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Cystic Fibrosis Newborn Screening: Prompt Care Improves Outcomes

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Research Support from: **Centers for Disease Control, Cystic Fibrosis Foundation**

I do not intend to discuss an unapproved/investigative use of a commercial product/device in my presentation..

I have not used artificial intelligence in the development of this presentation.

Objectives

- At the conclusion of this activity, learners will be able to:
  - Recognize the multisystem nature of cystic fibrosis, highlighting the critical importance of early diagnosis and treatment
  - Examine variation in cystic fibrosis newborn screening systems
  - Associate clinical outcomes from newborn screening for cystic fibrosis with delayed diagnosis/treatment and disparities based on construct of race and ethnicity

Join the Q&A and answer MOC part 2 questions at slido.com. Log in with the code #CENLApotpourri

Overview

Introduction to Cystic Fibrosis

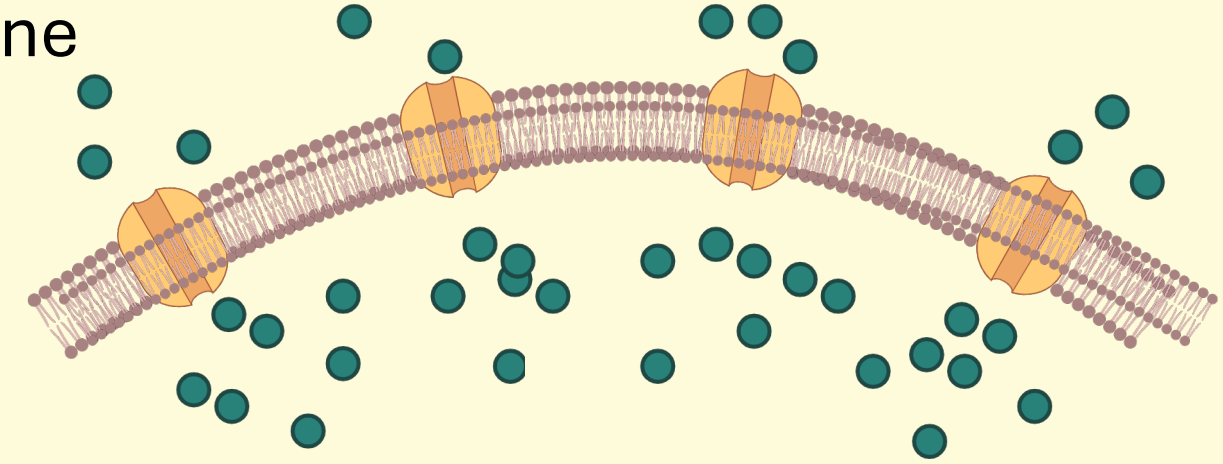
Cystic Fibrosis Newborn Screening

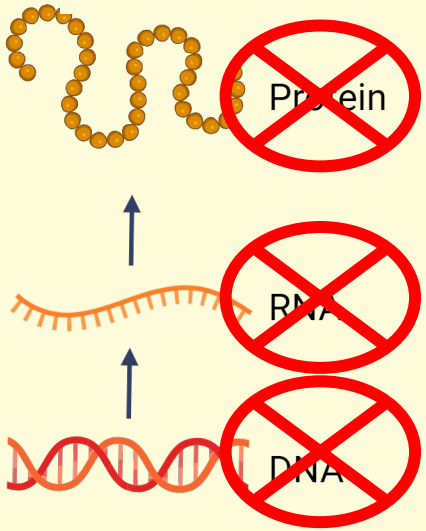
Outcomes in Cystic Fibrosis After Newborn Screening

Introduction to Cystic Fibrosis

CF: Autosomal Recessive Multi-System Disease

- Variants (aka mutations) in CFTR (CF transmembrane conductance regulator) gene





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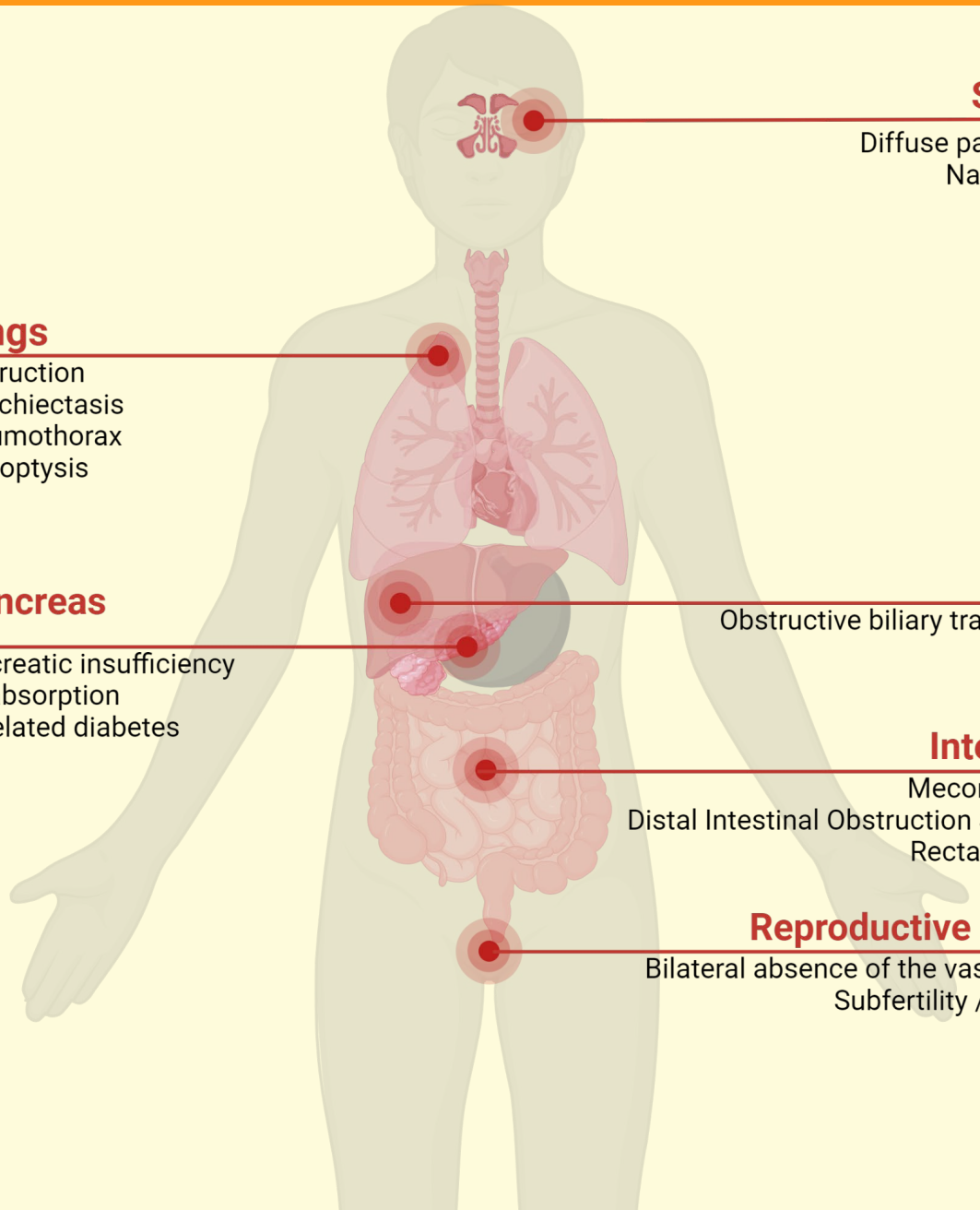
CF: Autosomal Recessive Multi-System Disease



Sinuses
Diffuse pansinusitis
Nasal polyps

Lungs
Obstruction
Bronchiectasis
Pneumothorax
Hemoptysis

Liver
Obstructive biliary tract disease

Pancreas
Pancreatic insufficiency
Malabsorption
CF related diabetes

Intestines
Meconium ileus
Distal Intestinal Obstruction Syndrome
Rectal Prolapse

Reproductive system
Bilateral absence of the vas deferens
Subfertility / Infertility

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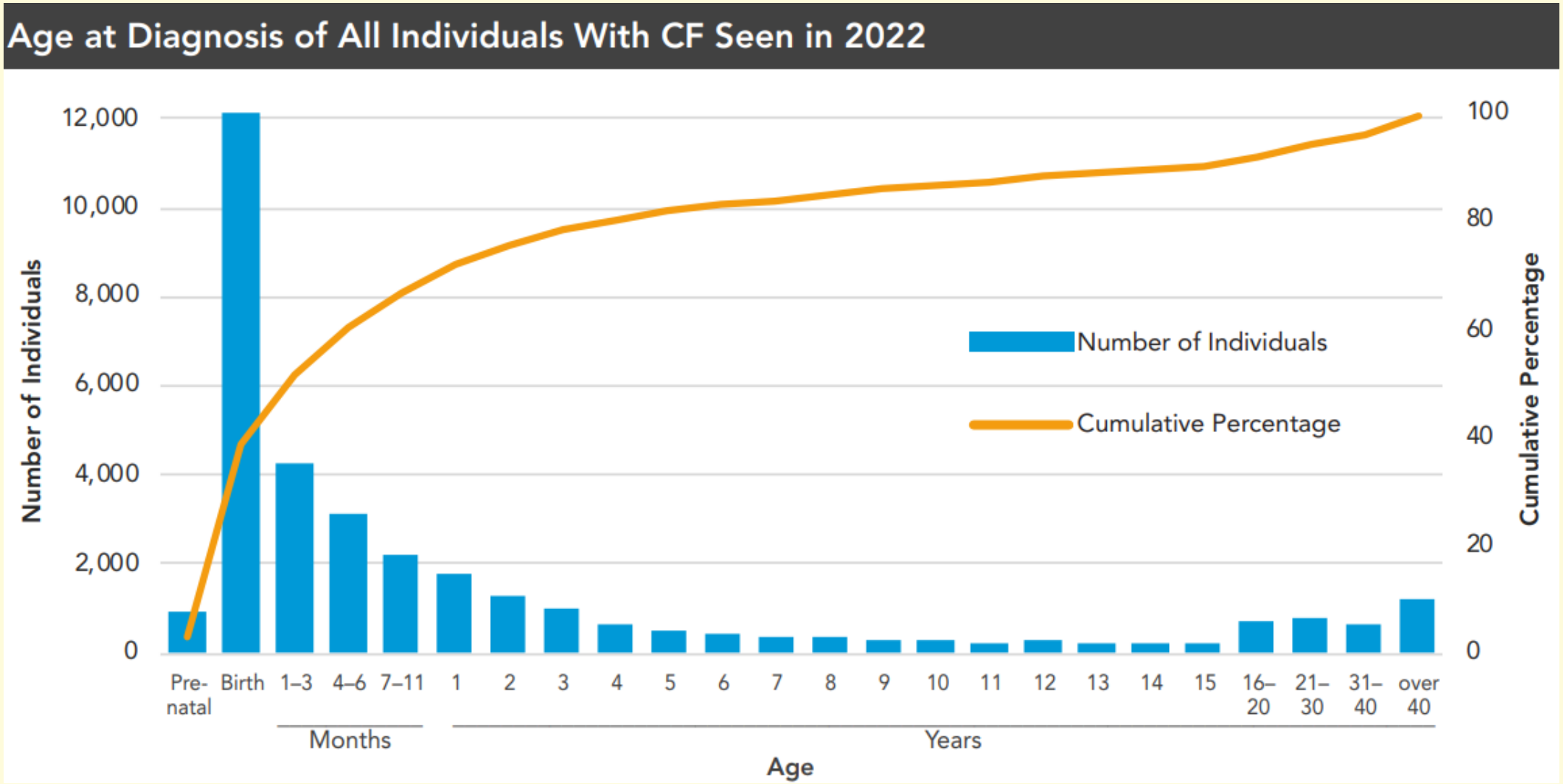
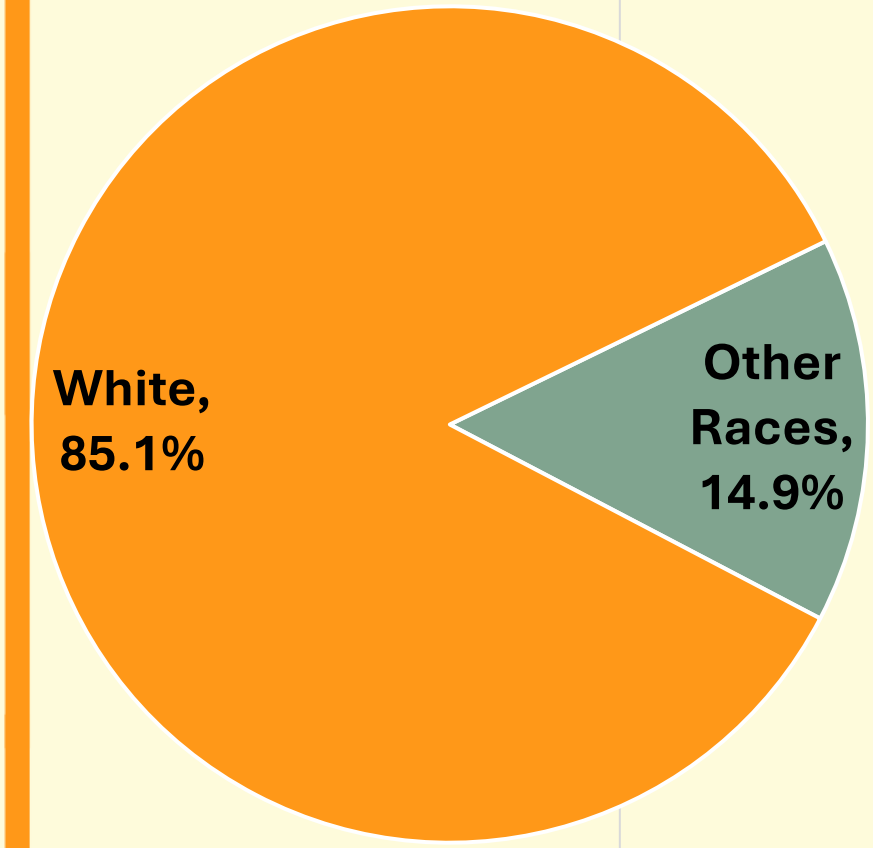
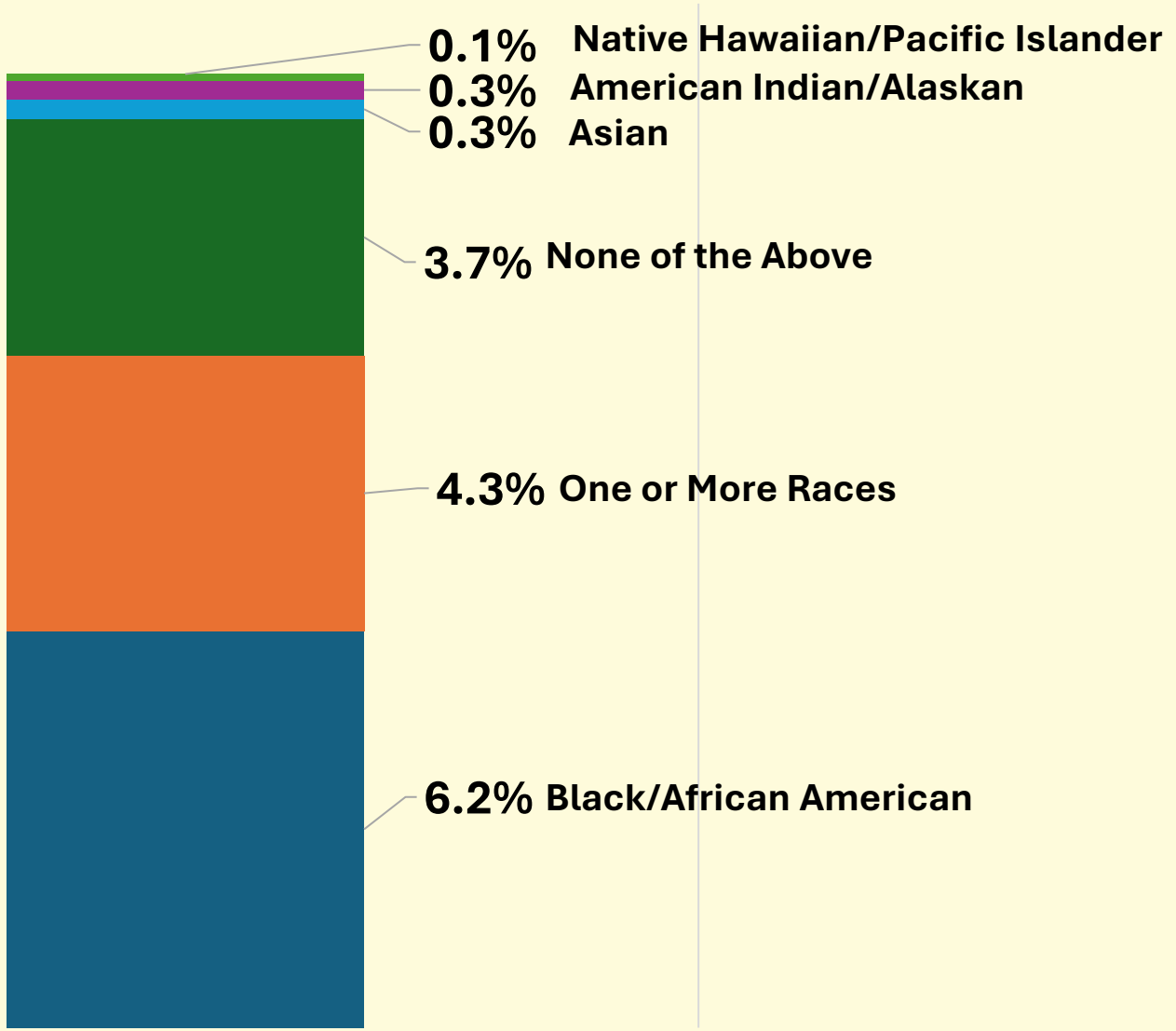
CF can be Diagnosed at All Ages



2022 Annual Data Report (accessed CFF.org July 2024)

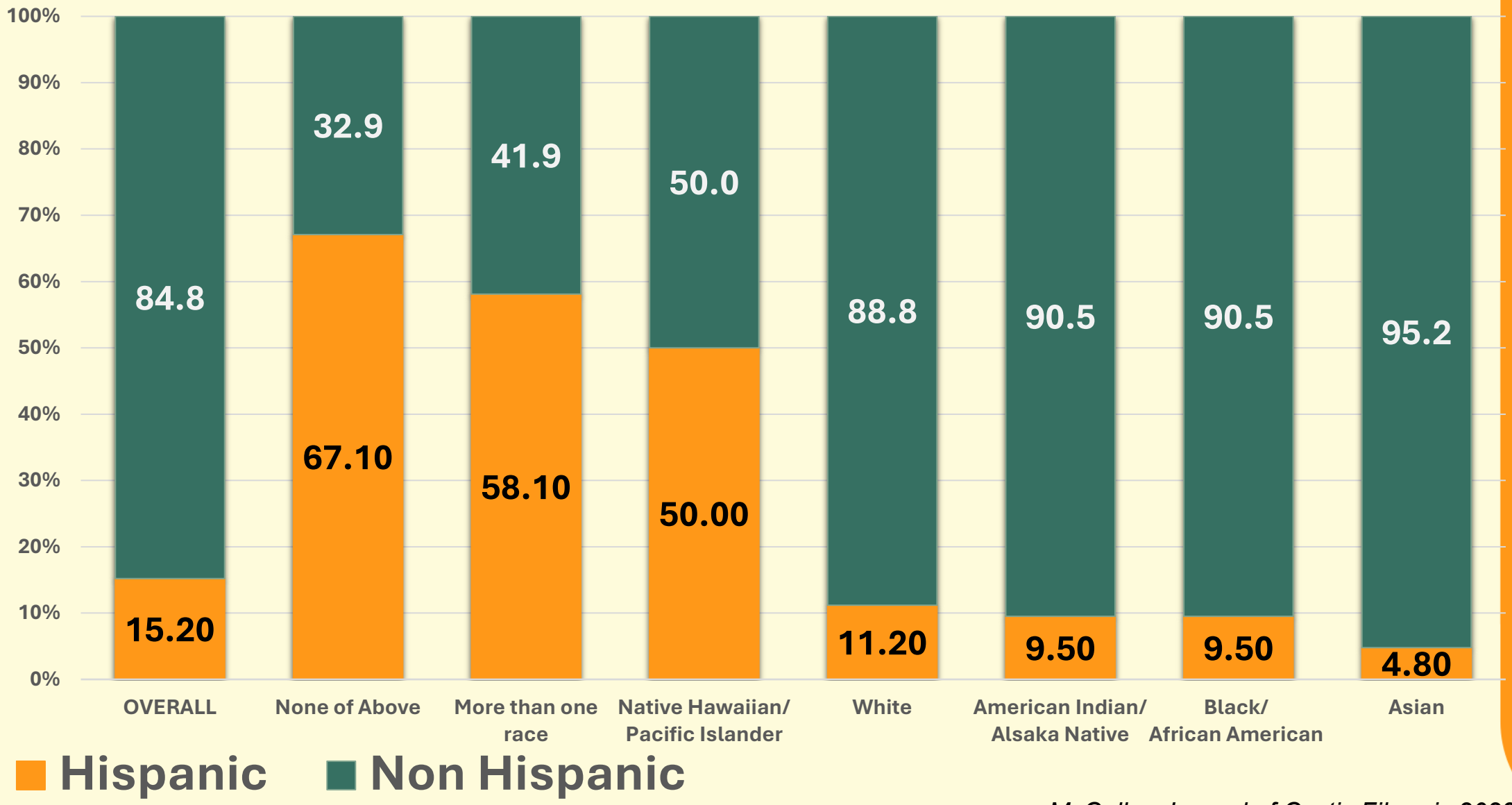
Race of Infants Born between 2010-2018, Diagnosed with CF after NBS





McColley Journal of Cystic Fibrosis 2022

Hispanic Ethnicity of Infants With CF Born 2010-2018



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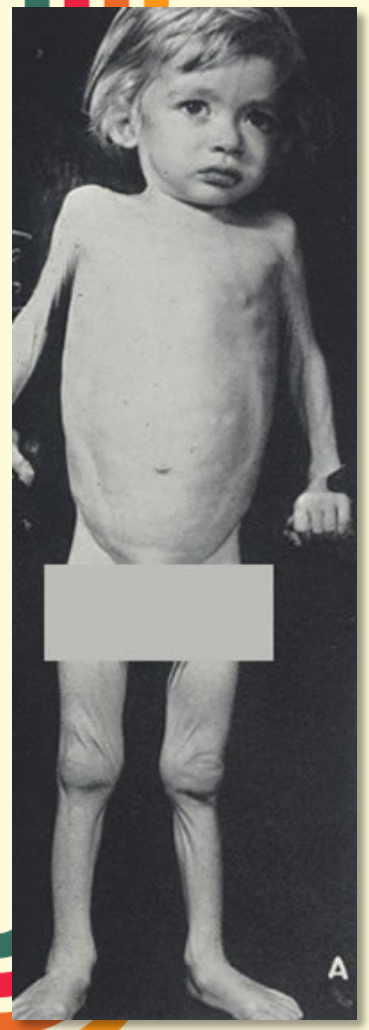
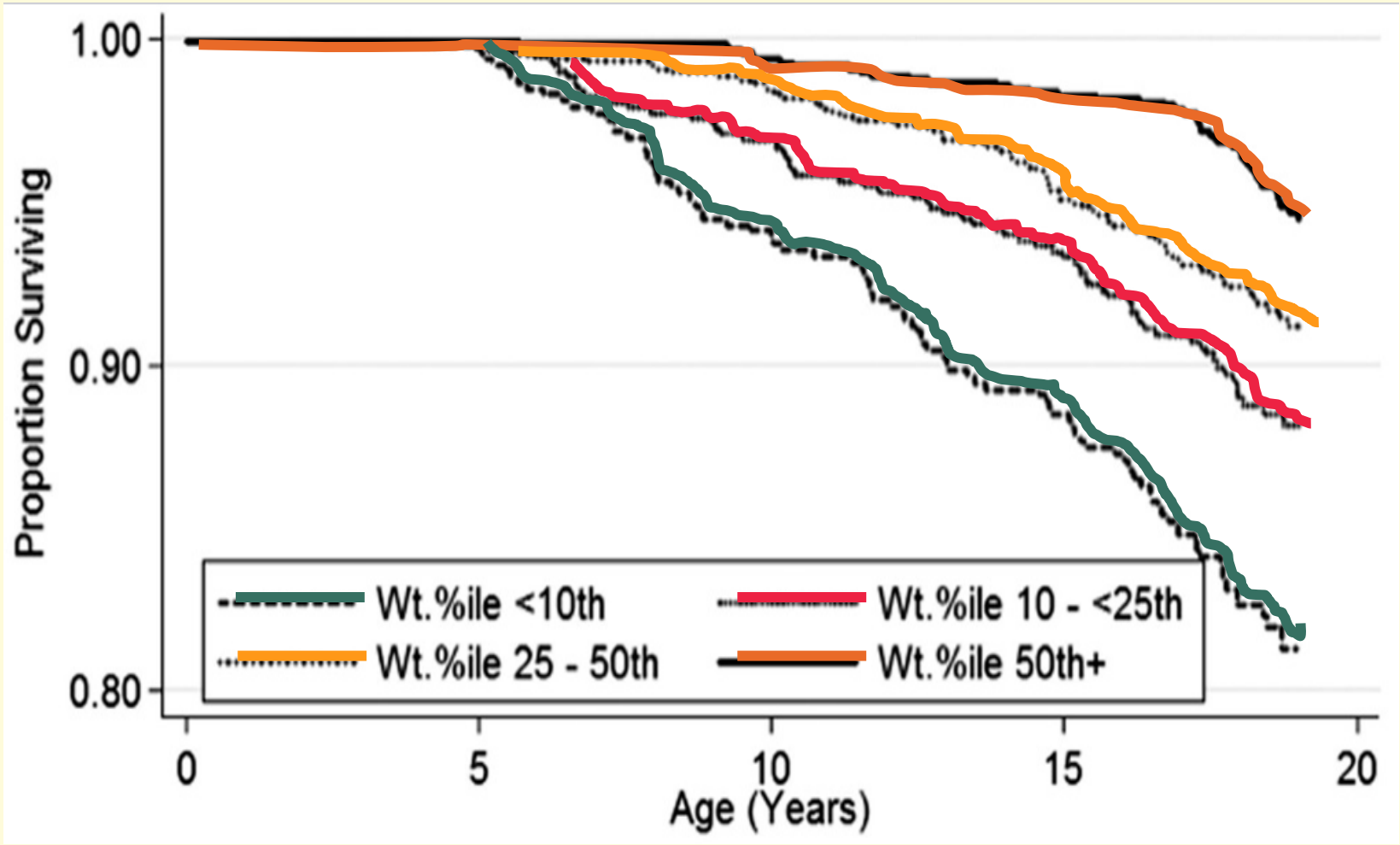
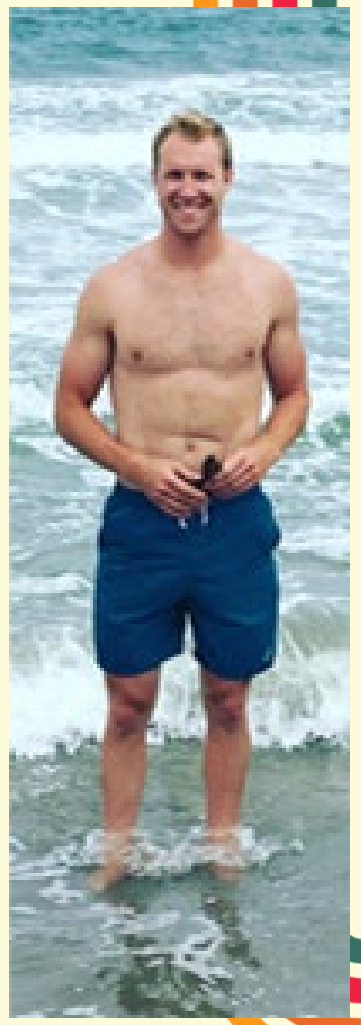
Without Treatment, CF is Fatal in Early Life

- Malnutrition, vitamin deficiency, hyponatremia can be **fatal in early infancy**
- Childhood weight and height percentiles **predict survival** to adulthood in CF





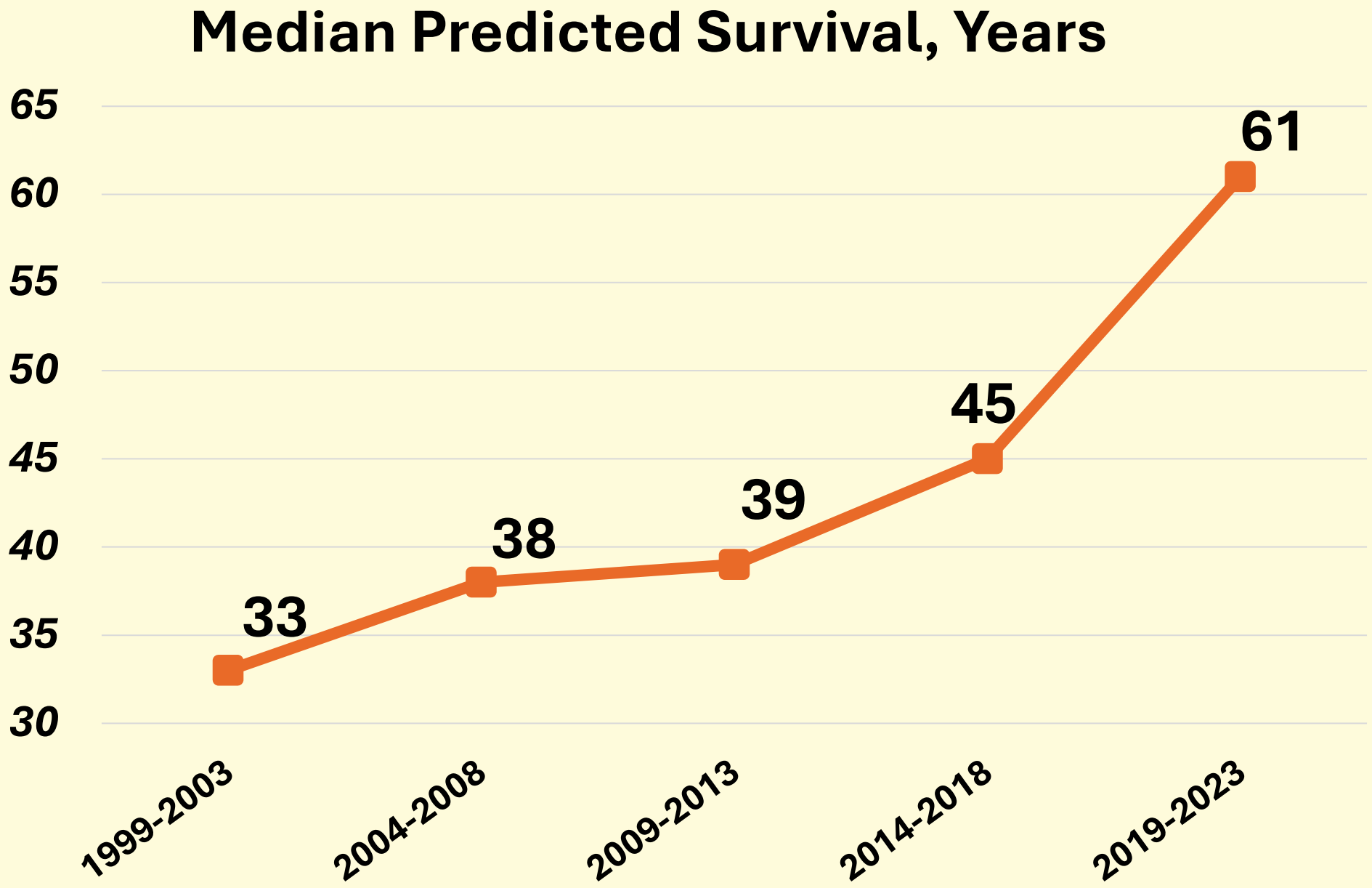


Yen J Peds 2013, cfmedicine.com (accessed August 2017), CFF.org (accessed September 2017)

Early Nutrition, Improved Lung Function

Sanders J Peds 2015

Accelerated Improvement in CF Life Expectancy

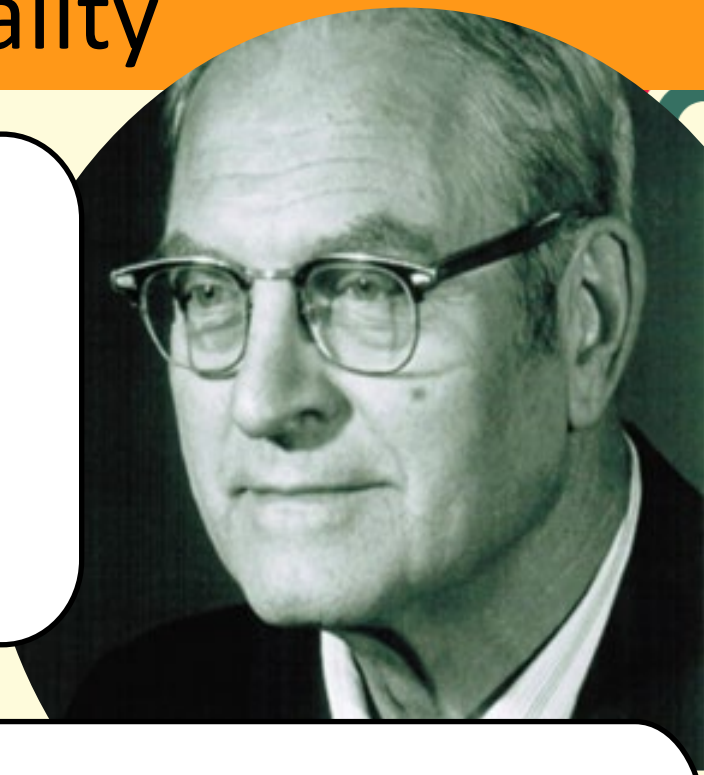


2023 Cystic Fibrosis Foundation Patient Registry Highlights (accessed CFF.org July 2024)

Cystic Fibrosis Newborn Screening

NBS Reduced Morbidity/Mortality

The history of NBS begins with Bob Guthrie, who developed an assay for phenylketonuria (PKU) and a filter paper card for blood collection



NBS tests infants for medical conditions that lead to significant morbidity or mortality and have a treatment that can improve outcomes

Primary Care Providers Play Crucial Role in NBS

NBS is standard for a growing number of disorders

Parent education in newborn period to increase

- Awareness
- Acceptance (to reduce refusal)
- Action (follow-up results, testing if positive screen)

Positive screens are common - Follow up promptly

Know state algorithms

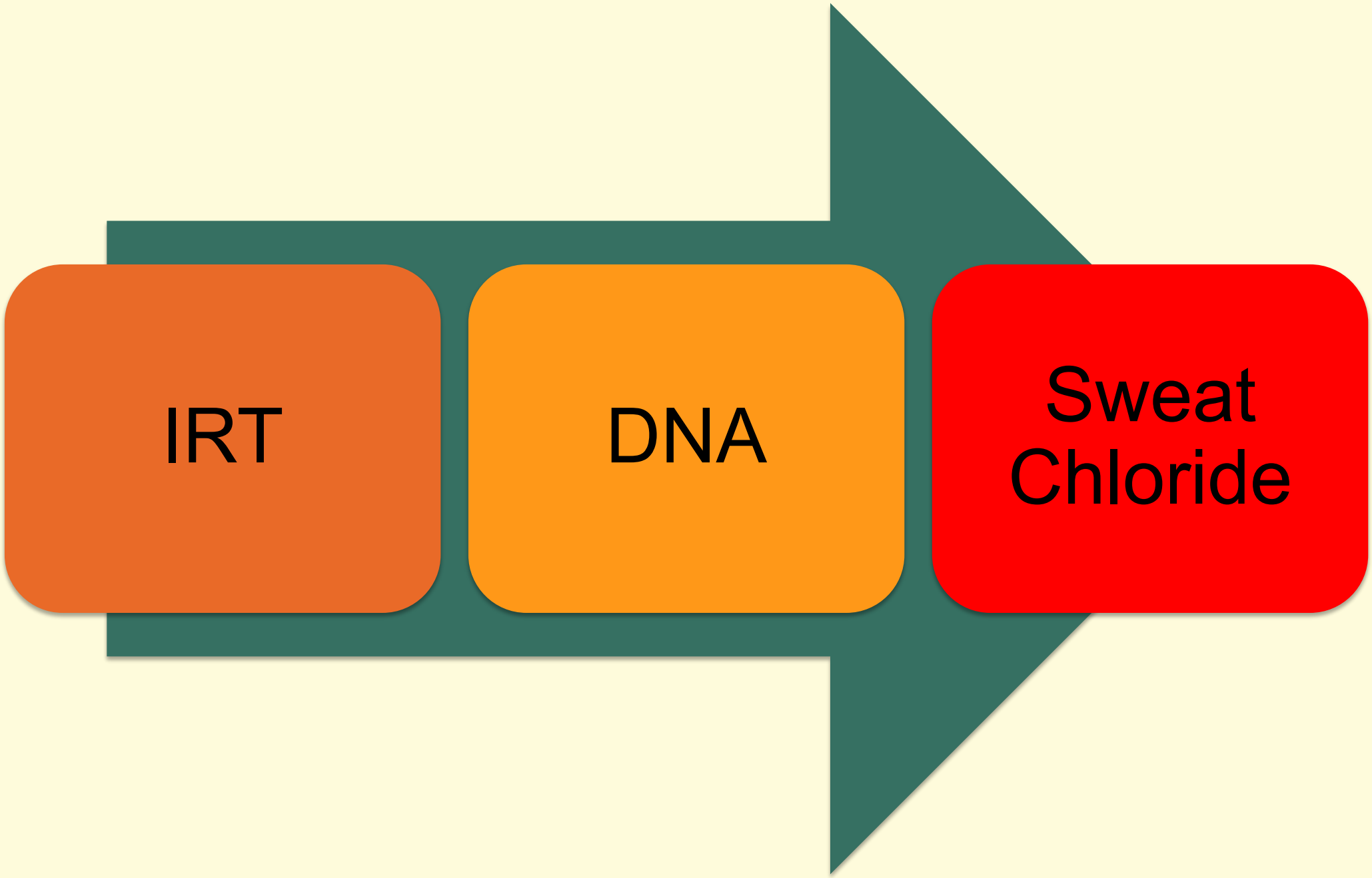
Act promptly when there is a positive (out of range) screening test result

CDC Recommend Screening for CF Addition in 2004

Evidence from randomized controlled trials, cohort studies, comparisons of screened patient versus unscreened, and registry data showed

“the magnitude of the health benefits from screening for CF is sufficient that states should consider including routine newborn screening for CF in conjunction with systems to ensure access to high quality care”

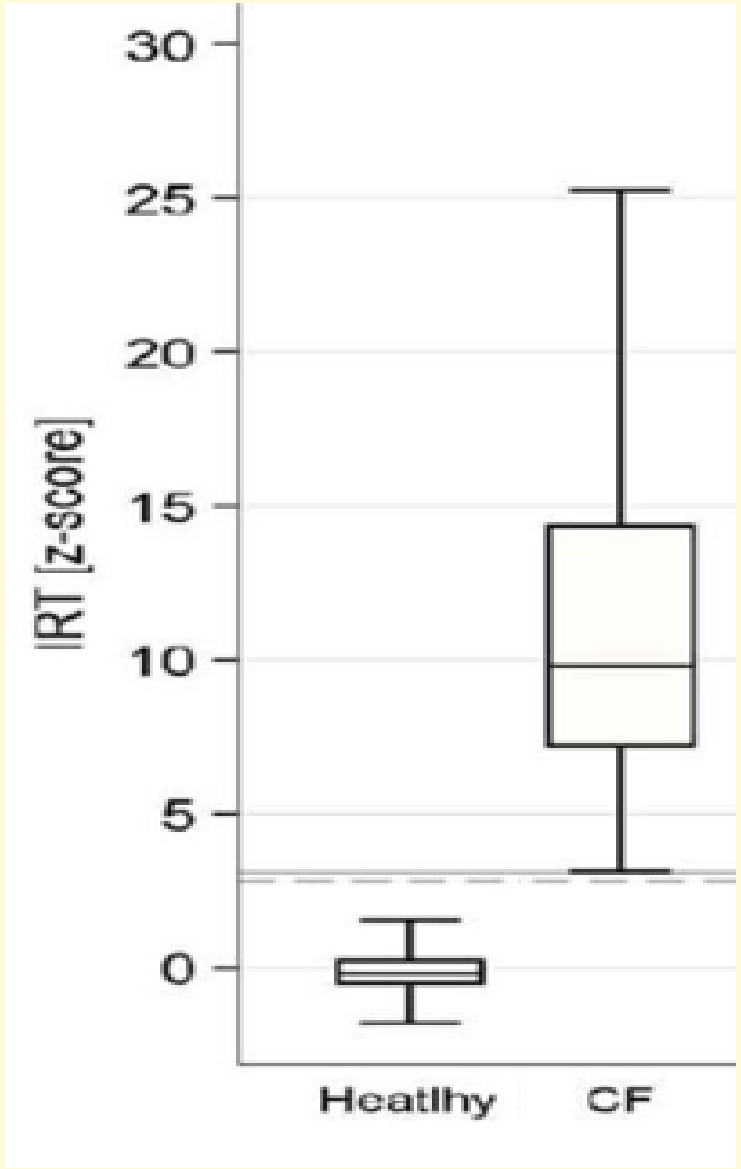
MMWR 2004


IRT
DNA
Sweat Chloride

Blood Spot Tested for IRT (immunoreactive trypsinogen)

IRT levels are 2 to 5 times higher in neonates with CF

Reflects leakage of pancreatic secretions into the blood during fetal life due to blockage of pancreatic ducts

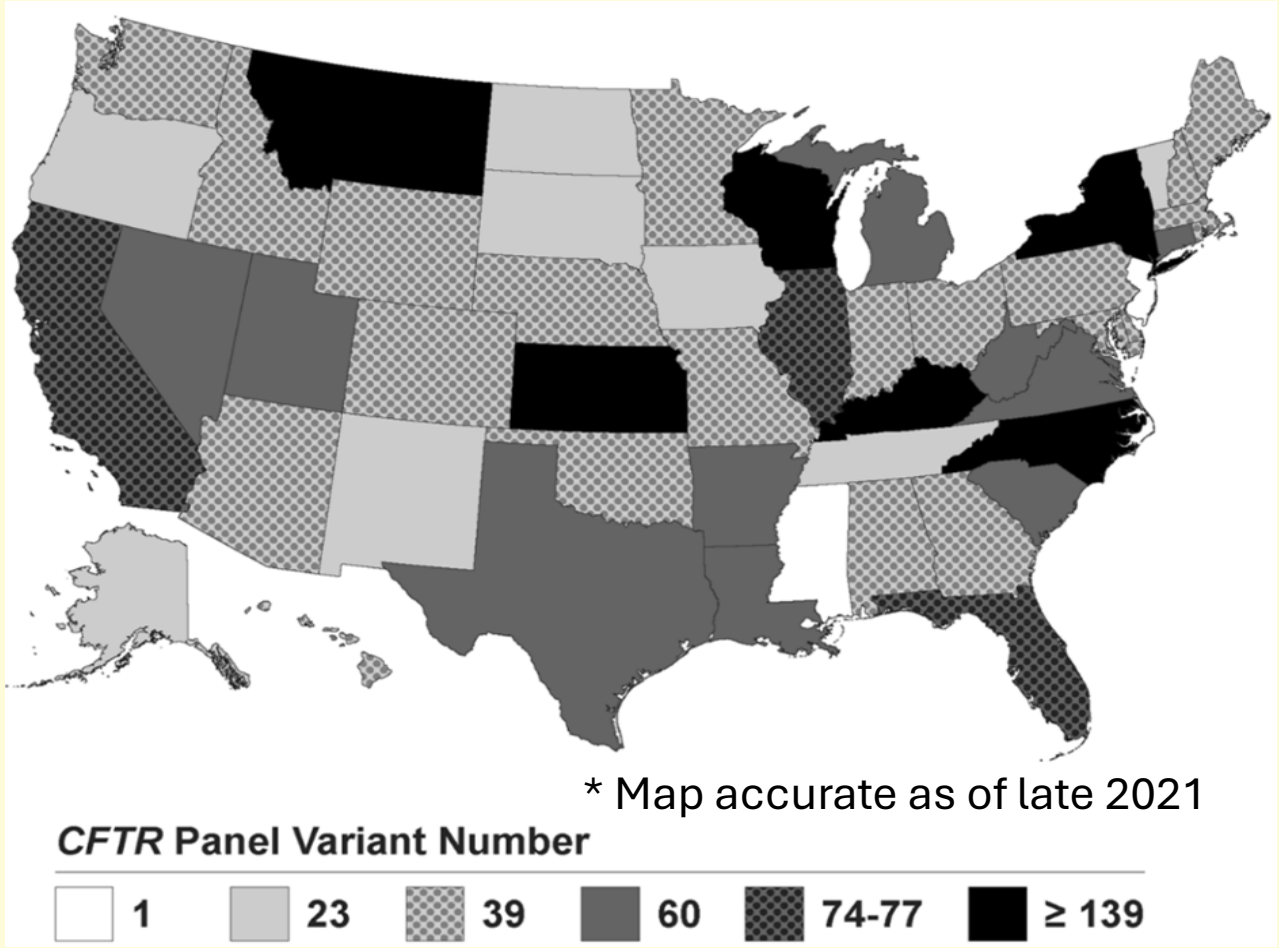


Fingerhut Arch Dis Child Fetal Neo Ed 2022

Elevated IRT Leads to Genetic Analysis

Bloodspot is analyzed for CFTR variants

Number of CFTR variants differs between states

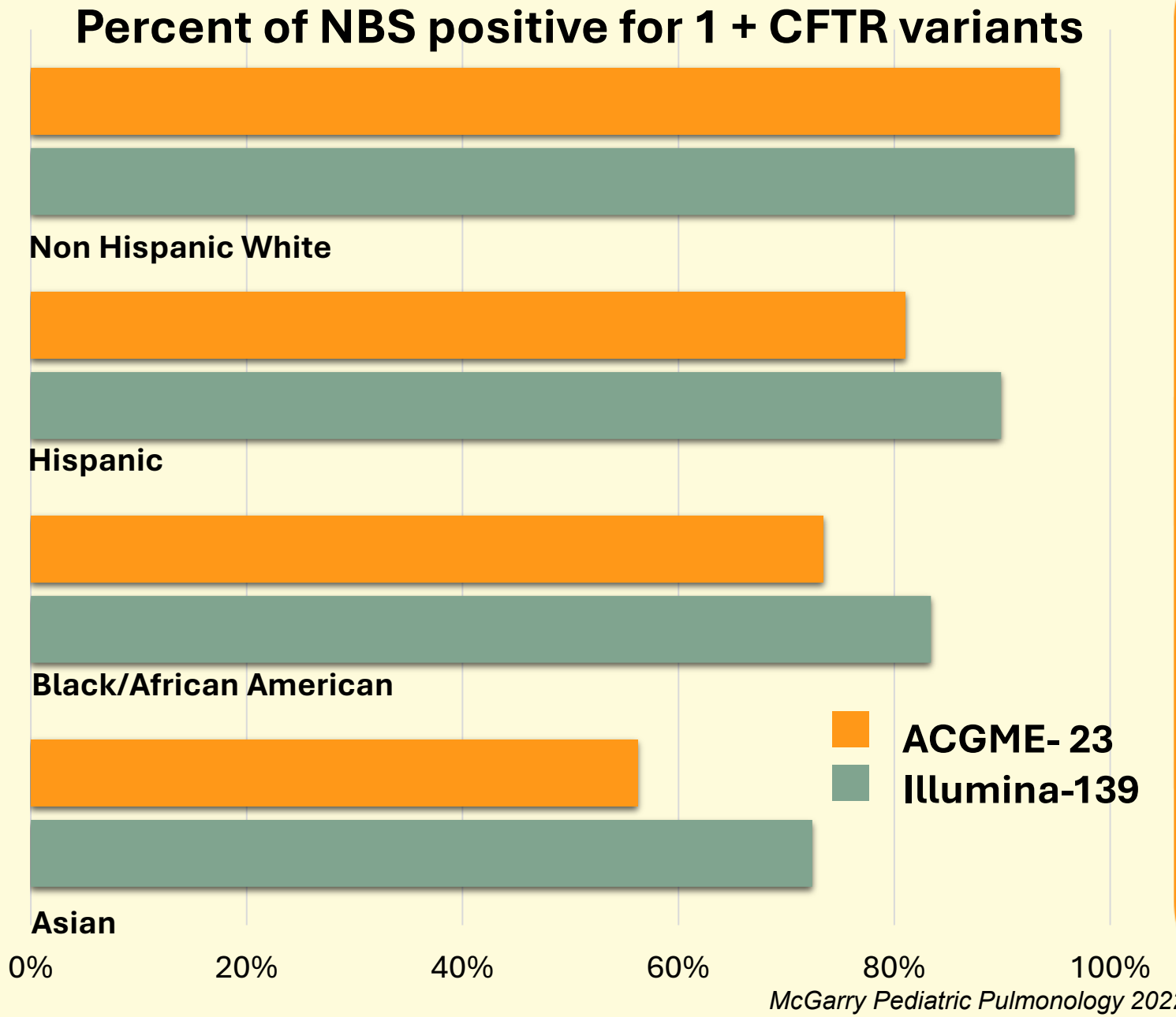


Rehani Pediatric Pulmonology 2022

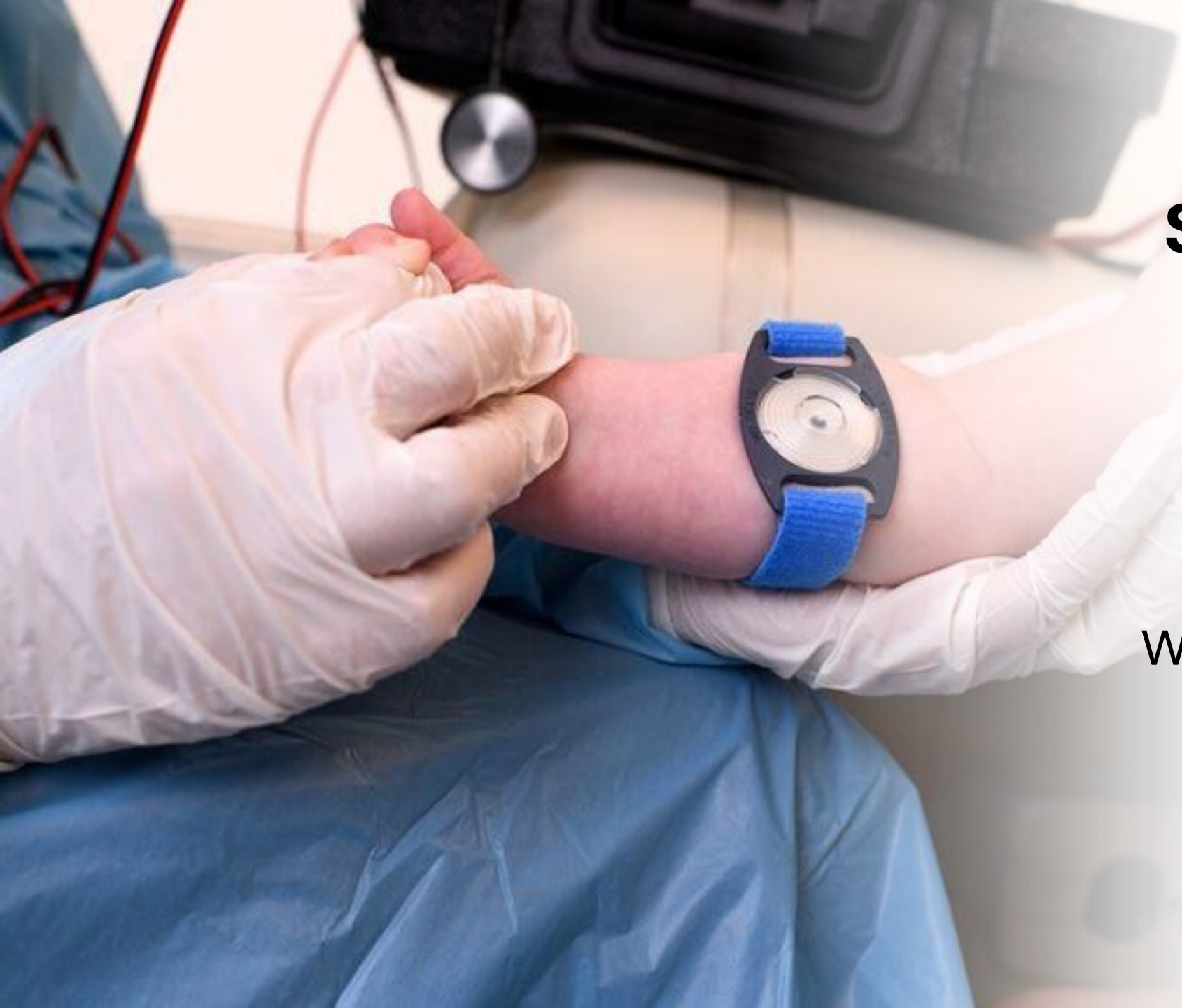
Variation in Genetic Analysis Leads to Disparities

When fewer CFTR variants analyzed, detection rate lower in minoritized populations

NBS panels under-represent CFTR variants seen in Black and Hispanic people with CF



McGarry Pediatric Pulmonology 2022



Sweat test is Diagnostic GOLD Standard is Sweat Chloride Test

Collaborate with CF Center

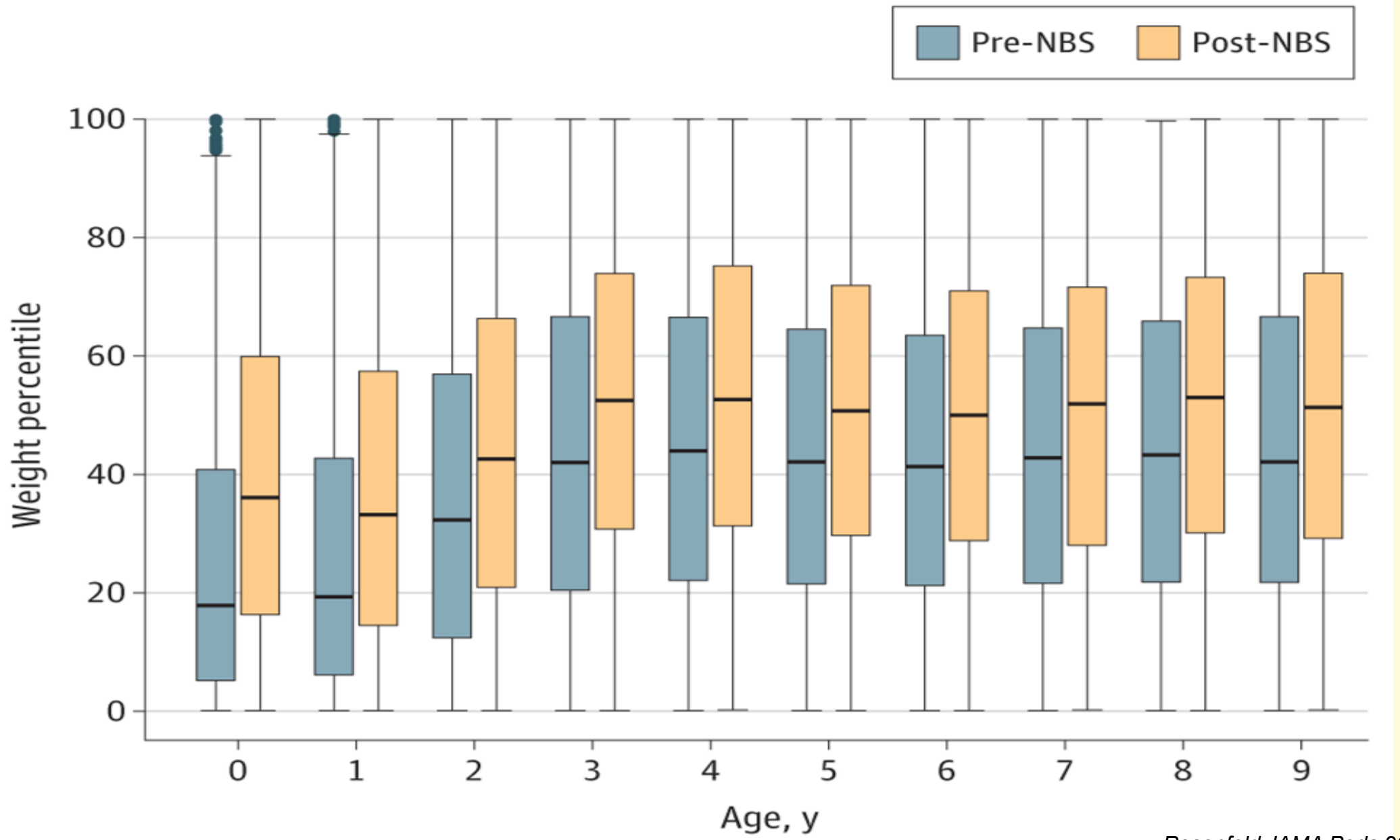
WHO needs sweat test?

-Abnormal NBS
(regardless of mutations found)

-Normal NBS with symptoms of CF or full sibling with CF

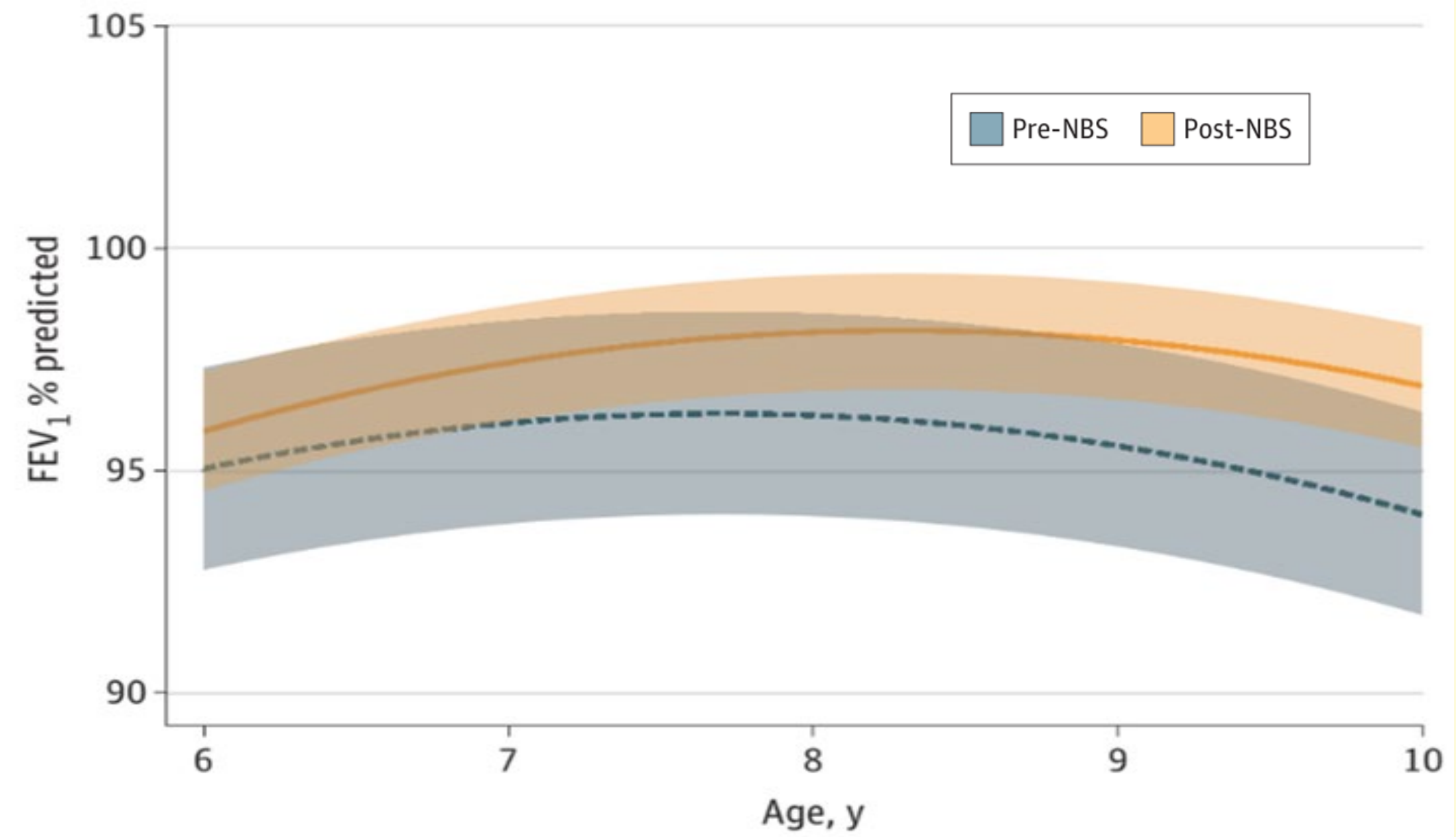
Outcomes in Cystic Fibrosis After Newborn Screening

Improved Nutrition after Implementation of CF NBS



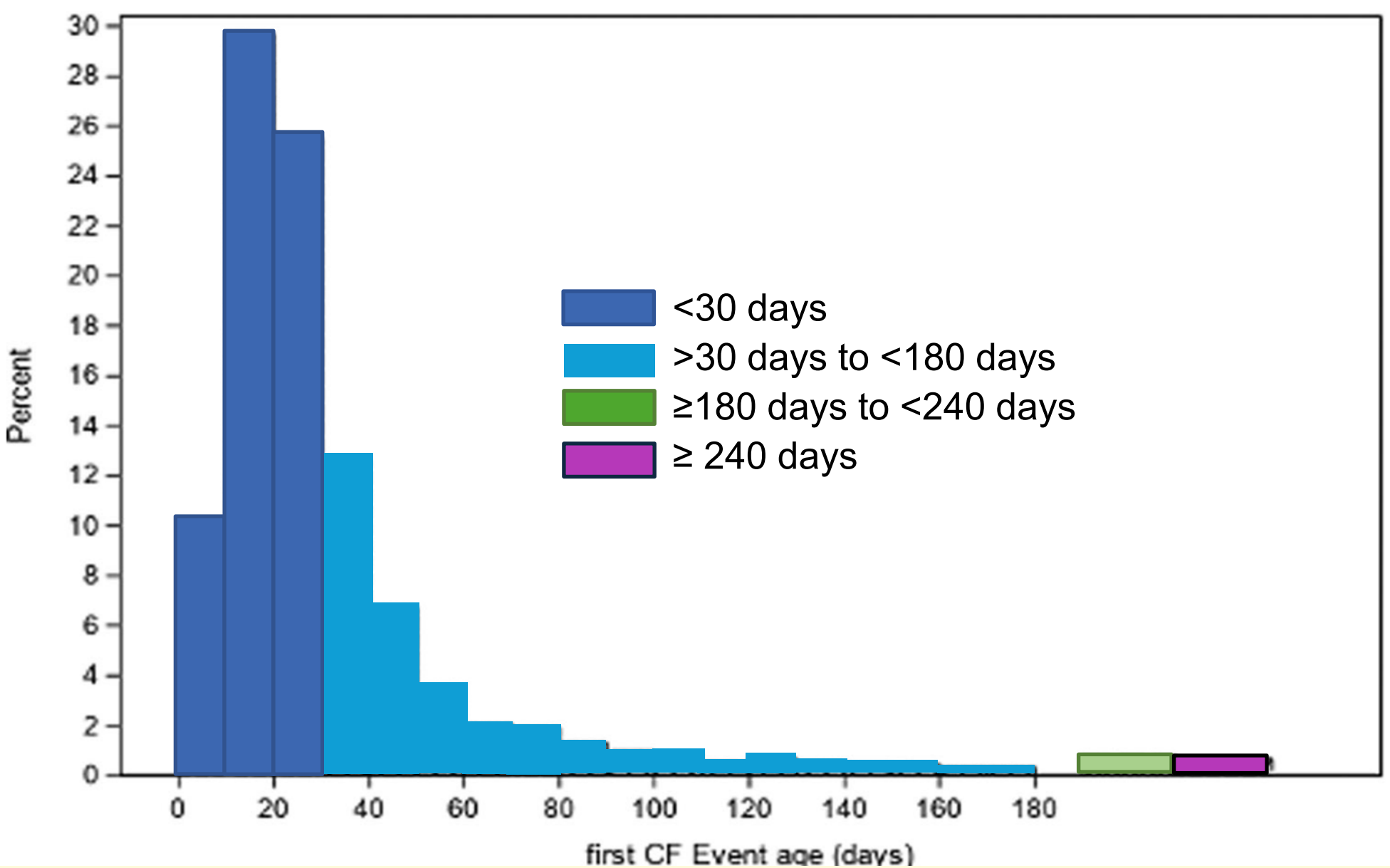
Rosenfeld JAMA Peds 2022

Pulmonary Function Improved since NBS Started



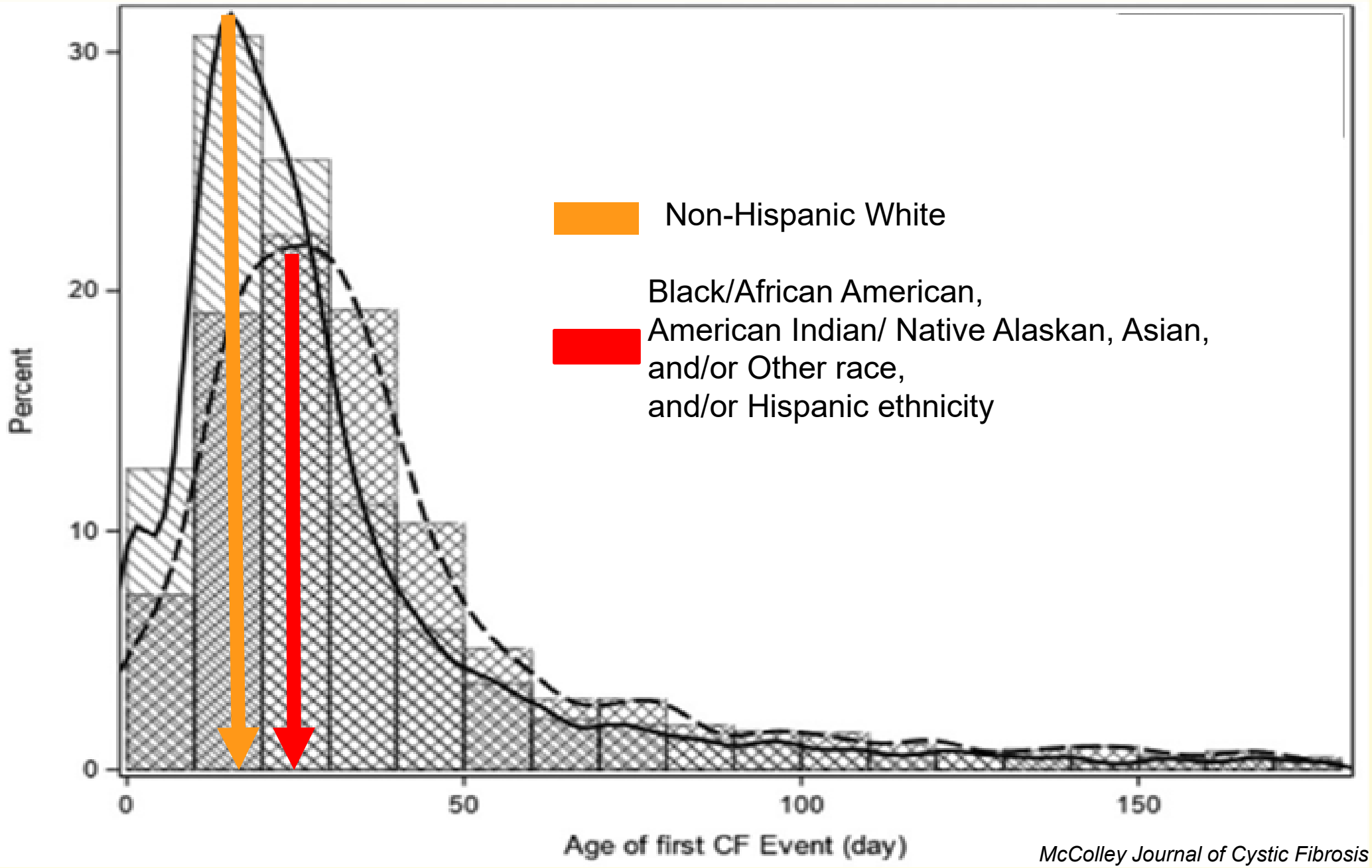
Rosenfeld JAMA Peds 2022

During 9 Years of NBS Delays to First Encounter Persists



Martiniano Ped Pulm 2021

Disparity in Follow-up by Race/Ethnicity



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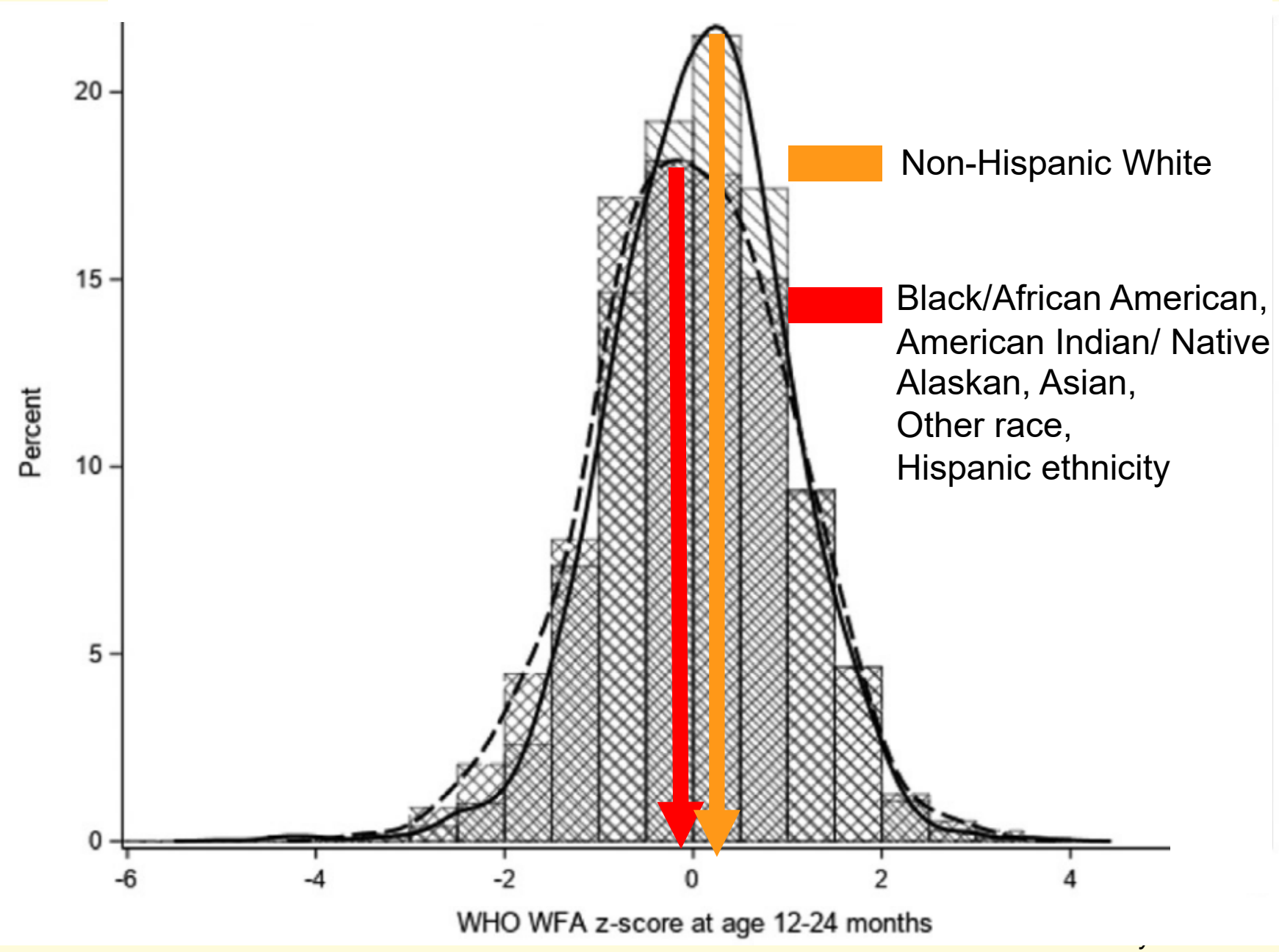
Delayed Age at First Event Associated with Worse Outcomes

- More respiratory symptoms
- More failure to thrive
- Nutrition outcomes
  - First Visit: Lower median height & higher frequency with height <10$^{th\%}$ile.
  - 12–24-month visit: Lower weight & height for age
- Higher rate of hospitalizations for pulmonary exacerbation

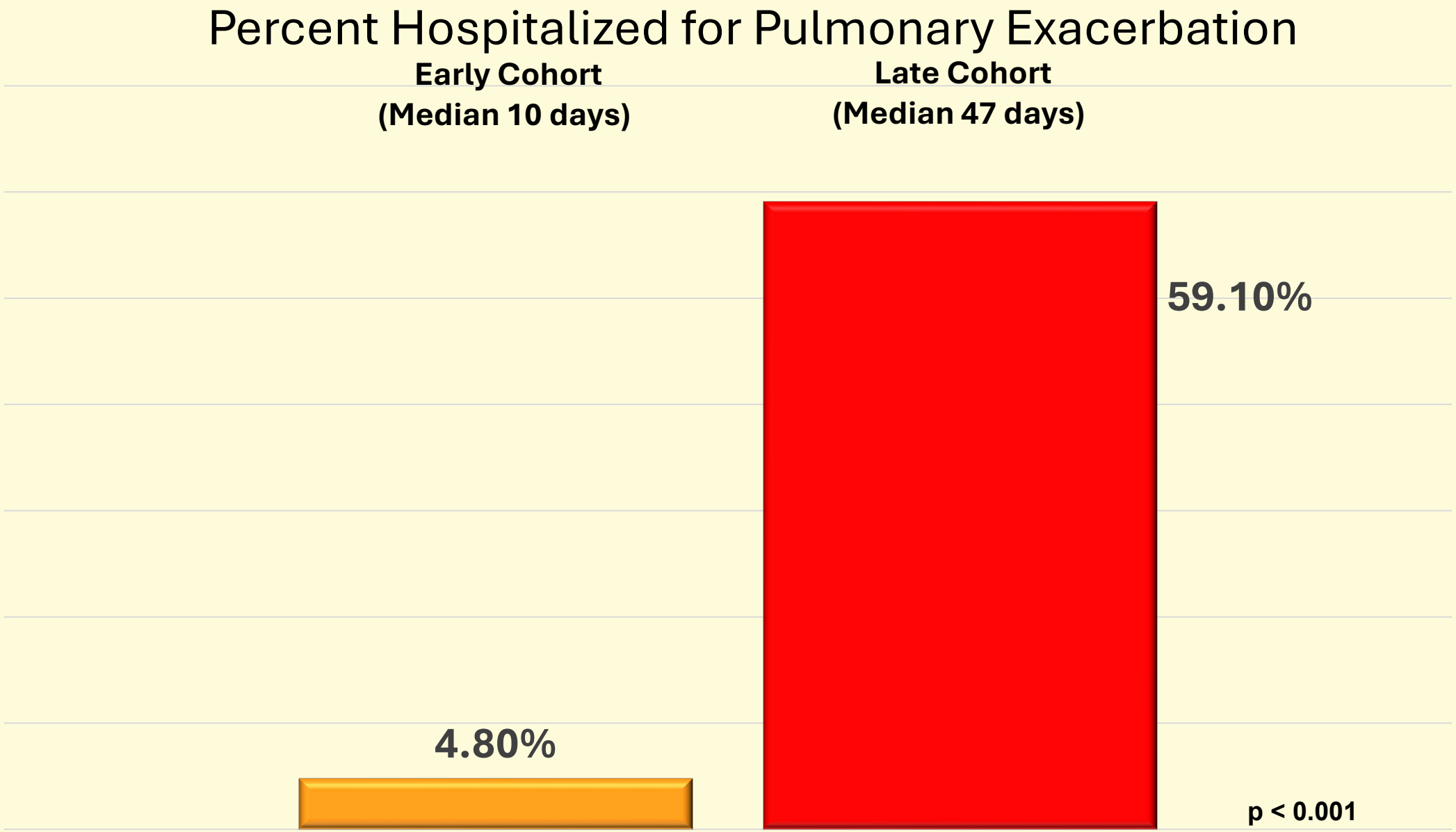
McColley Journal of Cystic Fibrosis 2022

Nutritional Gaps by Race/Ethnicity Continue Despite NBS



ystic Fibrosis 2022

Hospitalization for Exacerbation Higher in Delayed Care



Martiniano J Ped 2023

Summary

- CF is a multisystem disease affecting ALL ages, races, and ethnicities.
- Without treatment, CF is often fatal in early life.
- Life expectancy is increasing
- NBS contributes to earlier diagnosis and improves outcomes.
- Disparities persist, with delayed diagnosis and worse outcomes.

- **To optimize health and reduce disparities for infants and children with CF, immediate referral for diagnostic testing and treatment is needed.**