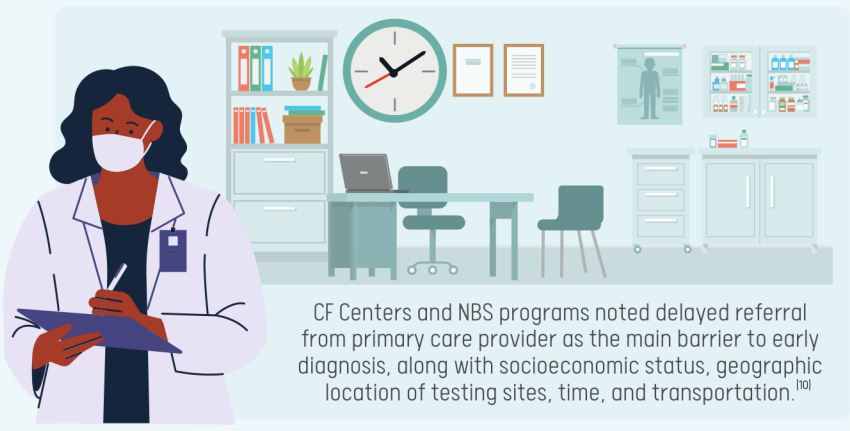
BLACK/AFRICAN AMERICAN & ASIAN

POPULATIONS EXPERIENCE THE HIGHEST RATE OF FALSE-NEGATIVE NBS *OR* DELAYED DIAGNOSIS AMONG NON-WHITE POPULATIONS. [1]

REFERRAL FOR EVALUATION AND TREATMENT SHOULD NOT BE DELAYED IN INFANTS WITH A POSITIVE NBS TEST.

40% of infants had weight-for-age percentiles below the 10th percentile at first CF Center visit from 2010-2018. [4]

CF CENTER & NBS PROGRAM-REPORTED BARRIERS



CF Centers and NBS programs noted delayed referral from primary care provider as the main barrier to early diagnosis, along with socioeconomic status, geographic location of testing sites, time, and transportation. [10]

1 FALSE NEGATIVE CAN HAPPEN FOR ALL INFANTS, EVEN WITH ELEVATED IRT OR DETECTION OF CFTR VARIANTS.

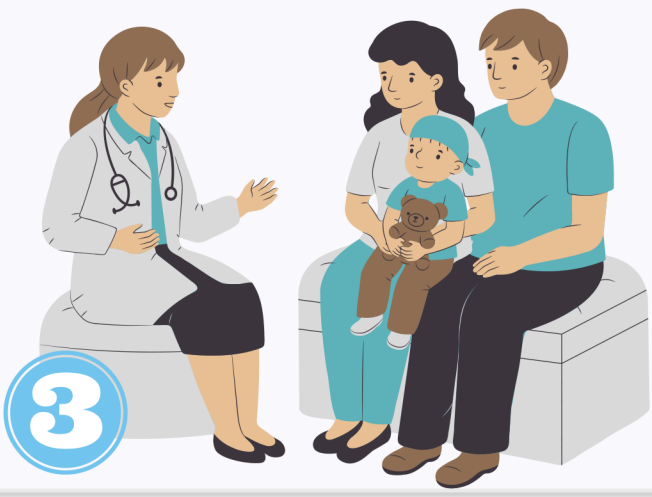
In addition to missed detection of CFTR variants in genetic panels, IRT cut off ranges can lead to false negative NBS. Infants with clinical signs - including bowel obstruction and failure to thrive - within the first month of life should be referred for evaluation **even with a negative NBS or detection of only one CFTR variant.** [2]

2 TIMELY EVALUATION AND TREATMENT IMPROVES HEALTH OUTCOMES.

Newborns with a positive NBS should be referred for a sweat test as soon as possible but no later than 28 days. Sweat testing can be done if weight is at or greater than 2 kilograms (4.4 pounds). Laboratory tests and DNA panels can also be run more frequently to allow results to be communicated quicker. [2]

CLEAR COMMUNICATION CAN REDUCE MISCONCEPTIONS AND IMPROVE UNDERSTANDING OF RISK AND OUTCOMES.

There are long held misconceptions that CF only affects infants of European ancestry. All infants can have CF. Ensuring that health providers and caregivers understand this and the benefit of earlier diagnosis is essential.



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CYSTIC FIBROSIS FOUNDATION ADDING TOMORROWS

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