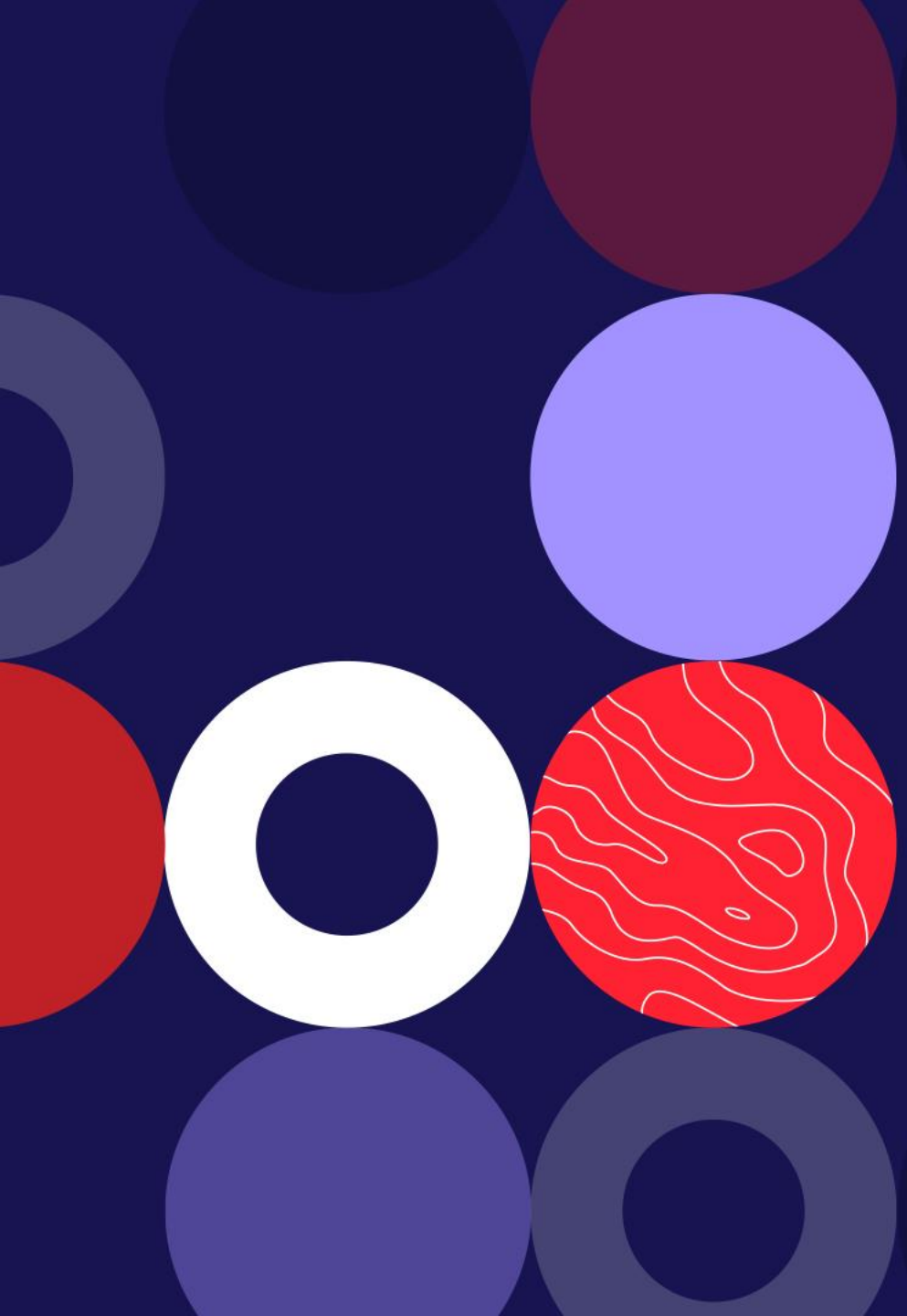

Practical Updates in Primary Care

Advances in the Identification and Management of Cystic Fibrosis: From Screening to Long-Term Care

Ahmet Uluer, DO, MPH

Director, Adult Cystic Fibrosis Program
Adult Director, Therapeutic Development Network
Division of Pulmonary and Critical Care
Brigham and Women's Hospital and Boston Children's Hospital
Assistant Professor, Harvard Medical School

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Atlantic Health Morristown Medical Center
Clinical Associate Professor, Sidney Kimmel Medical College

Supported by an educational grant from Vertex Pharmaceuticals Incorporated.

Disclosures

- **Ahmet Uluer, DO, MPH:** Grant Funding – Cystic Fibrosis Foundation, Therapeutics Development Network; PI (as faculty at an institution that is part of the CF TDN) – BiomX, Spirovant, Vertex

- **Rebecca A. Griffith, MD, FACP:** PI and Sub-PI – Cystic Fibrosis Foundation-funded studies; Grant Recipient – Cystic Fibrosis Foundation

  Atlantic Health receives financial compensation from Vertex Pharmaceuticals for my work as an investigator in trials

Learning Objectives

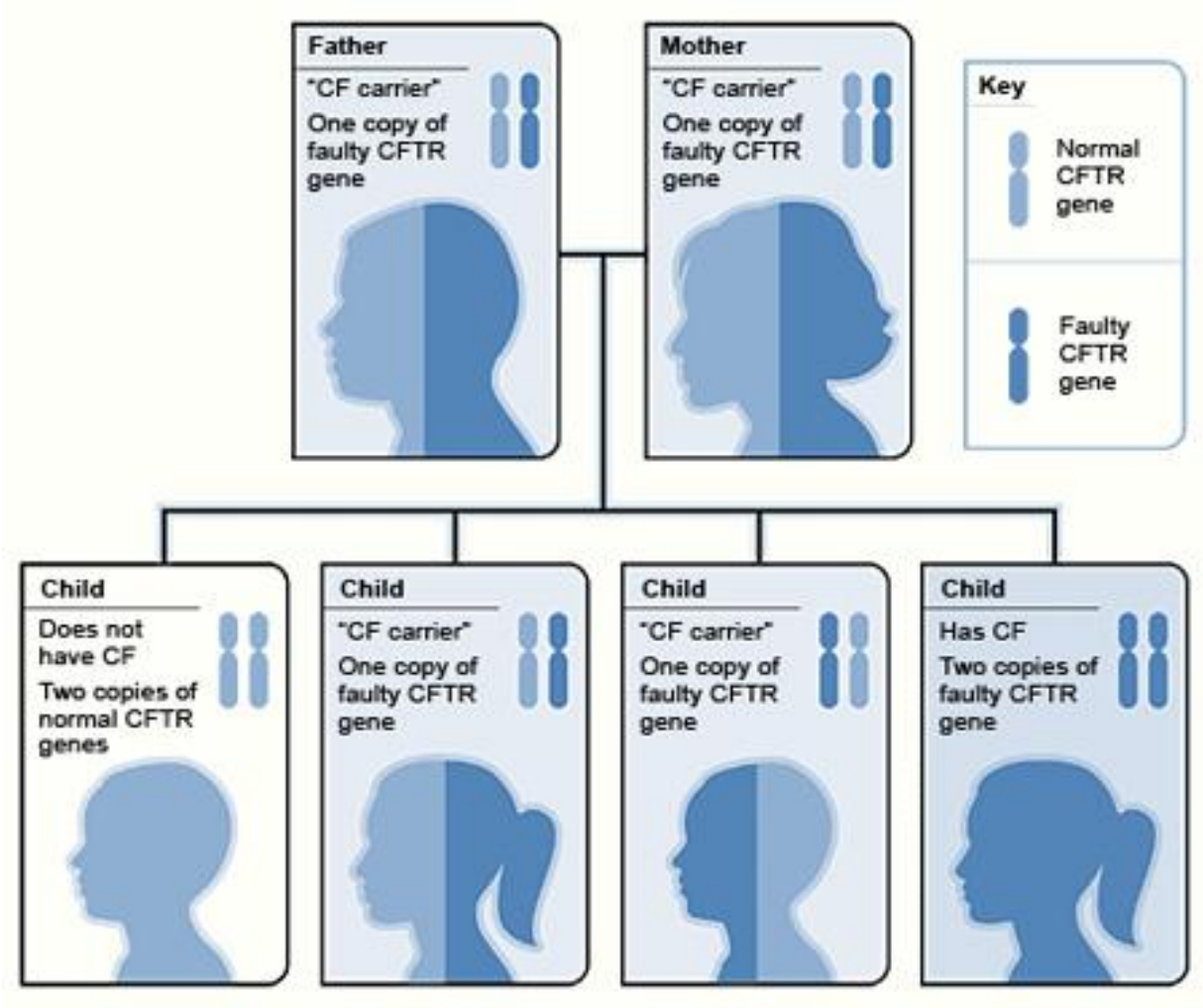
- Apply screening guidelines for the timely assessment and diagnosis of CF in pediatric and adult patients
- Assess efficacy and safety data on current and emerging therapies, including those targeting CFTR, for patients with CF
- Implement multidisciplinary and guideline-directed approaches to optimize the management of individuals with CF

CF = cystic fibrosis; CFTR = cystic fibrosis transmembrane conductance regulator.

Practical Updates in Primary Care

Cystic Fibrosis Overview: From Newborn Screening and Diagnosis to Targeted Therapies

CF: Inheritance Pattern



- Autosomal recessive inheritance associated with CFTR gene
- Most common genetic mutation is F508del in European ancestry
  - Less common in other parts of the world
- Estimated 162,000+ people with CF worldwide

Guo J, et al. *J Cyst Fibros*. 2022;21(3):456-462. Image source: https://commons.wikimedia.org/wiki/File:Cysticfibrosis02.png.

Cystic Fibrosis Symptoms

- Salty-tasting skin
- Persistent cough
- Frequent lung infections
- Chronic sinus infections
- Nasal polyps
- Wheezing or shortness of breath
- Inability to gain weight, poor growth
- Greasy, bulky stools or difficulty with bowel movements
- Male infertility

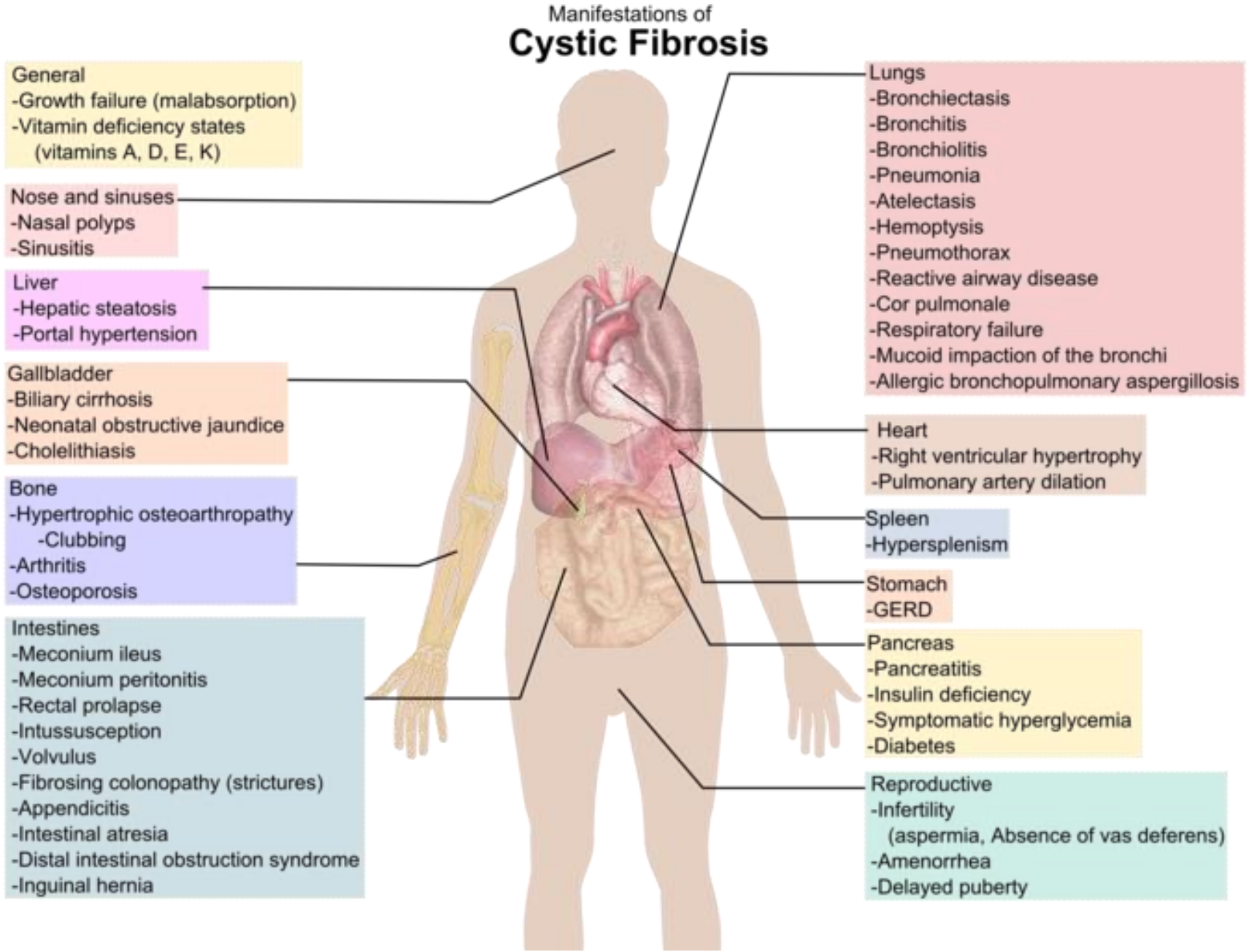
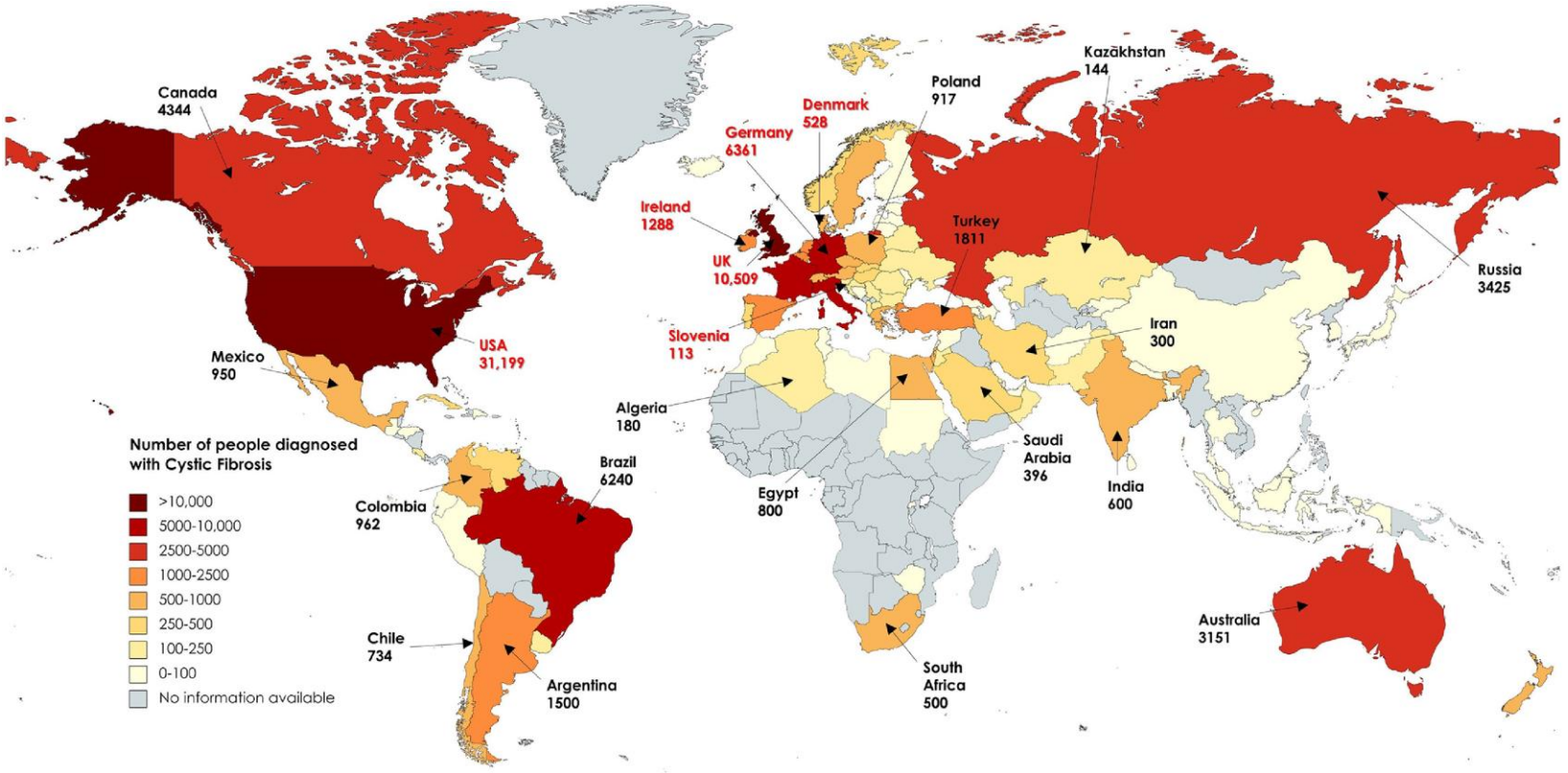
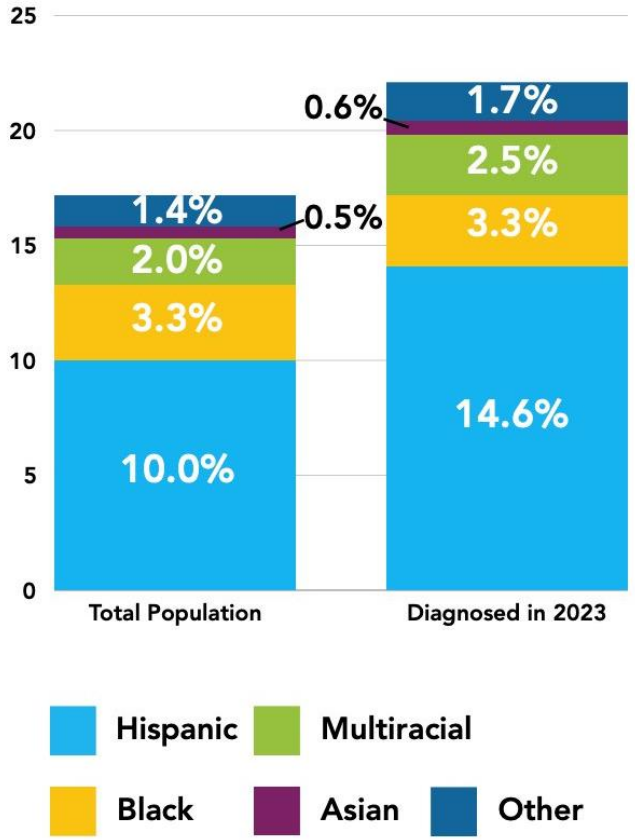


Image source: https://commons.wikimedia.org/wiki/File:Cystic_fibrosis_manifestations.svg.

Epidemiology

- Today: We are learning that the CF population is much more diverse every year
- The largest potential population (diagnosed + undiagnosed) is thought to be in India





Guo J, et al. *J Cyst Fibros*. 2022;21(3):456-462. Cystic Fibrosis Foundation (CFF). Accessed April 24, 2026. https://www.cff.org/sites/default/files/2025-05/2024-Patient-Registry-Highlights-Report.pdf.

Prevalence of the Most Common CFTR Mutations in pwCF

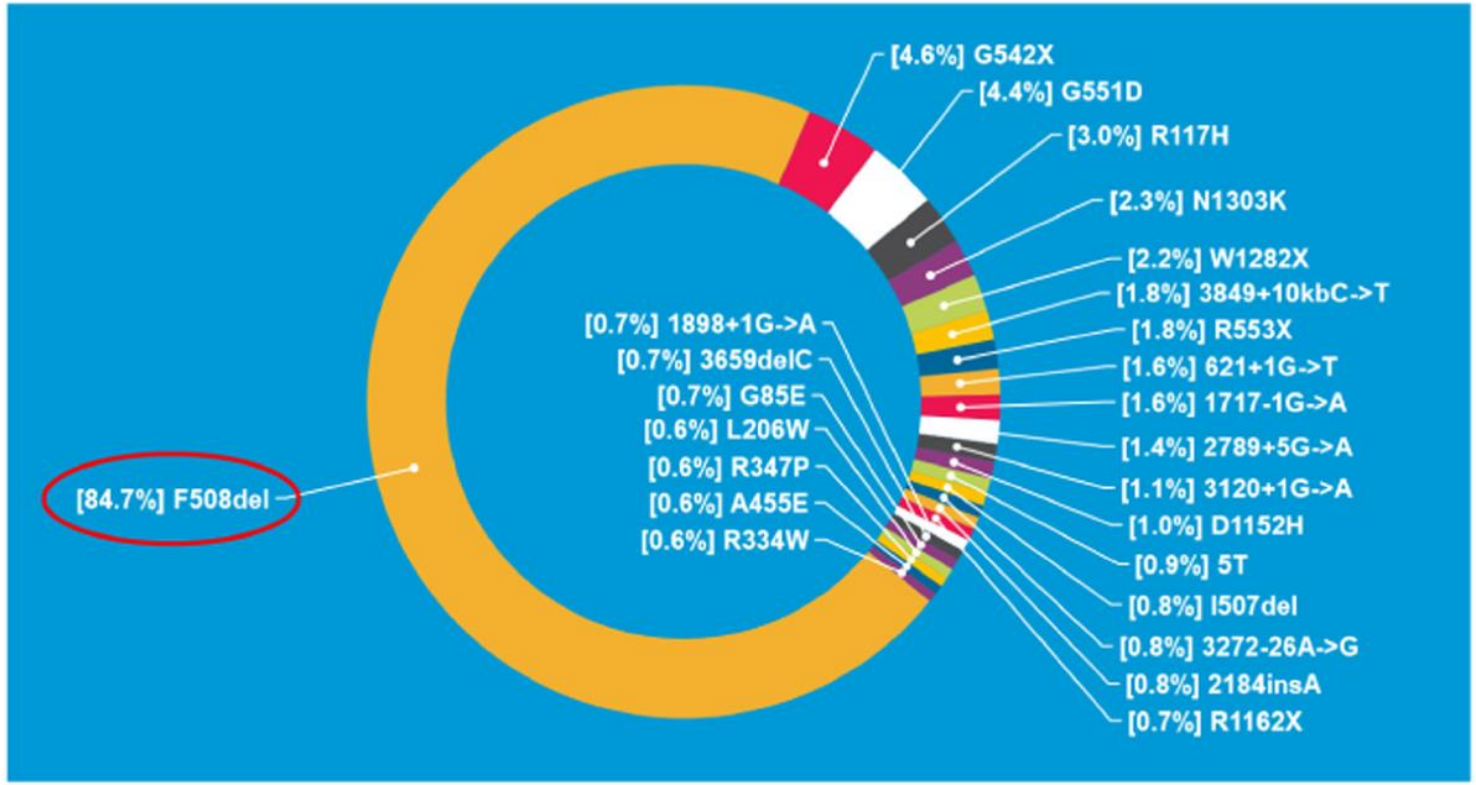


FIGURE 5 Prevalence of the most common CFTR mutations in people with CF seen in 2018.[20] CFTR, cystic fibrosis transmembrane conductance regulator

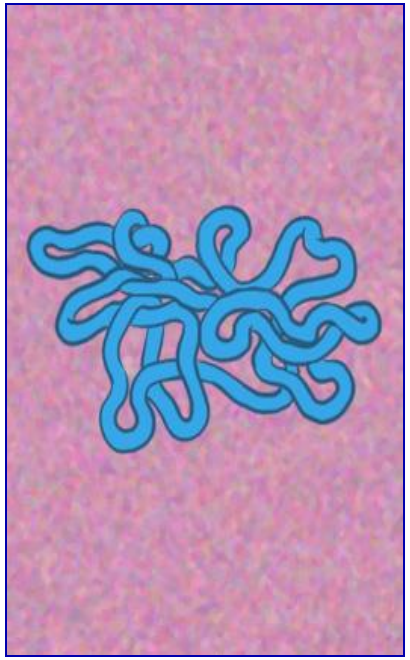
pwCF = people with CF.

Dave K, et al. *Pediatr Pulmonol*. 2021;56(Suppl 1):S79-S89.

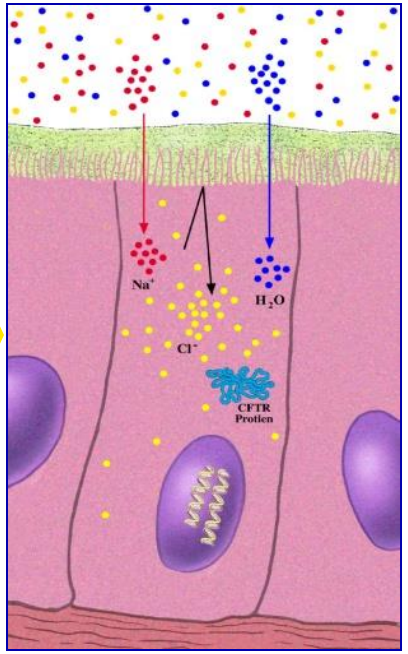
CF Pathophysiology

Abnormal Gene

Abnormal Protein

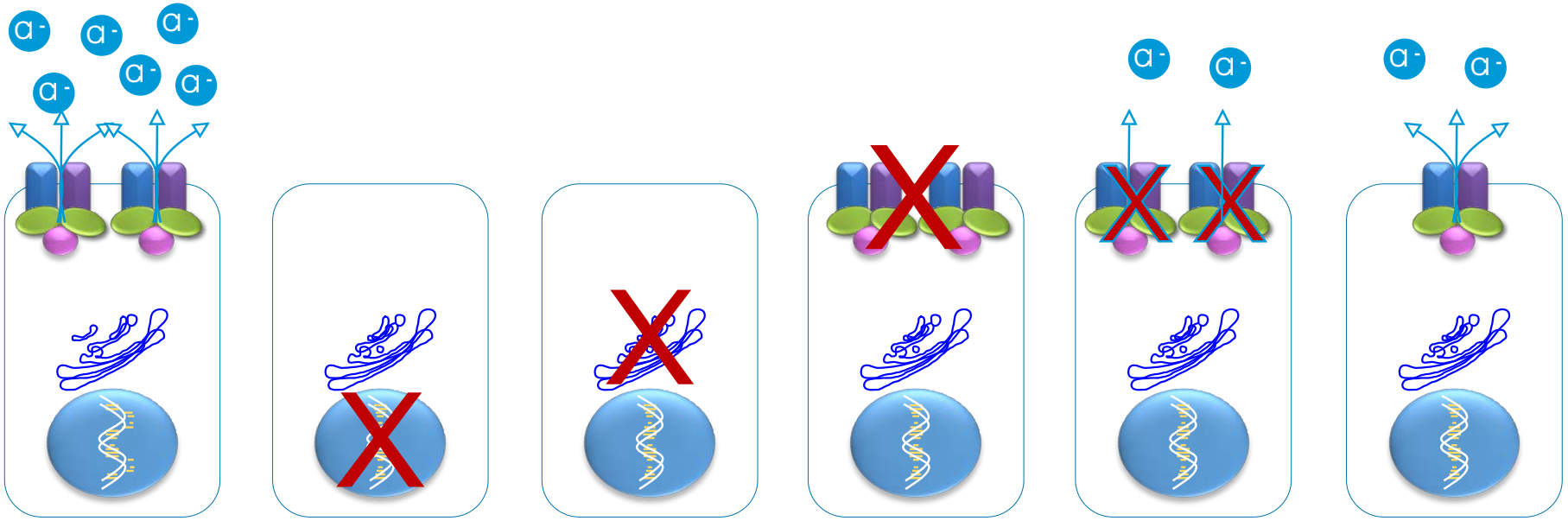


Altered Ion Transport (Cl^- and $HC0^-_3$) & Abnormal Mucus



Practical Updates in Primary Care

6 Classes of CFTR Mutations



	Normal	Class I	Class II	Class III	Class IV	Class V
DESCRIPTION	CFTR is created, reaches cell surface and functions properly, allowing transfer of chloride and water.	No functional CFTR created.	CFTR protein is created, but misfolded, keeping it from reaching the cell surface.	CFTR protein is created and reaches cell surface, but does not function properly.	The opening in the CFTR protein ion channel is faulty.	CFTR is created in insufficient quantities.
EXAMPLES		G542X W1282X R553X	F508del N1303K I507del	G551D S549N V520F	R117H D1152H R347P	3849+10kbC->T 2789+5G->A A455E

Adapted from: http://www.umd.be/CFTR/W_CFTR/gene.html.

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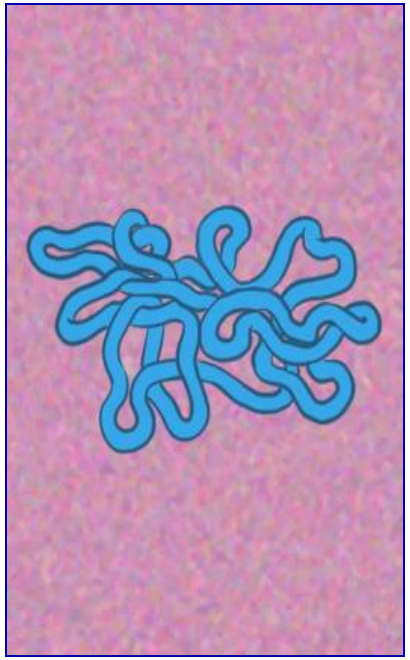
CF Pathophysiology

Abnormal Gene

Abnormal Protein



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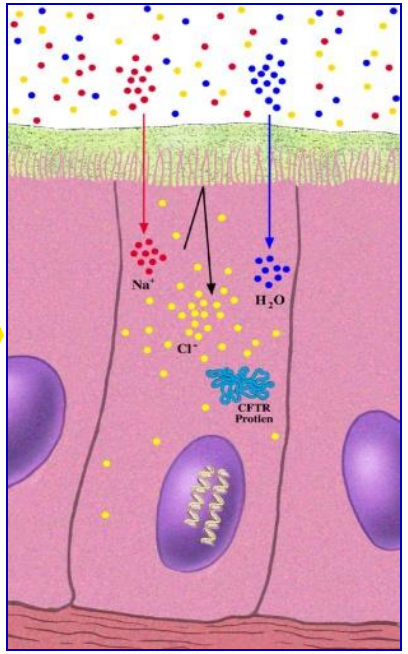
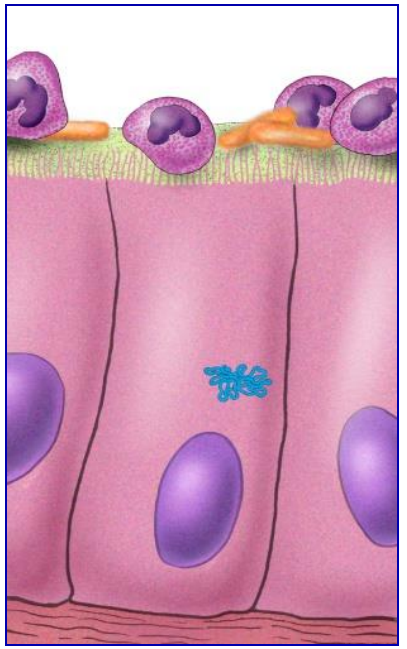


Infection & Inflammation



Organ Destruction & Respiratory Failure

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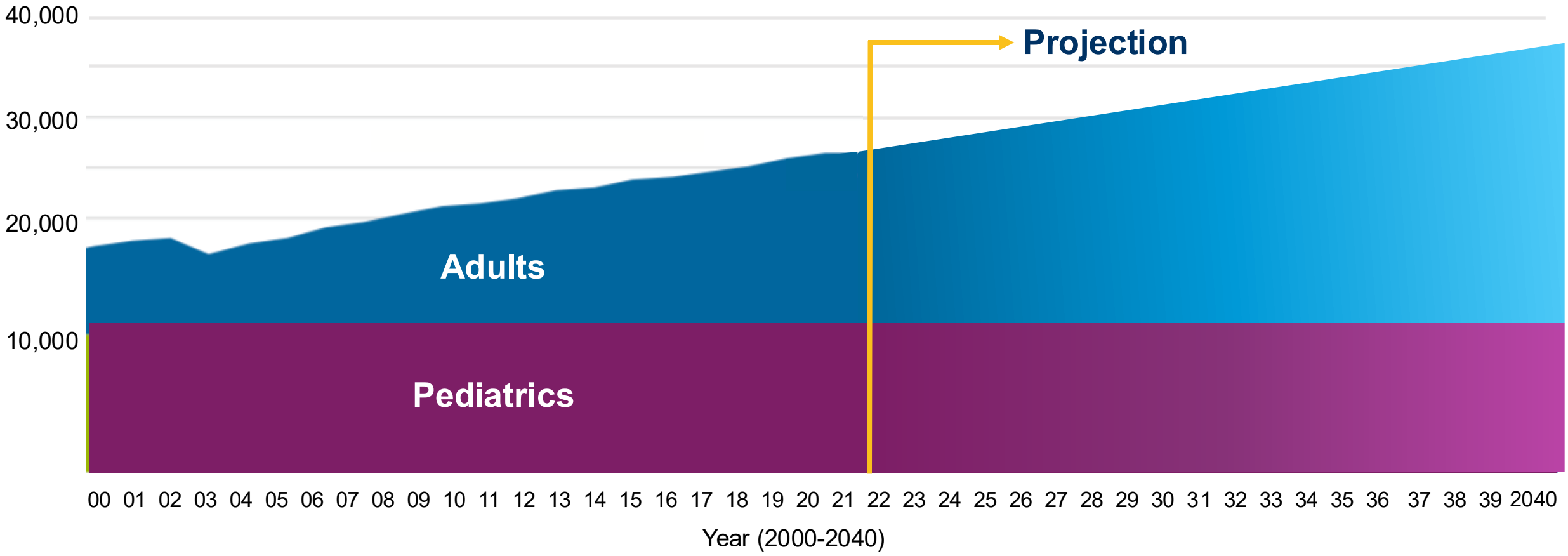
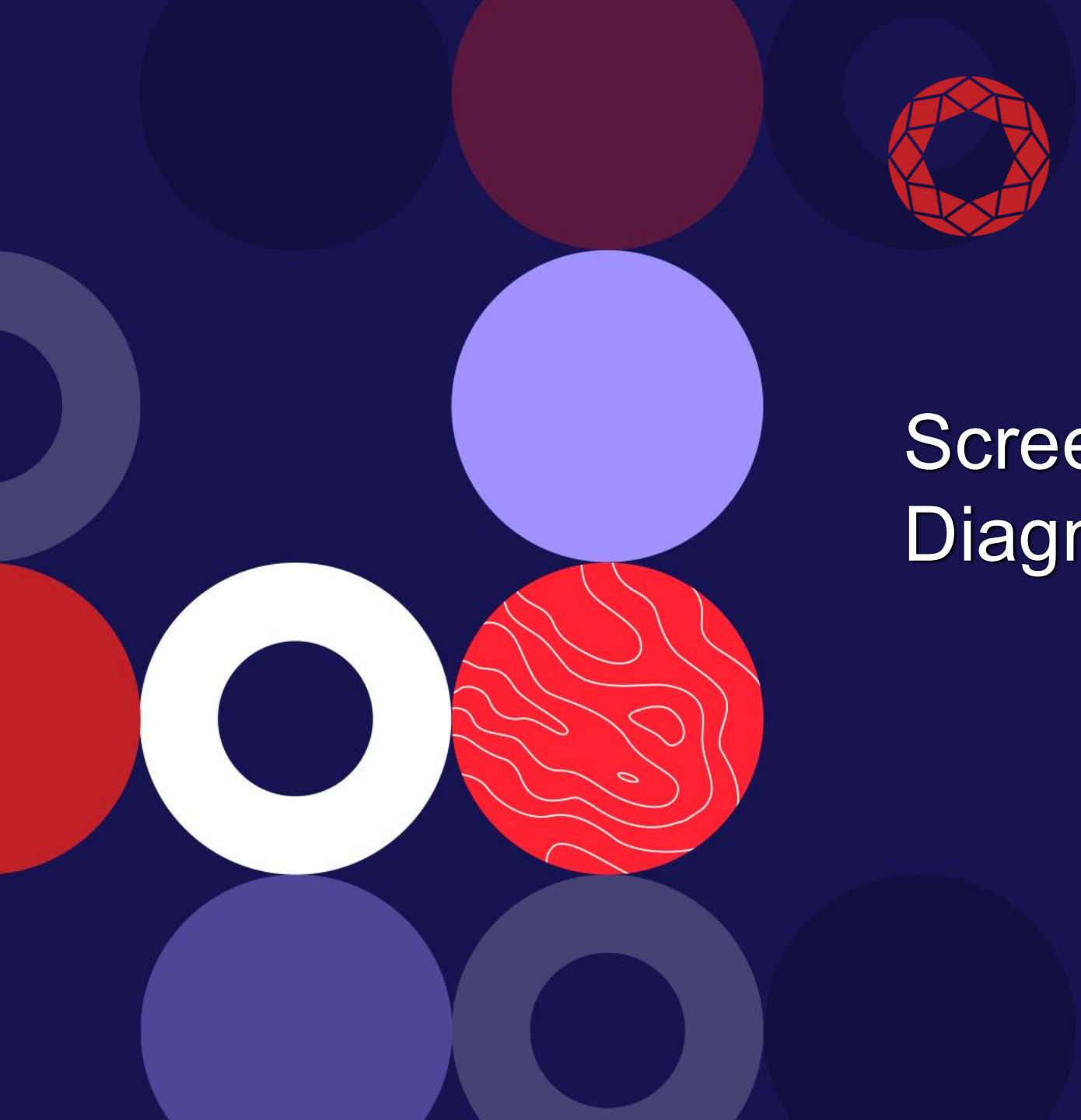
Projected Change in the US CF Population



Tullis L, et al. Presented at: NACFC; November 3-5, 2022; Philadelphia, PA.



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Screening & Diagnosis Guidelines

Sweat Chloride Test – Confirmation of Diagnosis

Sweat tubule defect discovered in 1952 after New York's August 1948 heat wave

Quantitative pilocarpine iontophoresis

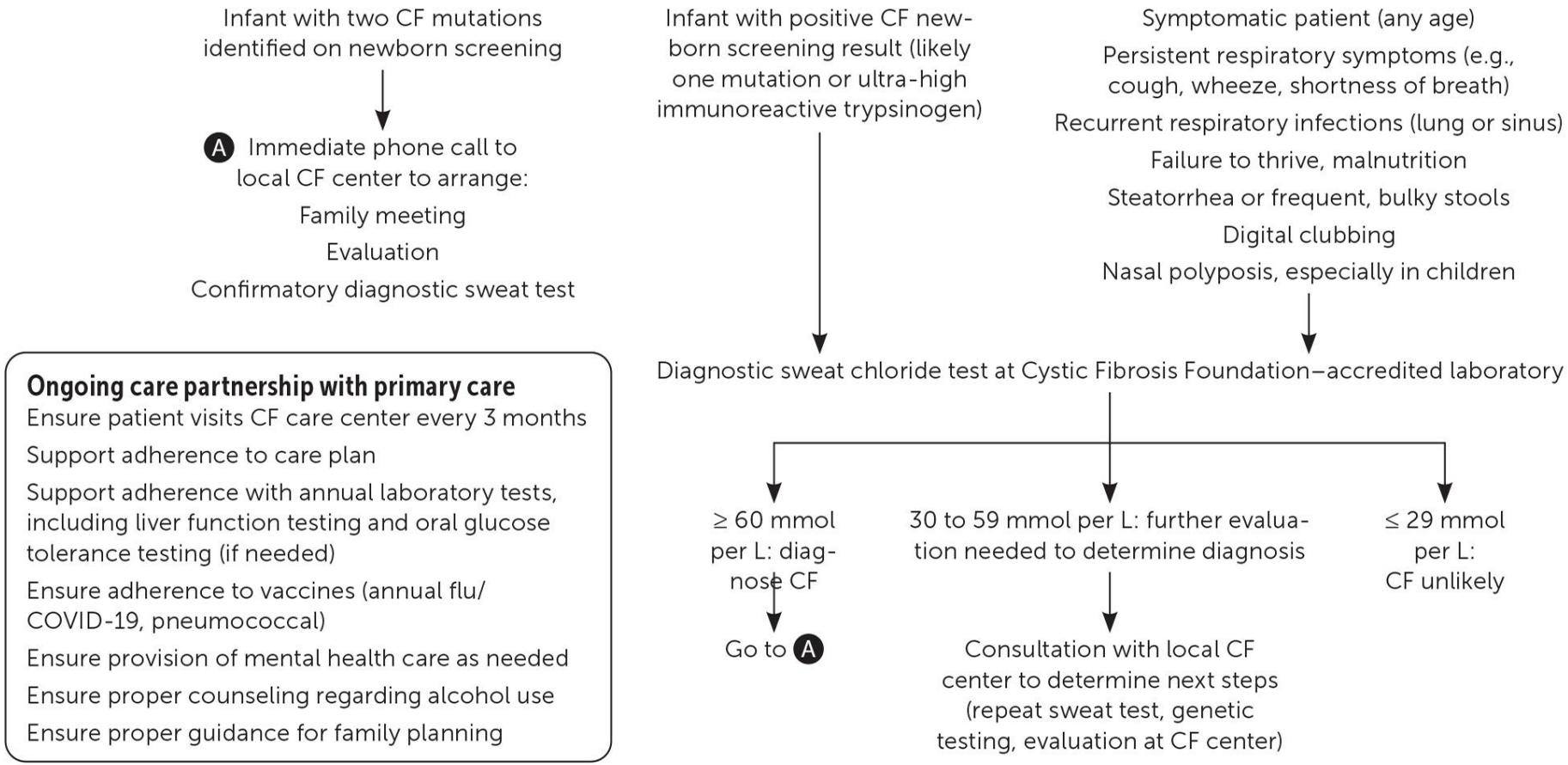
Traditional value for abnormal: >60 mmol/L

But <30 mmol/L is normal

Thus, any sweat chloride >30 mmol/L is consistent with a CF diagnosis as has been demonstrated in many infants with 2 pathogenic variants

Gibson LE, Cooke RE. *Pediatrics*. 1959;23(3):545-549. Farrell PM, Koscik RE. *Pediatrics*. 1996;97(4):524-528.

Diagnosis of CF and How PCPs Partner with Care



Note: This algorithm demonstrates a systematic process for diagnosing CF in patients identified by newborn screening or symptoms. In addition, it highlights key roles for the primary care physician in partnership with the patient and the Cystic Fibrosis Foundation. **Age and ethnicity should not be a barrier or consideration in proceeding with diagnostic testing.**

PCPs = primary care providers.

Collins MS, Mansilla-Rivera K. *Am Fam Physician*. 2024;109(5):388-390.

Practical Updates in Primary Care

Why Add CF to Newborn Screen?

Children who receive CF care early in life have better nutrition and are healthier than those who are diagnosed later

Early diagnosis and treatment can

Improve growth	Help improve lung health	Reduce hospital stays	Identify and eradicate bacteria early	Add years to life

CFF. Accessed April 24, 2026. https://www.cff.org/intro-cf/newborn-screening-cf.

What Is the Test?

All states start with testing the level of immunoreactive trypsinogen

Enzyme made by the pancreas and is normally in low levels

CF can cause elevated levels, but so can other conditions…

Some states reflex +IRT to a DNA test

Each state has a panel of genetic variants that it looks for in baby's DNA

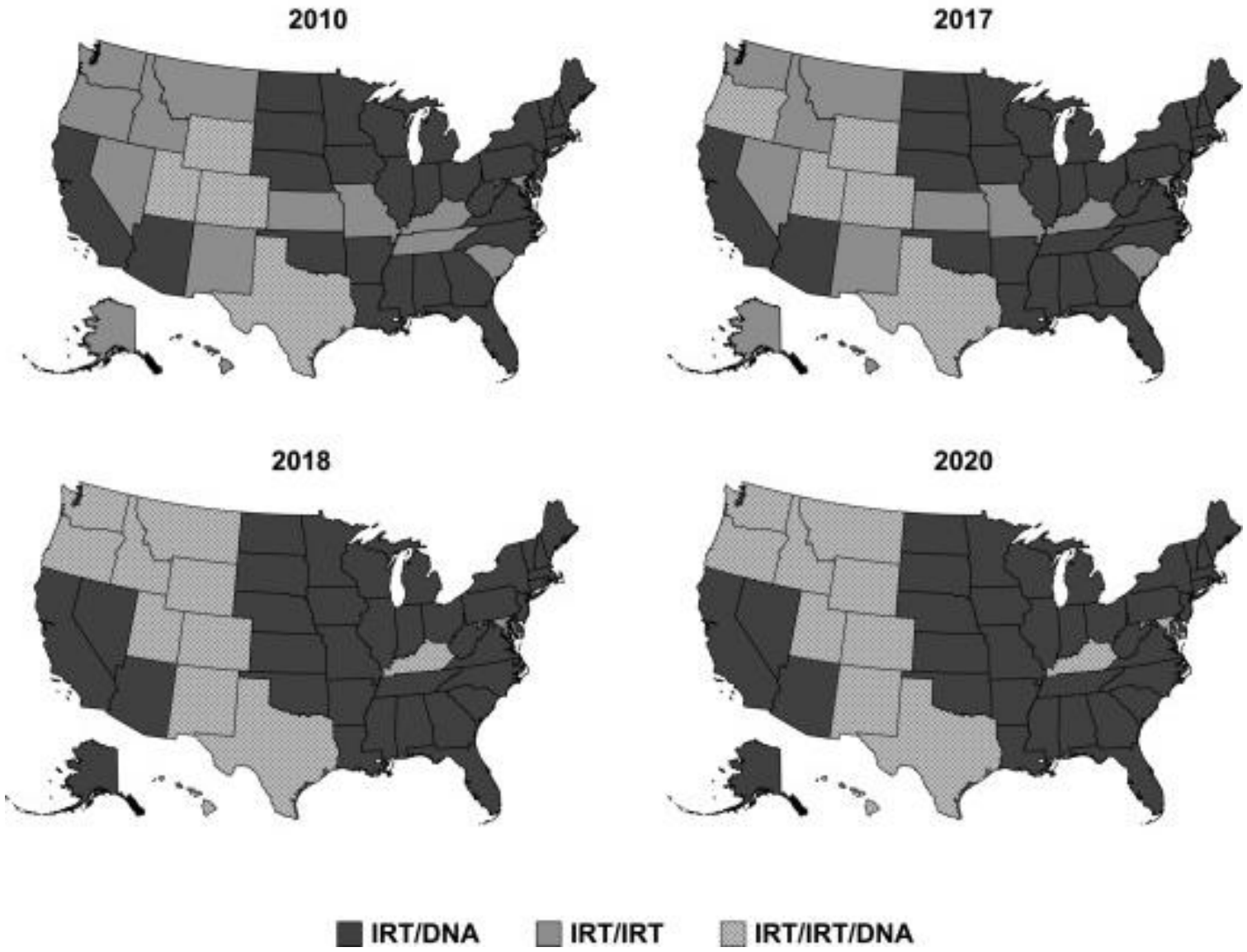
What type of testing does Massachusetts do?

* IRT → DNA (39-gene panel)

IRT = immunoreactive trypsinogen.

CFF. Accessed April 24, 2026. https://www.cff.org/intro-cf/newborn-screening-cf.

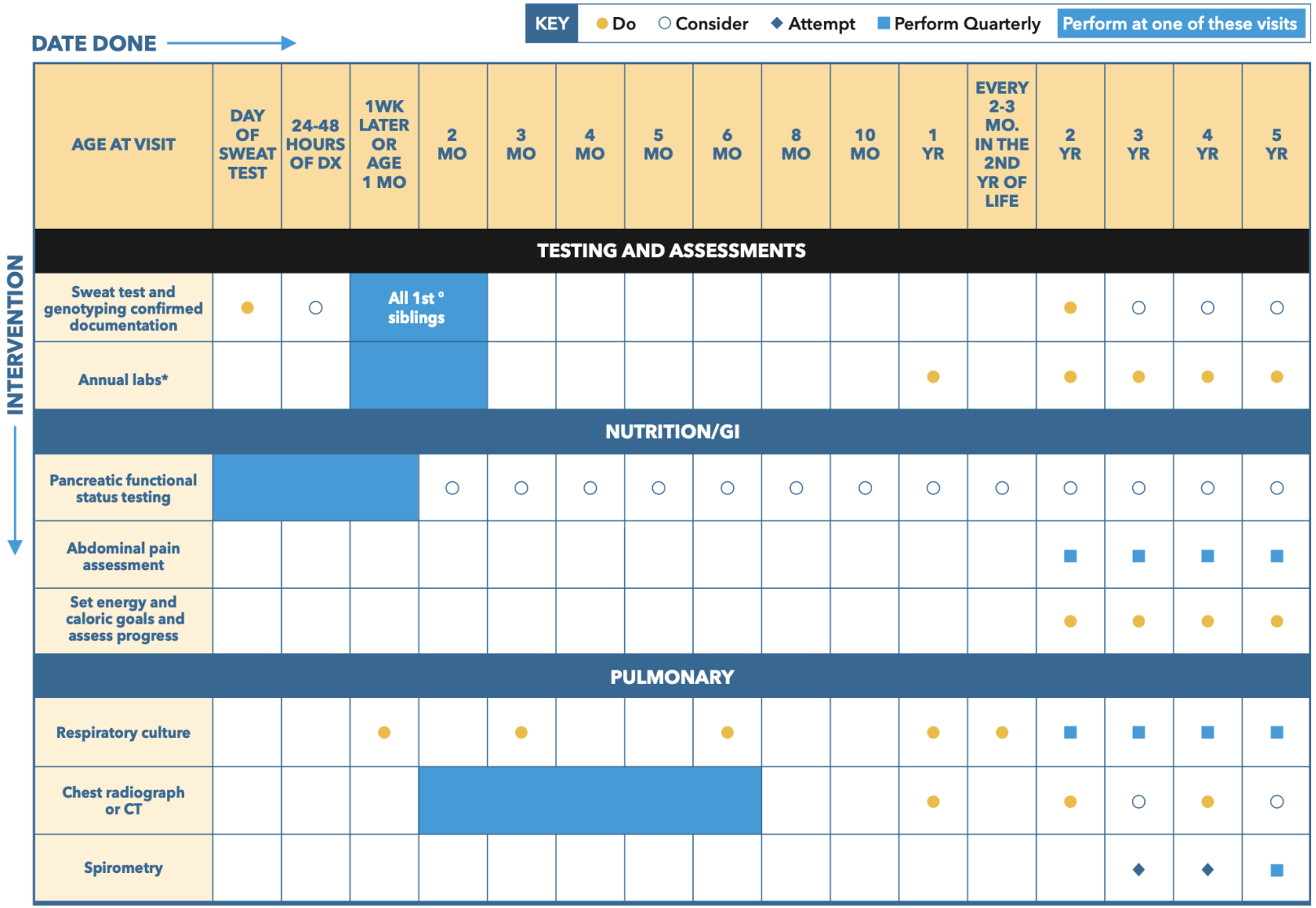
Evolution of CF Newborn Screening by Test



Farrell PM. *Mol Genet Metab*. 2023;140(1-2):107679.

Practical Updates in Primary Care

Clinical Care Guidelines for Newborns to 5 Years



KEY: ● Do ○ Consider ◆ Attempt ■ Perform Quarterly — Perform at one of these visits

DATE DONE →

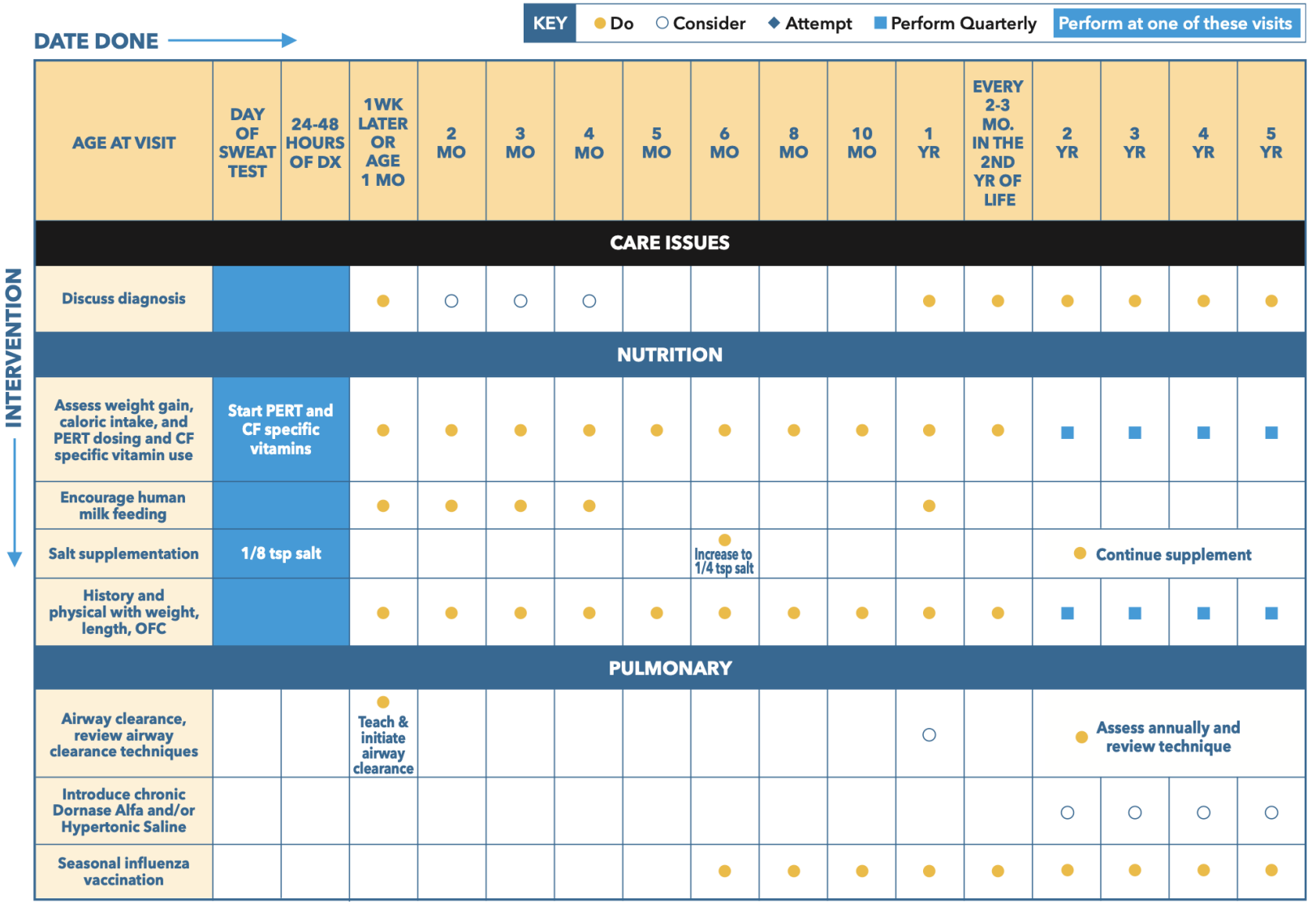
INTERVENTION ↓

AGE AT VISIT	DAY OF SWEAT TEST	24-48 HOURS OF DX	1WK LATER OR AGE 1 MO	2 MO	3 MO	4 MO	5 MO	6 MO	8 MO	10 MO	1 YR	EVERY 2-3 MO. IN THE 2ND YR OF LIFE	2 YR	3 YR	4 YR	5 YR
TESTING AND ASSESSMENTS																
Sweat test and genotyping confirmed documentation	●	○	All 1st ° siblings										●	○	○	○
Annual labs*			■	■	■	■	■	■	■	■	●		●	●	●	●
NUTRITION/GI																
Pancreatic functional status testing	■	■	■	○	○	○	○	○	○	○	○	○	○	○	○	○
Abdominal pain assessment													■	■	■	■
Set energy and caloric goals and assess progress													●	●	●	●
PULMONARY																
Respiratory culture			●		●			●			●	●	■	■	■	■
Chest radiograph or CT				■	■	■	■	■			●		●	○	●	○
Spirometry														◆	◆	■

CFF. Accessed April 24, 2026. https://www.cff.org/medical-professionals/infant-care-clinical-care-guidelines/.

Practical Updates in Primary Care

Clinical Care Guidelines for Newborns to 5 Years



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CARE ISSUES																
Discuss diagnosis			●	○	○	○					●	●	●	●	●	●
NUTRITION																
Assess weight gain, caloric intake, and PERT dosing and CF specific vitamin use	Start PERT and CF specific vitamins		●	●	●	●	●	●	●	●	●	●	■	■	■	■
Encourage human milk feeding			●	●	●	●					●					
Salt supplementation	1/8 tsp salt							● Increase to 1/4 tsp salt					● Continue supplement			
History and physical with weight, length, OFC			●	●	●	●	●	●	●	●	●	●	■	■	■	■
PULMONARY																
Airway clearance, review airway clearance techniques			● Teach & initiate airway clearance								○		● Assess annually and review technique			
Introduce chronic Dornase Alfa and/or Hypertonic Saline													○	○	○	○
Seasonal influenza vaccination								●	●	●	●	●	●	●	●	●

*Annual labs include: Vitamin levels A,D, E, prothrombin time, serum electroytes BUN creatine glucose, complete blood count, AST/ALT/GGT/ Bili, albumin, ALP

CFF. Accessed April 24, 2026. https://www.cff.org/medical-professionals/infant-care-clinical-care-guidelines/.

Practical Updates in Primary Care

Specific Areas of Focus for PCPs

Expedite follow-up of positive NBS tests

Nutrition, growth, and development
- Encourage/support breastfeeding for the first 6 months
- Ensure salt supplements are taken daily (potentially life-saving)
- Inform the CF center about poor growth

Respiratory infection
- Recognize subtle early symptoms (insidious lung disease onset)
- Notify the CF center of changes such as persistent cough
- Employ a lower threshold for antibiotics prescription, and prescribe longer courses

Anticipatory guidance and counseling
- Inquire about adherence to CF treatments at each primary care visit
- Discuss the importance of adequate dietary sources of Ca, vitamin D, and fat-soluble vitamins
- Convey optimism and hope while encouraging full, high-quality lives

Psychosocial
- Expect CF to create psychosocial challenges (family stress)
- Emphasize that contact with other patients with CF increases risk for cross-infections
- Explain reproductive aspects with parental input at an appropriate age

Terlizzi V, Farrell PM. *Curr Probl Pediatr Adolesc Health Care*. 2024;54(6):101637.

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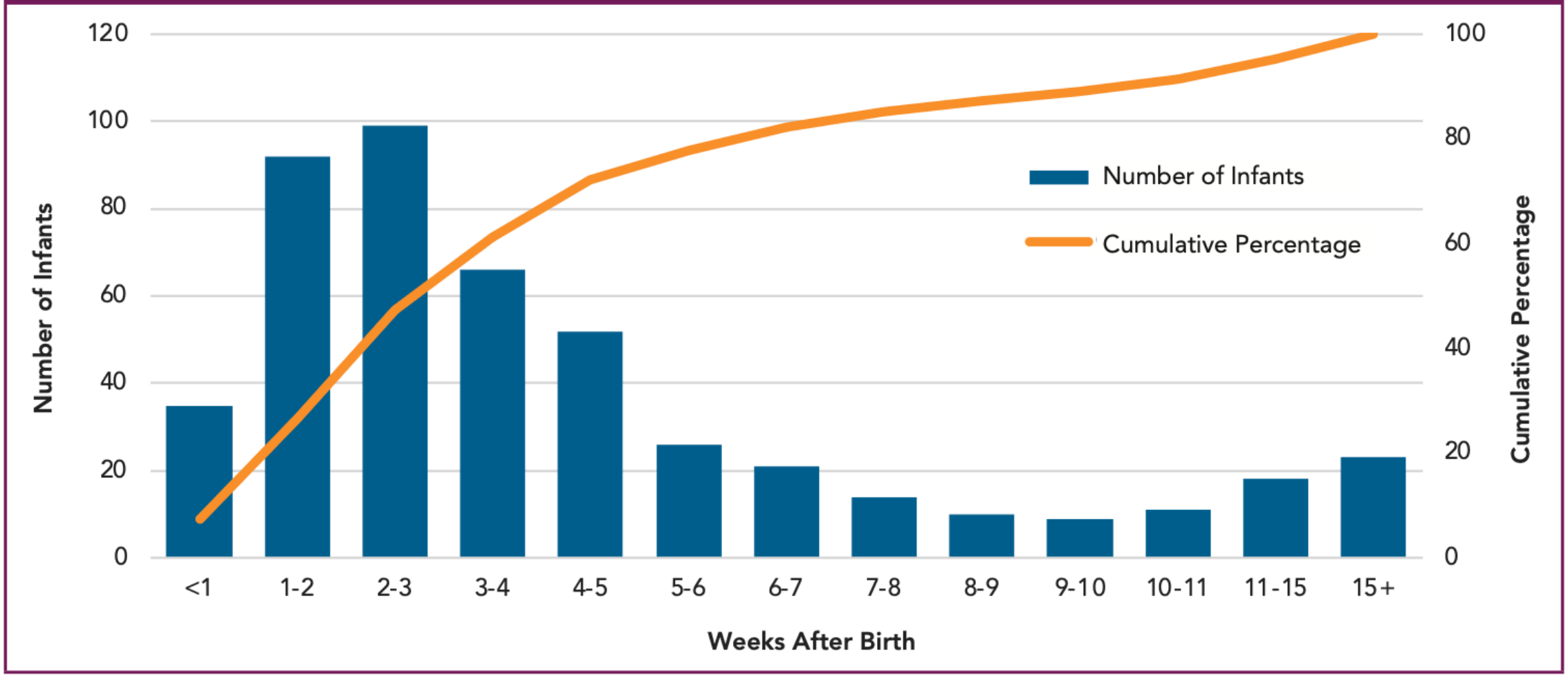
Time to First Clinic Encounter, Care Episode, or Sweat Test for Infants With CF Born in 2023 and Detected by Newborn Screening (n=487)



CFF. Accessed April 27, 2026. https://www.cff.org/sites/default/files/2024-09/2023-Patient-Registry-Annual-Data-Report.pdf.

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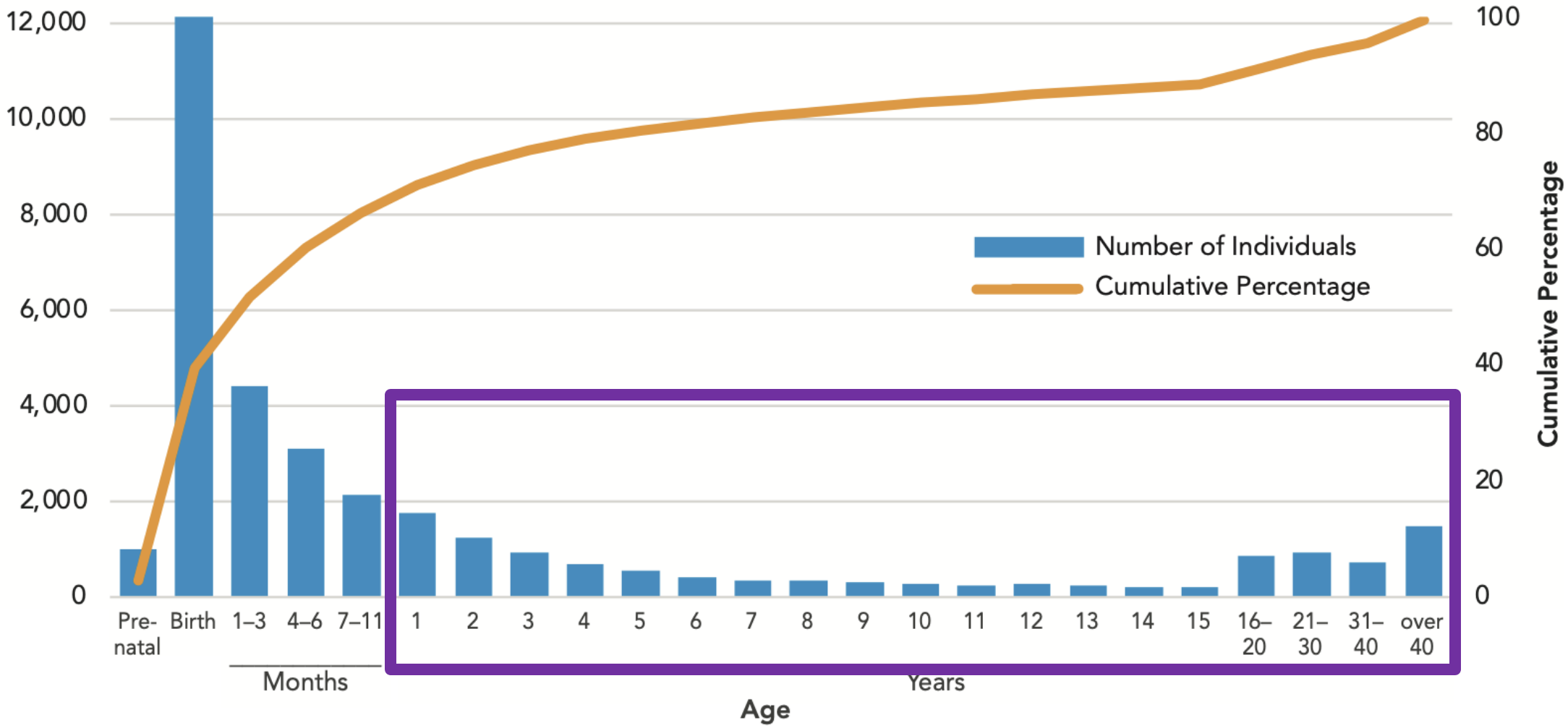
Age of Diagnosis of All Individuals Seen with CF in 2024



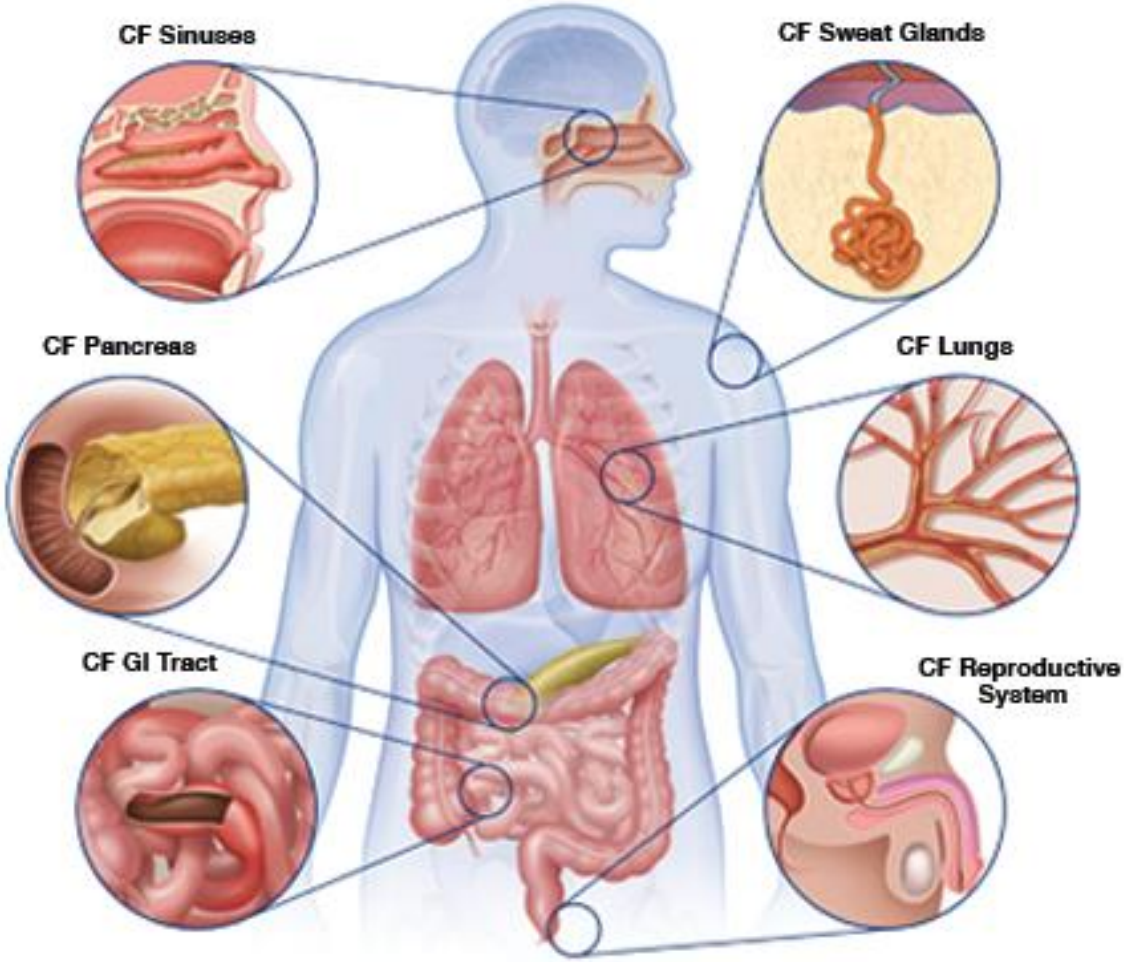
CFF. Accessed April 27, 2026. https://www.cff.org/sites/default/files/2024-09/2023-Patient-Registry-Annual-Data-Report.pdf.

2024 CF Registry Report – Symptoms at Diagnosis

Symptoms Reported at CF Diagnosis	All Individuals (%)	Diagnosed in 2024 (%)	Diagnosed in 2024 Age < 1 (%)[A]	Diagnosed in 2024 Age ≥ 1 (%)
Number of Individuals (n)	**33,989**	**843**	**520**	**323**
Asymptomatic				
DNA analysis	15.8	31.6	29.1	35.6
Family history	13.8	10.8	10.7	11.2
Newborn (neonatal) screening	32.7	55.8	86.2	N/A
Prenatal screening (CVS[B], amniocentesis)	2.7	4.7	7.3	N/A
Symptomatic				
Acute or persistent respiratory abnormalities	31.9	17.6	1.2	44.0
CBAVD[C] or infertility/GU[D] abnormalities	1.0	3.9	<0.1	10.2
Digital clubbing	0.6	1.2	<0.1	3.1
Edema	0.5	0.1	<0.1	0.3
Electrolyte imbalance	2.5	0.1	<0.1	0.3
Failure to thrive/malnutrition	23.5	5.9	3.5	9.9
Liver problems	0.9	0.2	<0.1	0.6
Meconium ileus/other intestinal obstruction	15.6	7.4	11.5	N/A
Nasal polyps/sinus disease	3.8	4.5	<0.1	11.8
Rectal prolapse	2.2	0.6	0.2	1.2
Steatorrhea/abnormal stools/malabsorption	18.2	4.6	3.1	7.1
Other	5.5	8.8	1.7	20.1

CFF. Accessed April 27, 2026. https://www.cff.org/media/38406/download?inline.

Late CF Clinical Manifestations



- CBAVD
- Recurrent/Chronic Pancreatitis
- Nasal polyposis/sinusitis
- Bronchiectasis/recurrent respiratory infections
- CF associated organisms
- ABPA
- Digital clubbing
- Diabetes
- Heat/electrolyte imbalance/cramping
- Liver disease
- Osteoporosis/osteopenia
- DIOS/recurrent constipation
- Colorectal cancer/other GI malignancy
- CAD
- Family History

Adapted from: Wilmott R, et al. *Kendig's Disorders of the Respiratory Tract in Children*. 9th ed. Elsevier; 2019.

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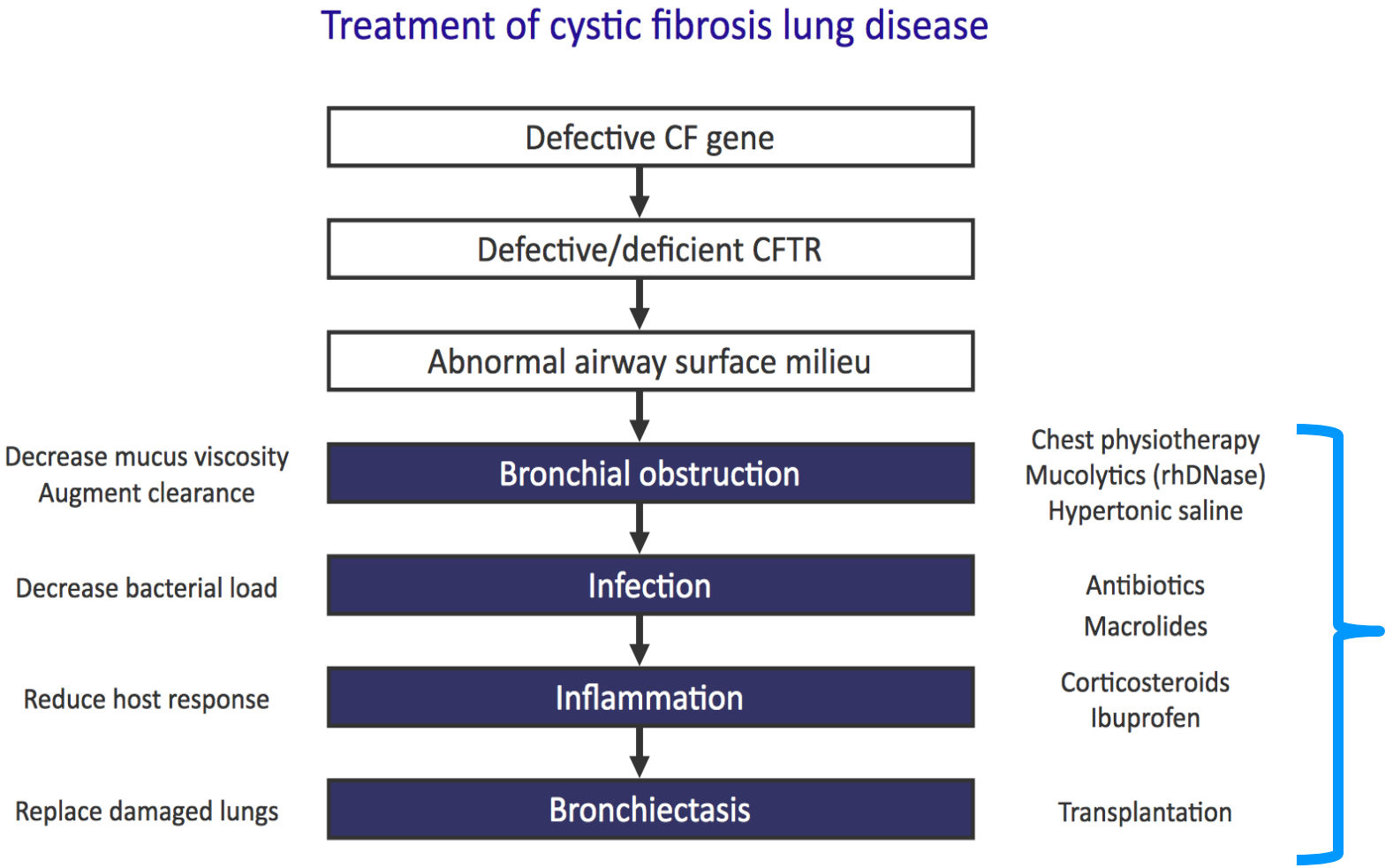
Targeted Therapies

Traditional Therapeutic Approaches for CF Lung Disease



Until recently, treatments focused here

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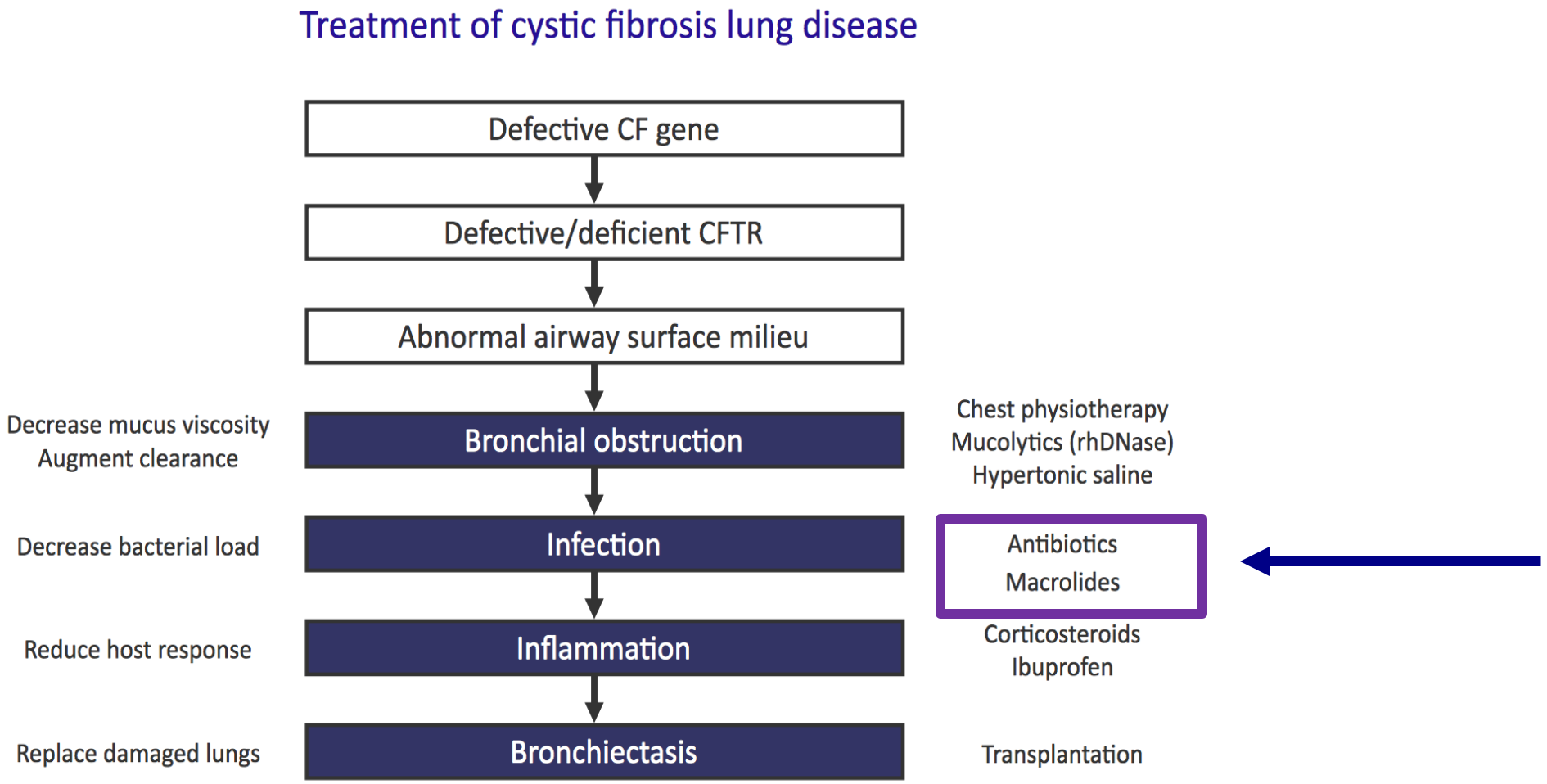
Davis PB, et al. *Am J Respir Crit Care Med*. 1996;154(5):1229-1256.

Traditional Therapeutic Approaches for CF Lung Disease



Davis PB, et al. *Am J Respir Crit Care Med*. 1996;154(5):1229-1256.

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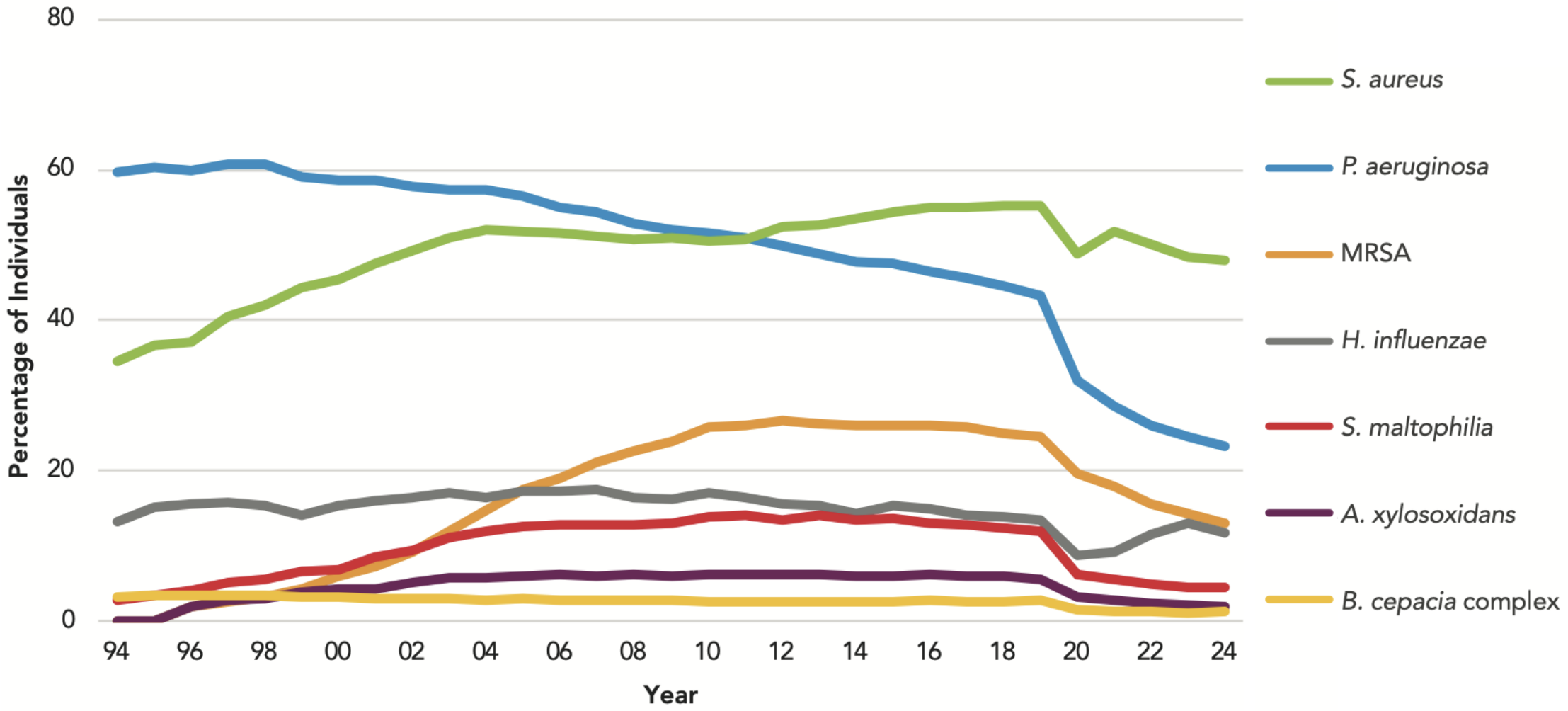
Prevalence of Respiratory Microorganisms in 2024



CFF 2024 Annual Registry Report

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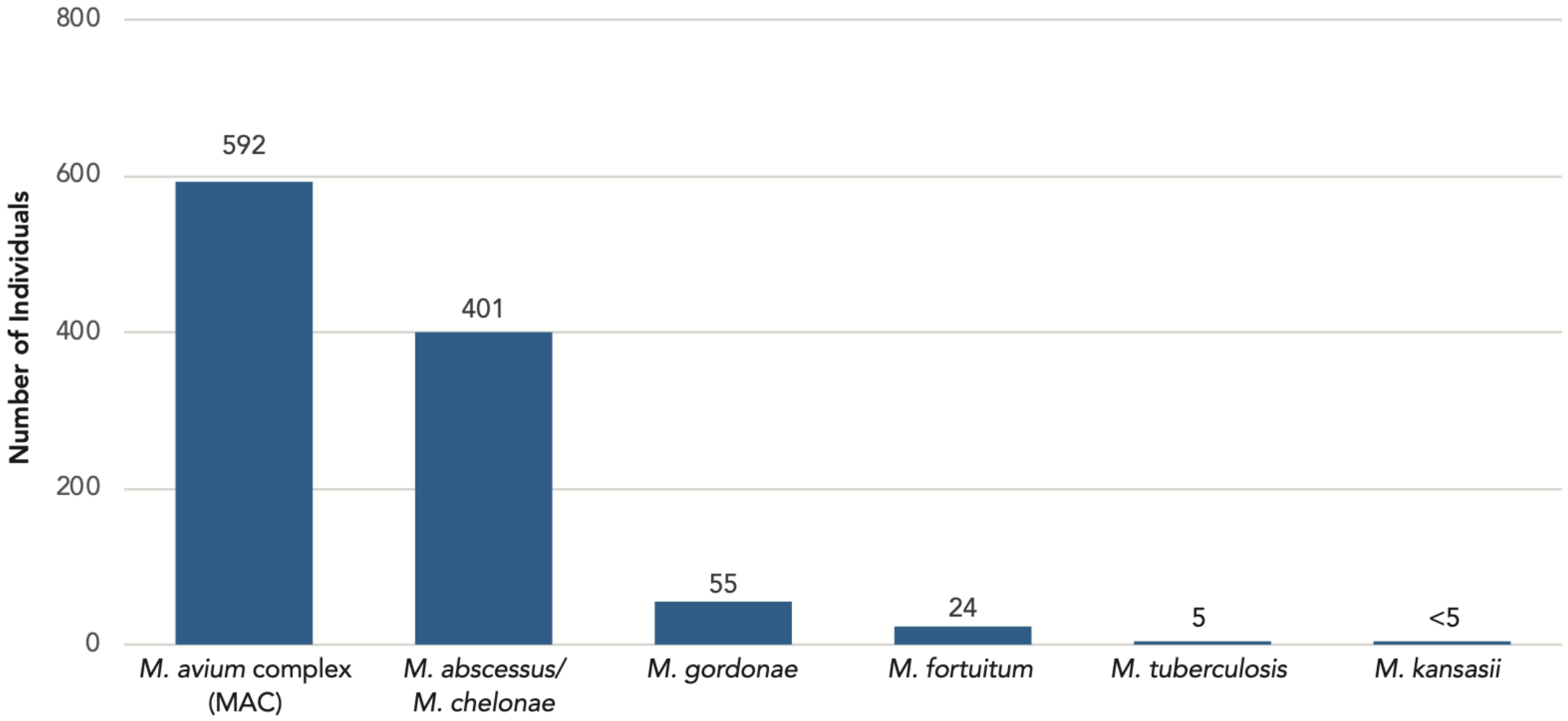
Non-Tuberculous Mycobacteria in 2024



CFF. Accessed April 27, 2026. https://www.cff.org/media/38406/download?inline.

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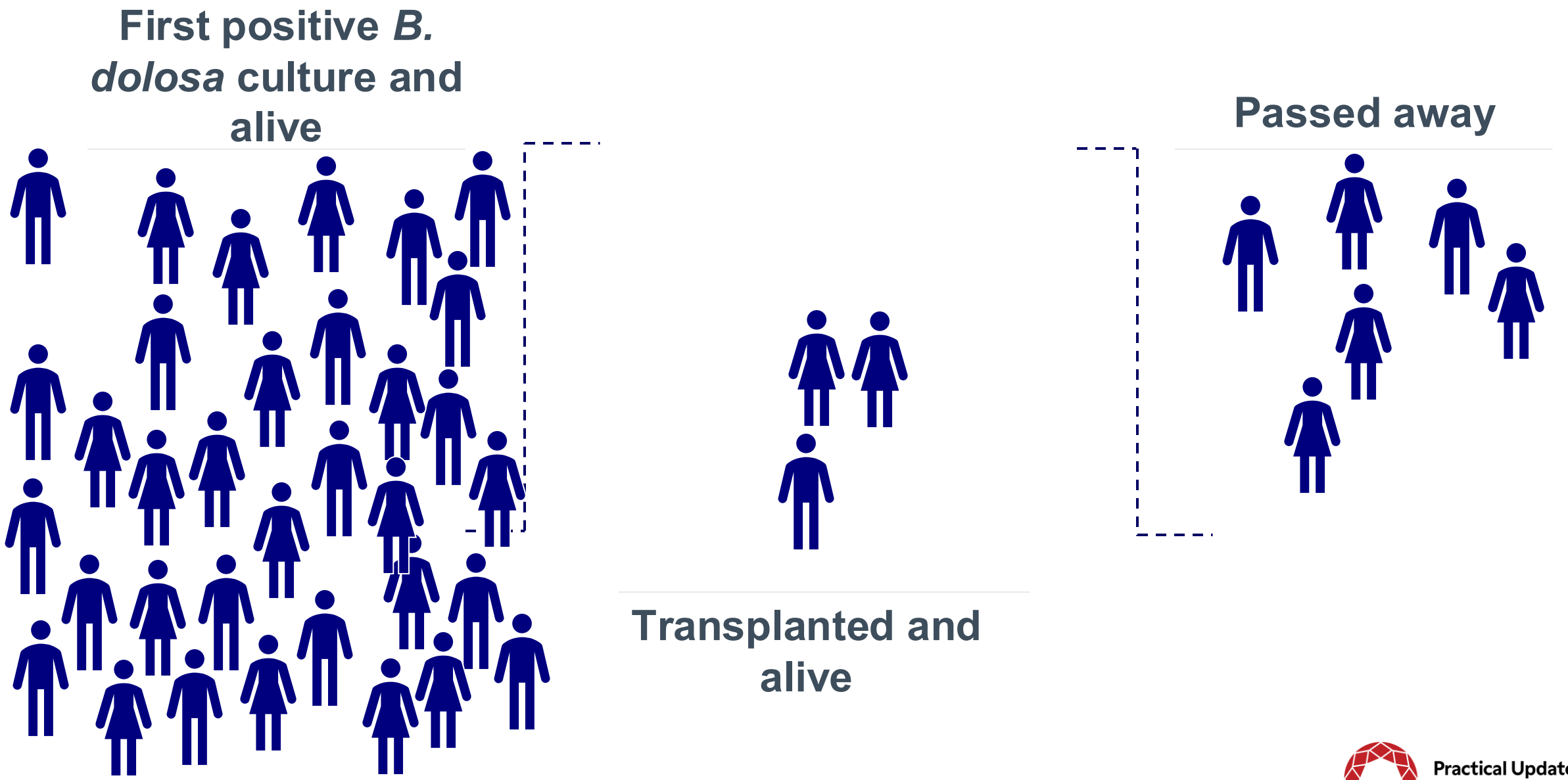
“Cepacia Syndrome”

- Fatal combination of necrotizing pneumonia, worsening respiratory failure, and bacteremia
- Previously associated with genomovar III (*B. cenocepacia*) but documented with other BCC organisms as well
- Bacteremia typically irreversible
- Rapid pulmonary deterioration
  - Pleural effusion
  - Necrotizing pneumonia and lung abscess
- Death
  - 62-100% of patients

BCC = Burkholderia cepacia complex.

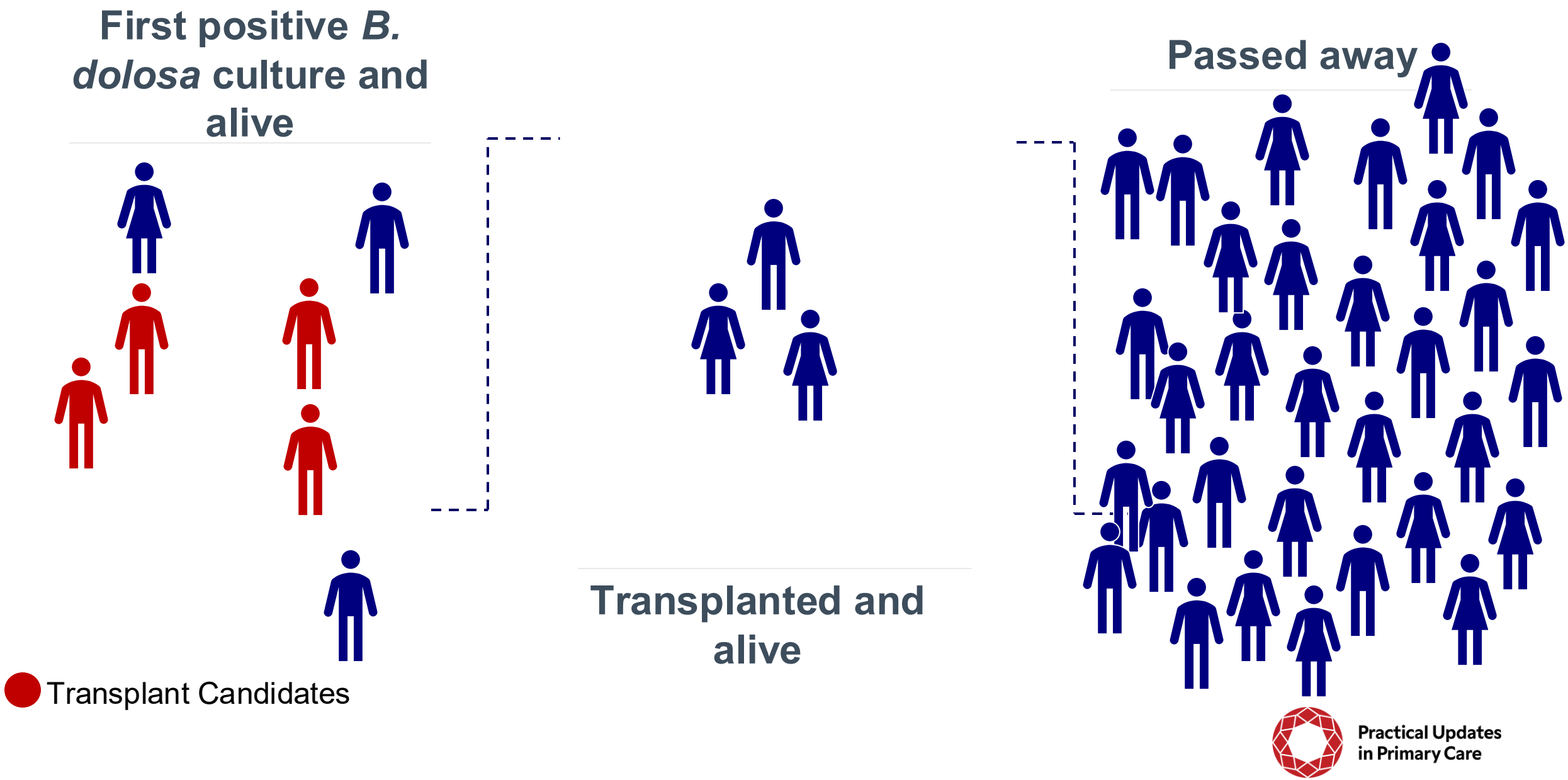
Kalferstova L, et al. J Clin Microbiol. 2015;53(5):1515-1522.

The Human Face of an Epidemic—2003

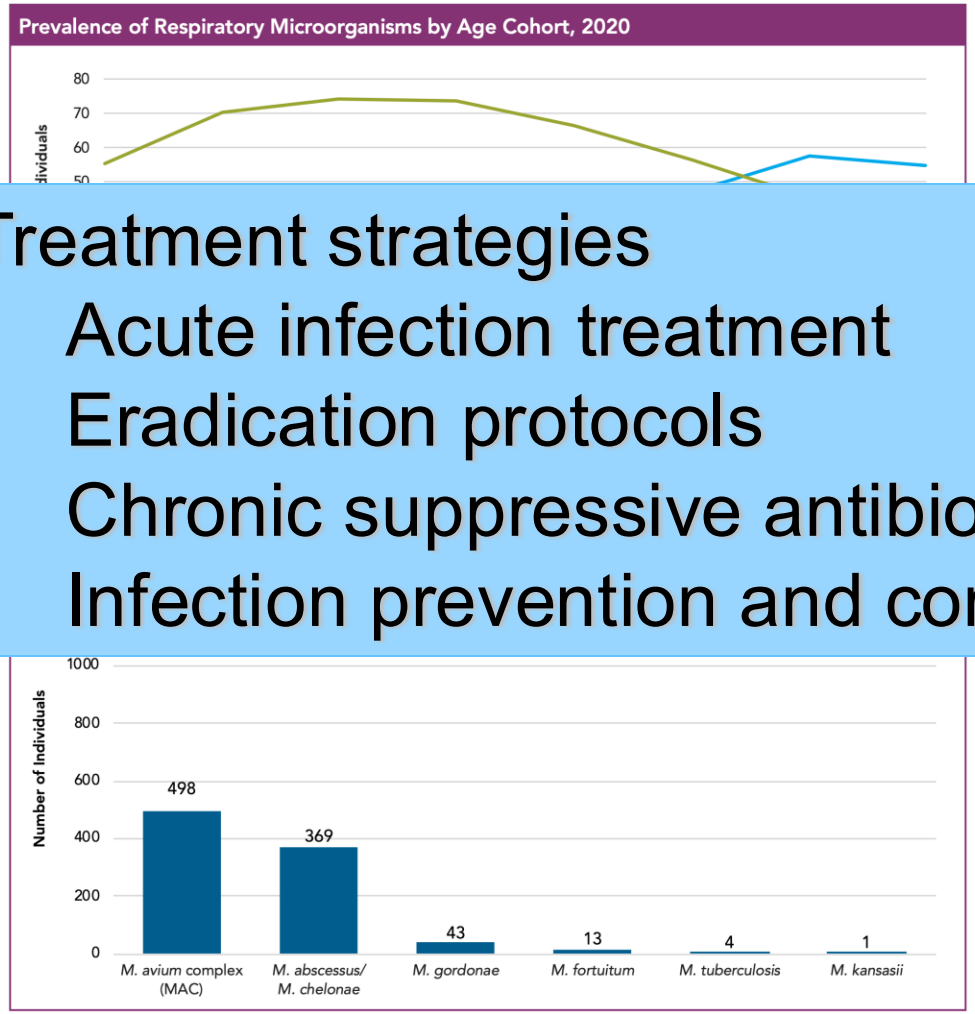


Practical Updates in Primary Care

Present Day

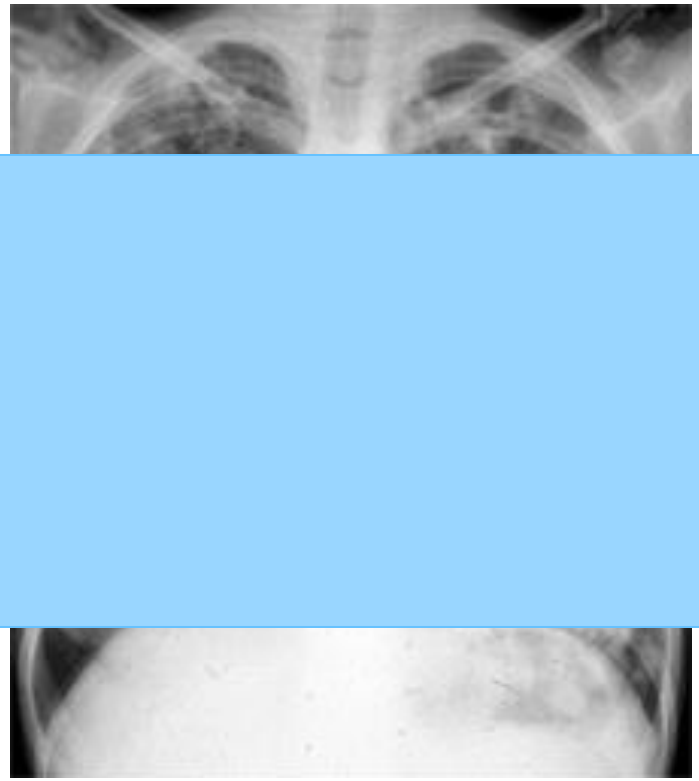

First positive *B. dolosa* culture and alive
Transplanted and alive
Passed away
Transplant Candidates
Practical Updates in Primary Care

Management of Acute Infectious Exacerbations



Treatment strategies

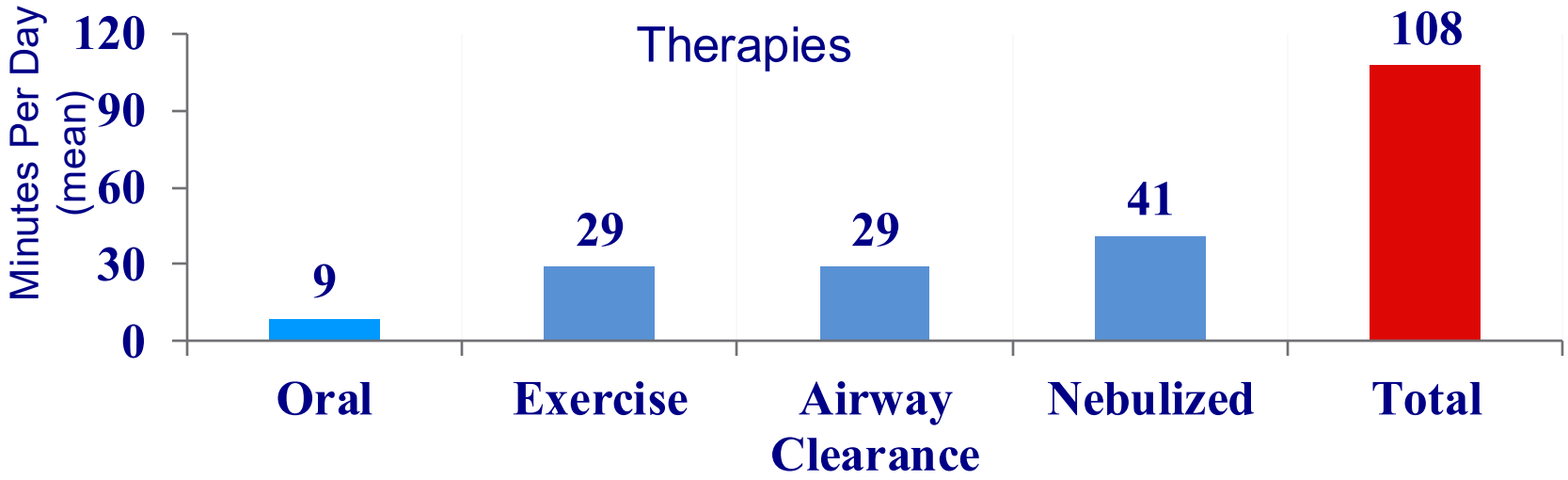
- Acute infection treatment
- Eradication protocols
- Chronic suppressive antibiotics
- Infection prevention and control



Pulmonary disease is major cause of morbidity and mortality

CFF. Accessed April 27, 2026. https://www.cff.org/sites/default/files/2021-11/2020%20Annual%20Data%20Report.pdf.

High Treatment Burden in CF



Medications	Median (Range)
# of Oral Medications	3 (0-7)
# of Nebulized Medications	2 (0-5)
# of Inhaled Medications (MDI)	1 (0-4)
# of Total Medications	7 (0-20)

CF-related observational and interventional studies

PROMISE

- Observational study looking at impact of triple combination CFTR modulator

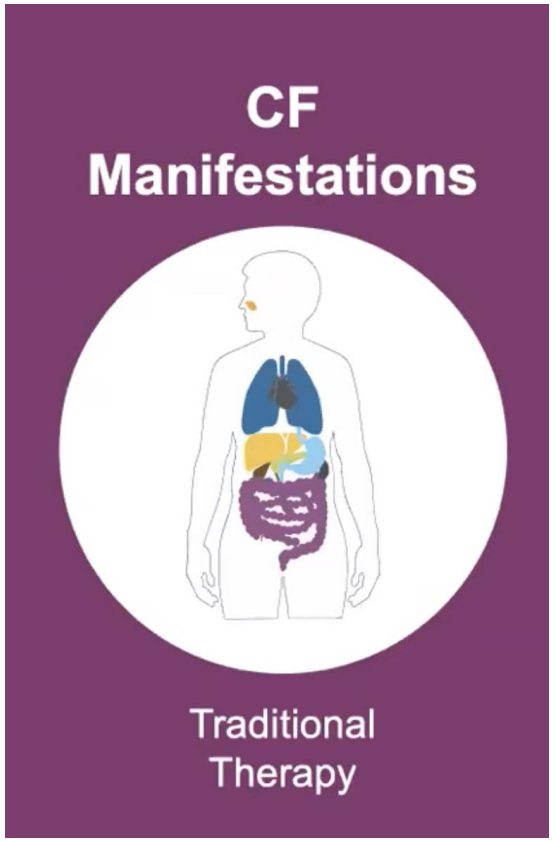
SIMPLIFY

- Interventional study of discontinuing either hypertonic saline or dornase alfa while on triple-combination CFTR modulator

MDI = metered dose inhaler.

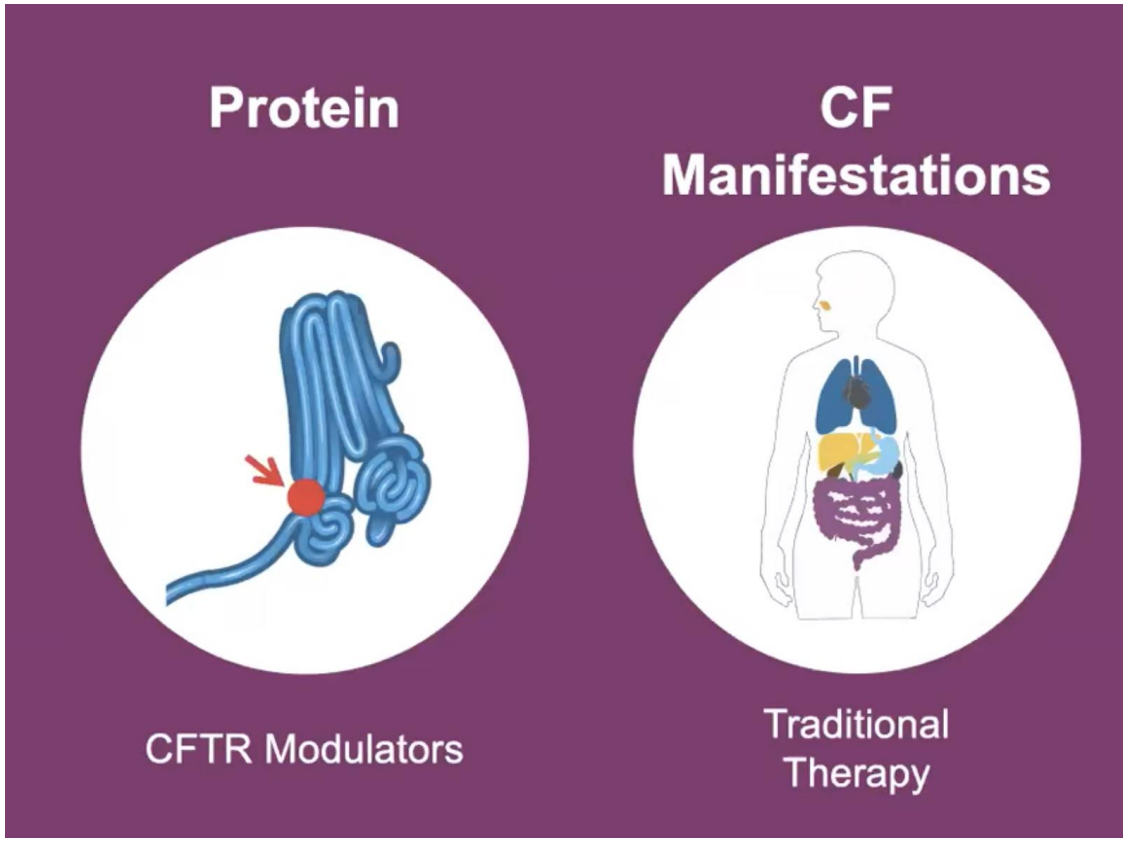
Sawicki GS, et al. *J Cyst Fibros*. 2009;8(2):91-96.

New Therapeutic Approach to CF



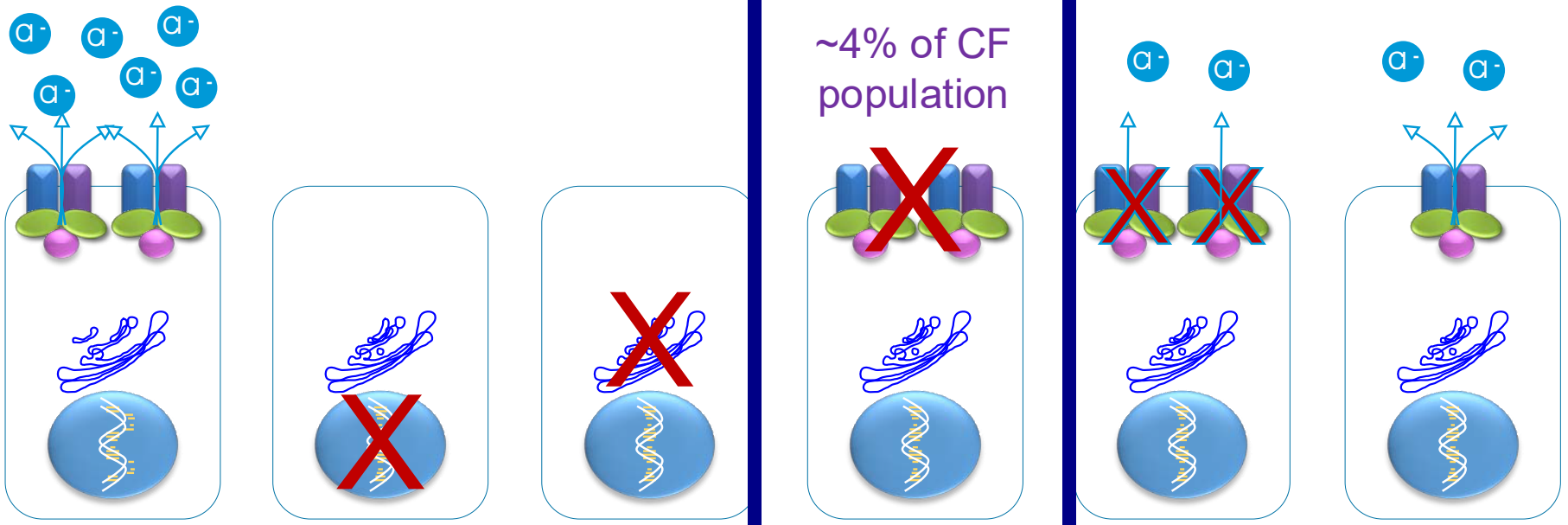
Courtesy of CFF.

New Therapeutic Approach to CF



Courtesy of CFF.

5 (or 6) Classes of CFTR Mutations

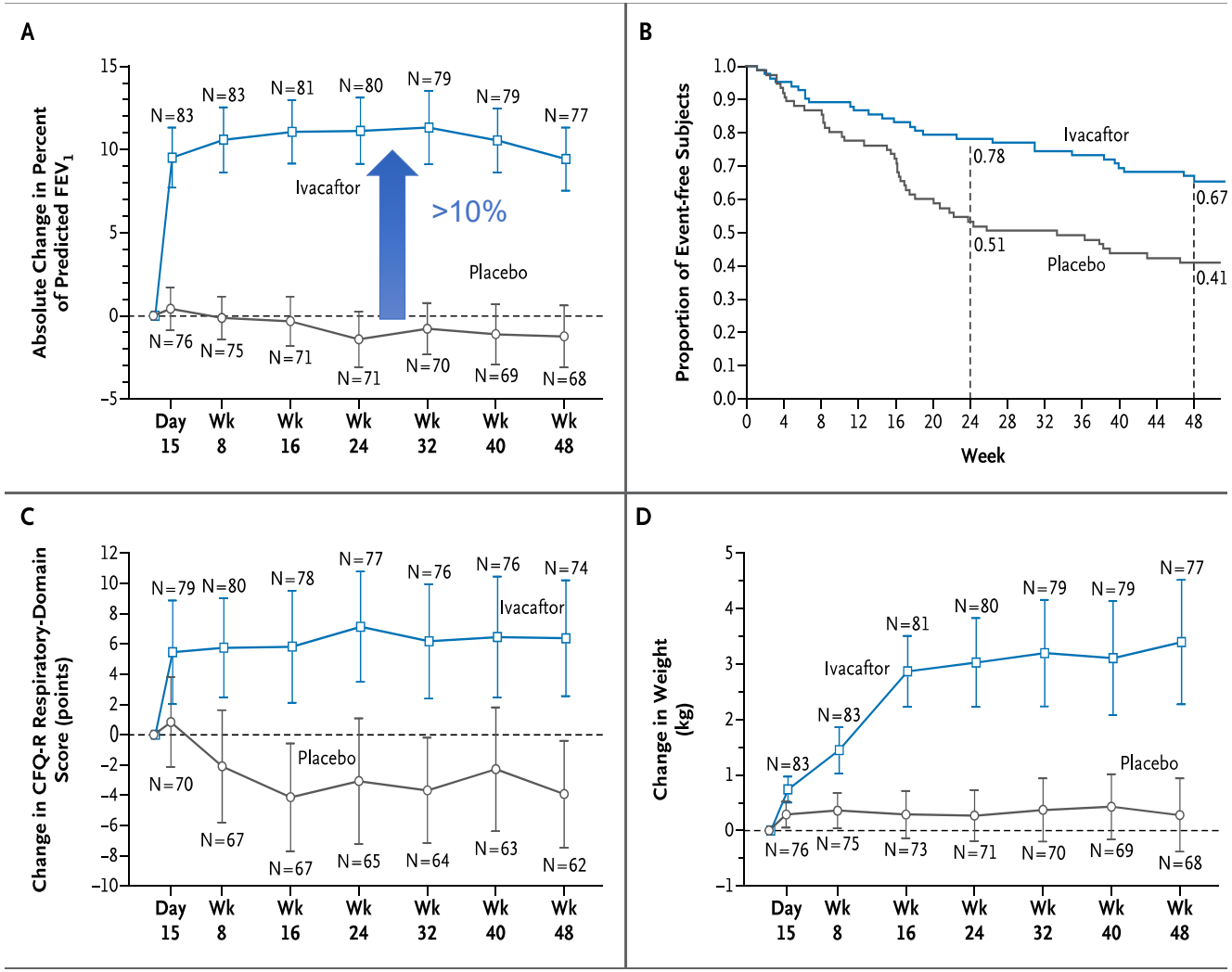


	Normal	Class I	Class II	Class III	Class IV	Class V
DESCRIPTION	CFTR is created, reaches cell surface and functions properly, allowing transfer of chloride and water.	No functional CFTR created.	CFTR protein is created, but misfolded, keeping it from reaching the cell surface.	CFTR protein is created and reaches cell surface, but does not function properly.	The opening in the CFTR protein ion channel is faulty.	CFTR is created in insufficient quantities.
EXAMPLES		G542X W1282X R553X	F508del N1303K I507del	G551D S549N V520F	R117H D1152H R347P	3849+10kbC->T 2789+5G->A A455E

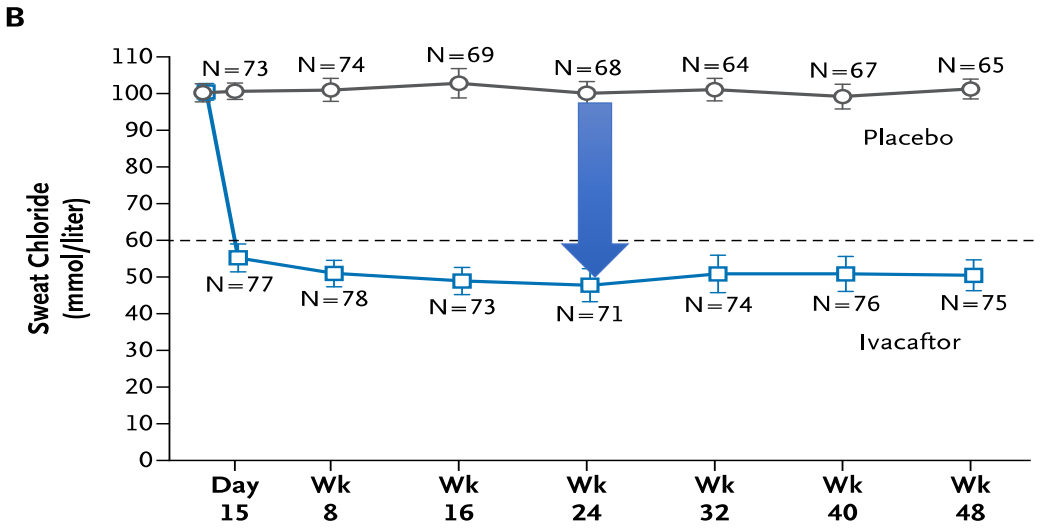
Adapted from: http://www.umd.be/CFTR/W_CFTR/gene.html.

Practical Updates in Primary Care

Ivacaftor and Health Outcomes



	Ivacaftor	Placebo
Absolute change from baseline through Week 24 in the ppFEV$_1$ (percentage points)	10.4	-0.2
Treatment difference (percentage points)	10.6 *P*<0.001	

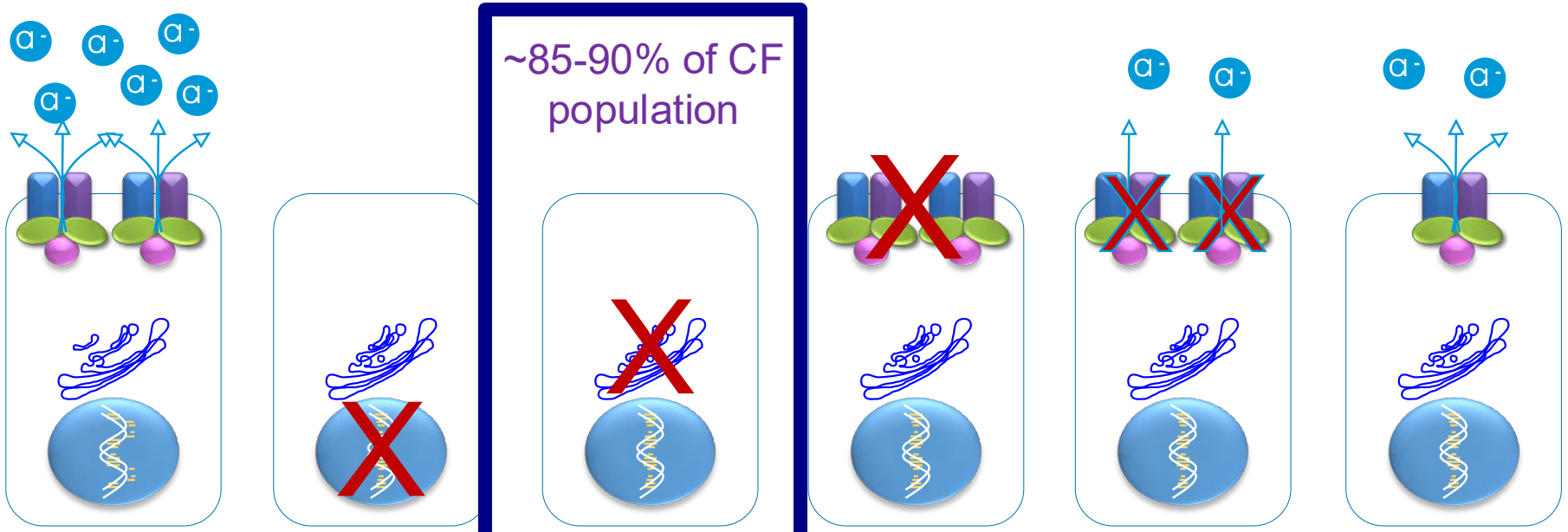


CFQ-R = Cystic Fibrosis Questionnaire-Respiratory; FEV = forced expiratory volume.

Ramsey BW, et al. *N Engl J Med*. 2011;365(18):1663-1672.

Practical Updates in Primary Care

5 (or 6) Classes of CFTR Mutations



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Adapted from: http://www.umd.be/CFTR/W_CFTR/gene.html.

Practical Updates in Primary Care

October 21, 2019

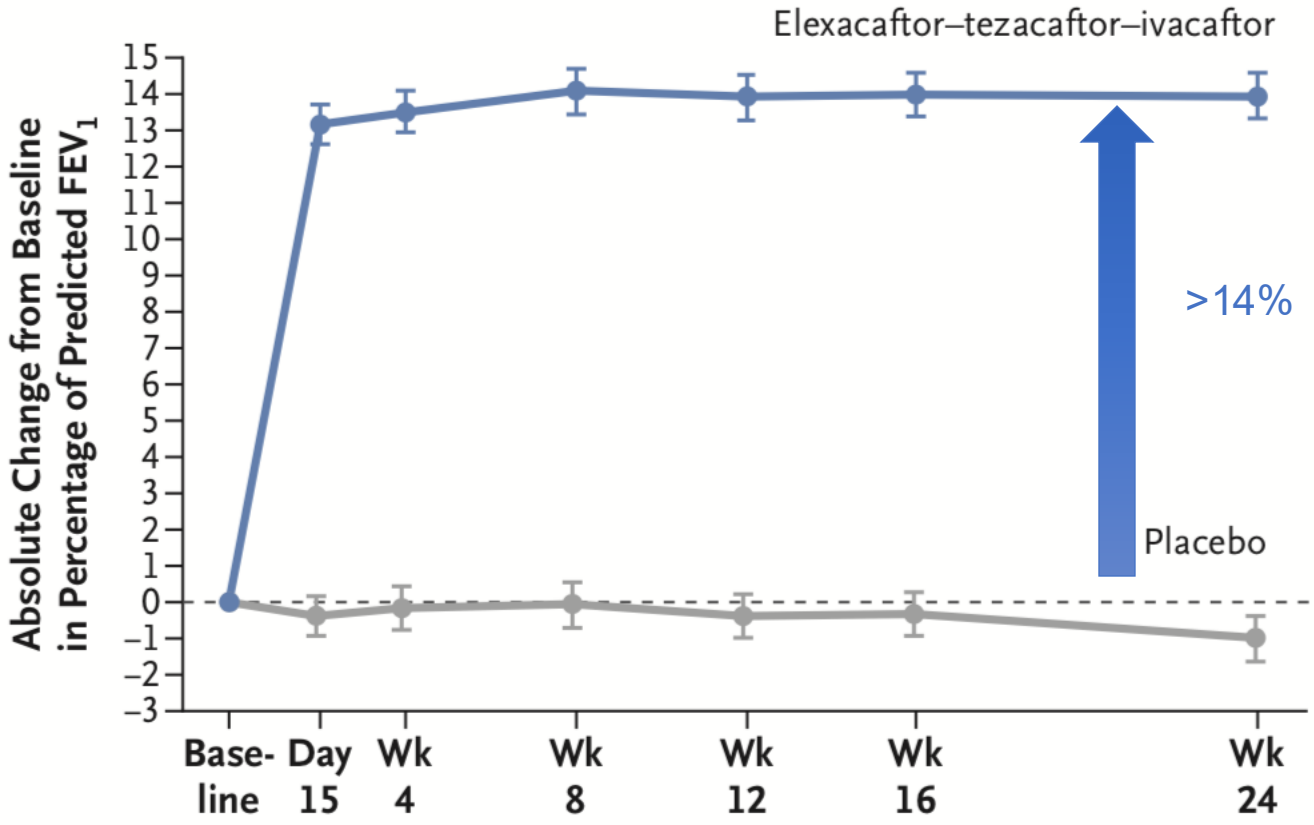
FDA approved elexacaftor/tezacaftor/ivacaftor for those with at least one copy F508del

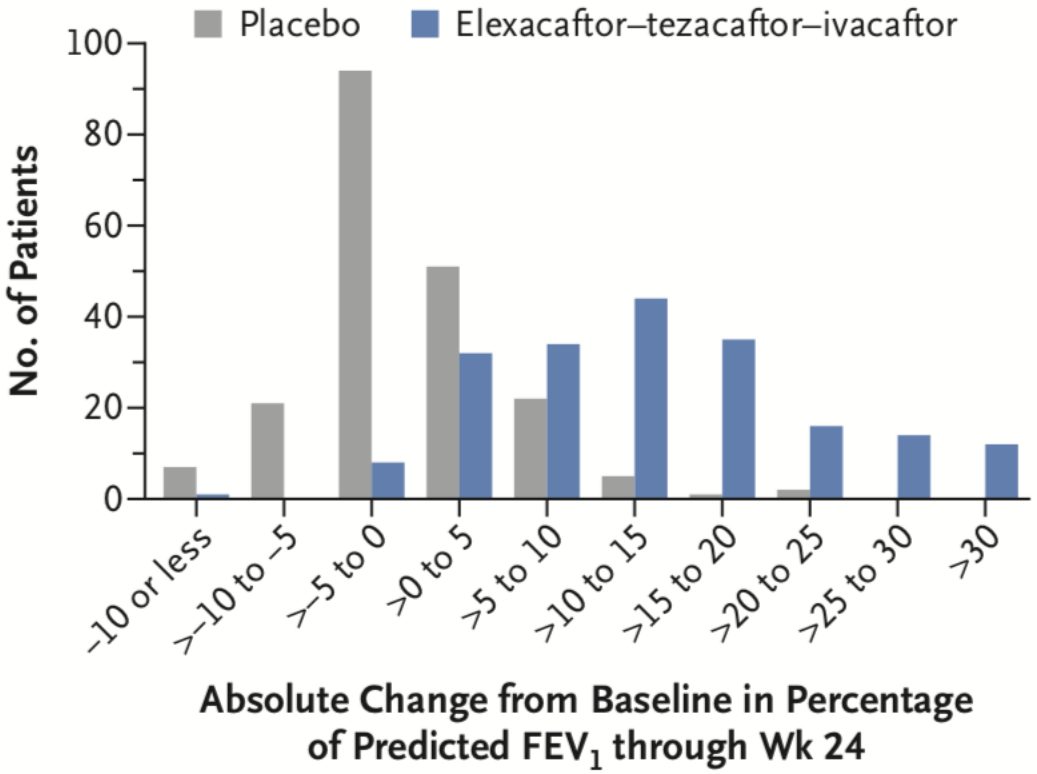
(preceded by lumacaftor/ivacaftor in 2015 and tezacaftor/ivacaftor and ivacaftor in 2018)

FDA. Accessed April 27, 2026. https://www.accessdata.fda.gov/scripts/cder/daf/.

Elaxacaftor/Tezacaftor/Ivacaftor

Lung Function Response (FEV_1)



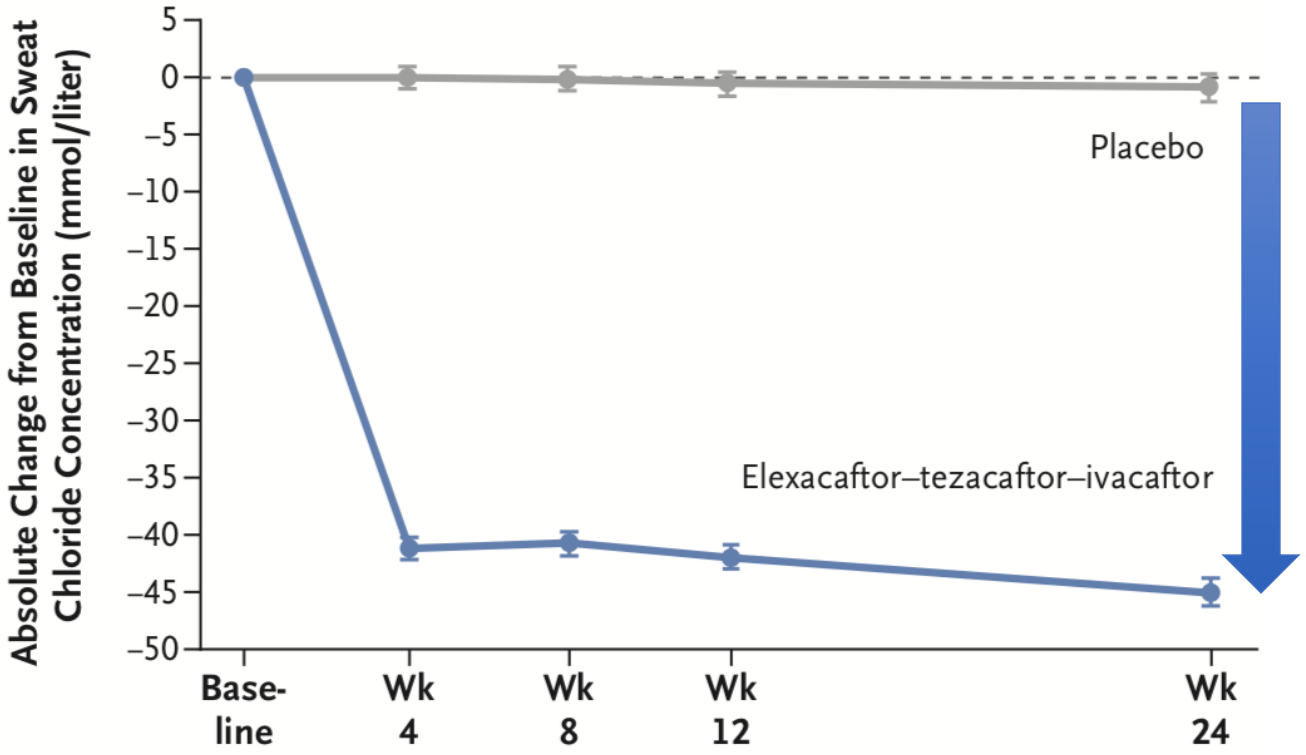


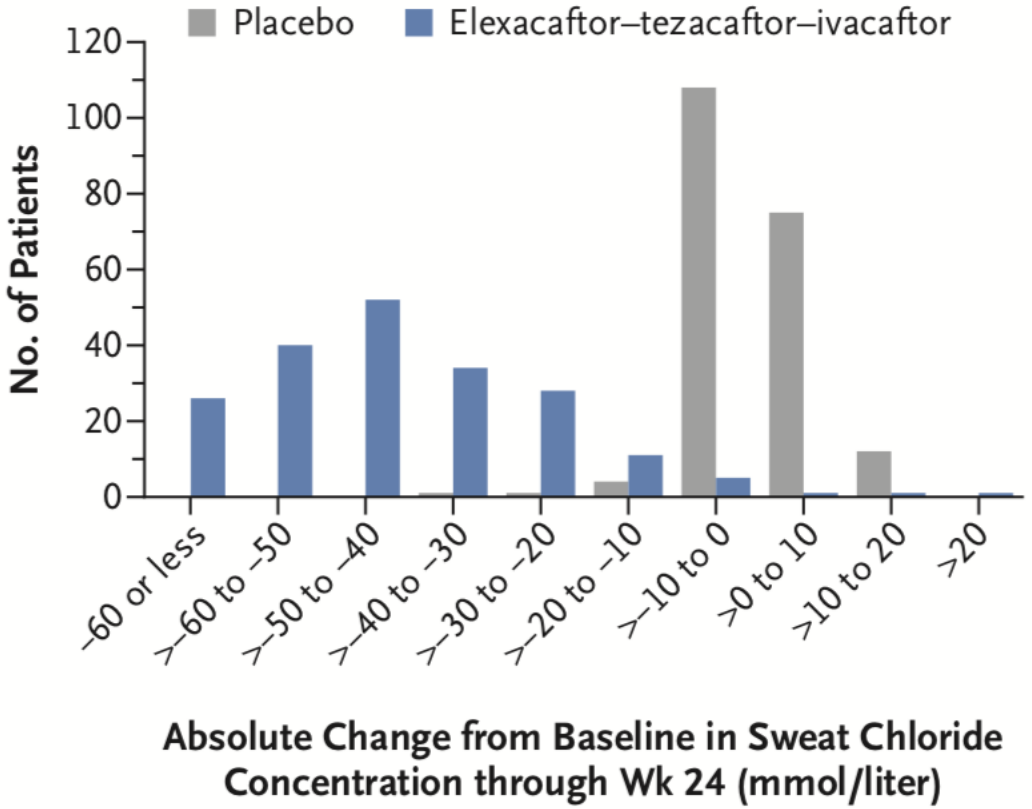
Middleton PG, et al. *N Engl J Med*. 2019;381(19):1809-1819.

Practical Updates in Primary Care

Elaxacaftor/Tezacaftor/Ivacaftor

Sweat Chloride Response (mmol/liter)



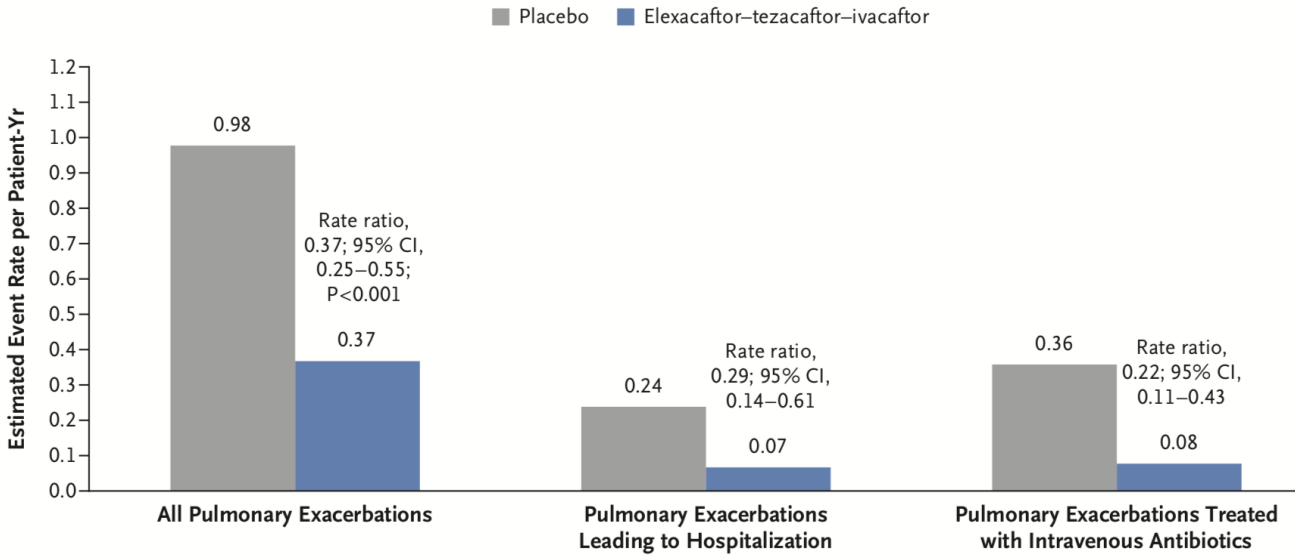


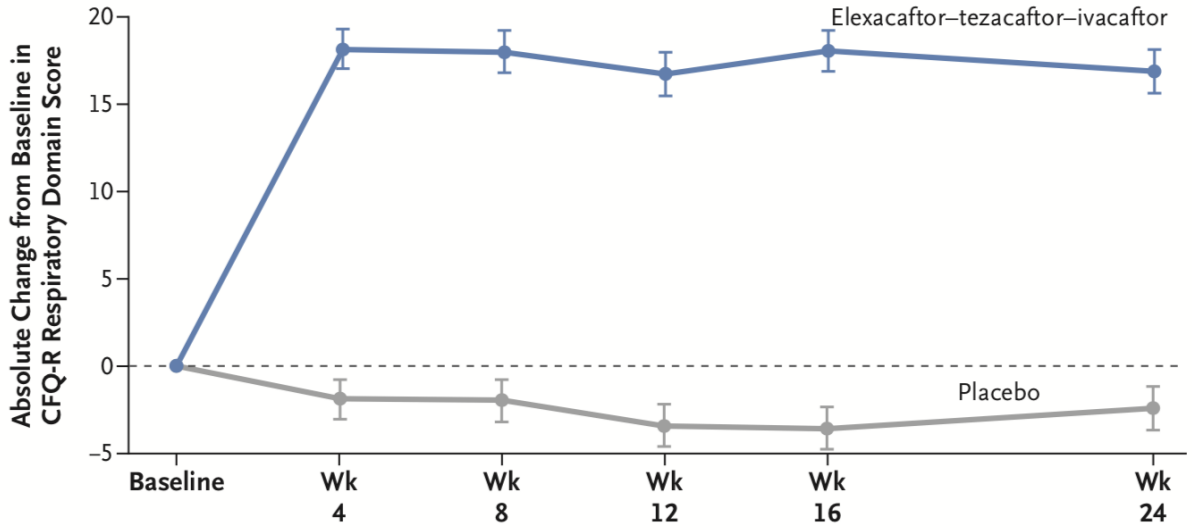
Middleton PG, et al. *N Engl J Med*. 2019;381(19):1809-1819.

Practical Updates in Primary Care

Elaxacaftor/Tezacaftor/Ivacaftor

Exacerbations and CFQ-R





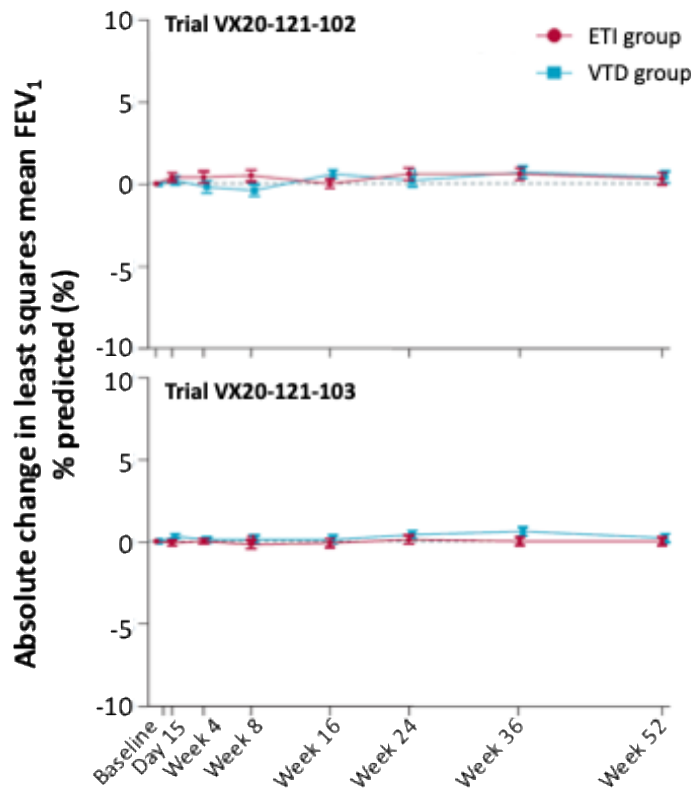
Middleton PG, et al. *N Engl J Med*. 2019;381(19):1809-1819.

Practical Updates in Primary Care

SKYLINE: Vanzacaftor/Tezacaftor/Deutivacaftor

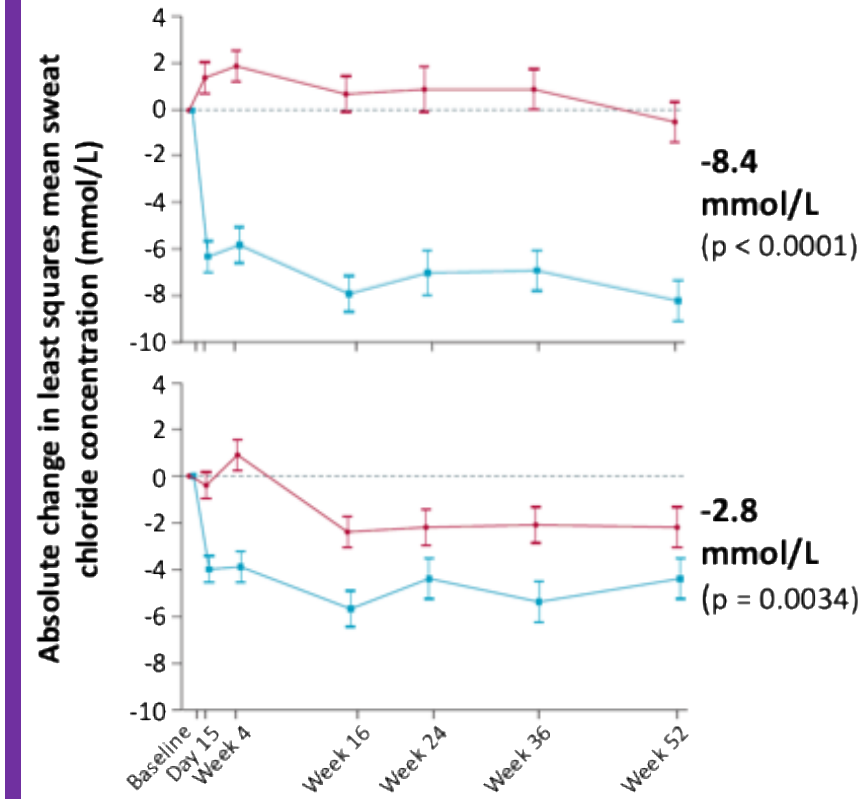
Two randomized, active-controlled, double-blind, phase 3 trials of twice daily (BID) ETI vs once daily (QD) VTD

ppFEV$_1$



QD VTD noninferior to BID ETI in terms of FEV$_1$

Sweat Chloride



Mean sweat chloride lower on VTD vs ETI

Adverse Events (AEs)

Comparable rates of AEs, 85% are mild to moderate

Most commons AEs

- Infective PEx
- Cough
- COVID-19
- Nasopharyngitis
- Headache
- URTI

VTD is comparably effective to ETI and dosed once daily vs twice daily

Insufficient data about how VTD impacts long-term complications, mental health

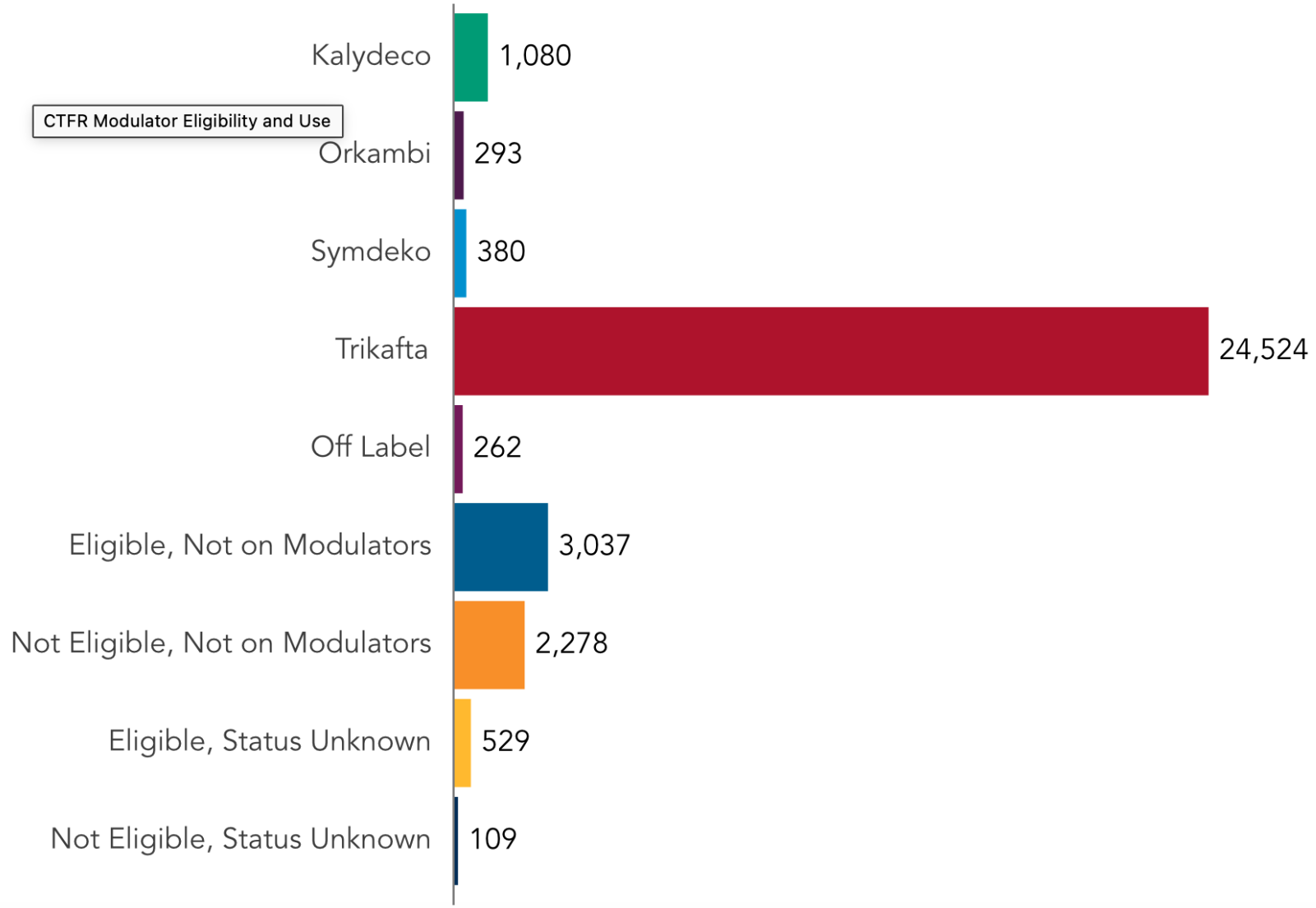
PEx = Pulmonary Exacerbation; VTD = Vanzacaftor-Tezacaftor-Deutivacaftor; ETI = Elexacaftor-Tezacaftor-Ivacaftor; FEV$_1$ = Forced Expiratory Volume per 1 second; SD = Standard Deviation; SE = Standard Error; CI = Confidence Interval

Slide courtesy of Tullis E.

Keating C, et al. *Lancet Respir Med*. 2025;13(3):256-271.

Practical Updates in Primary Care

Not All CF Patients Eligible for Modulators



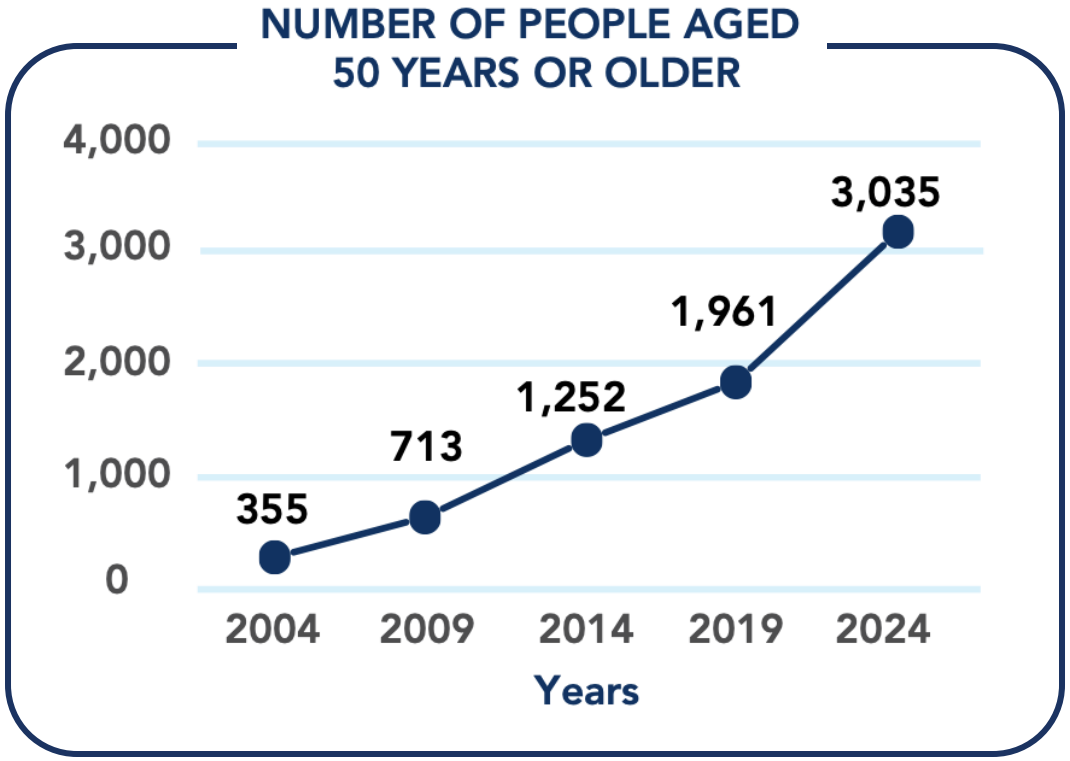
Cystic Fibrosis Foundation (CFF) [www.cff.org]. Last updated 2025. https://www.cff.org/medical-professionals/2024-patient-registry-highlights.

CF Foundation Patient Registry

More Adult-Focused Outcomes

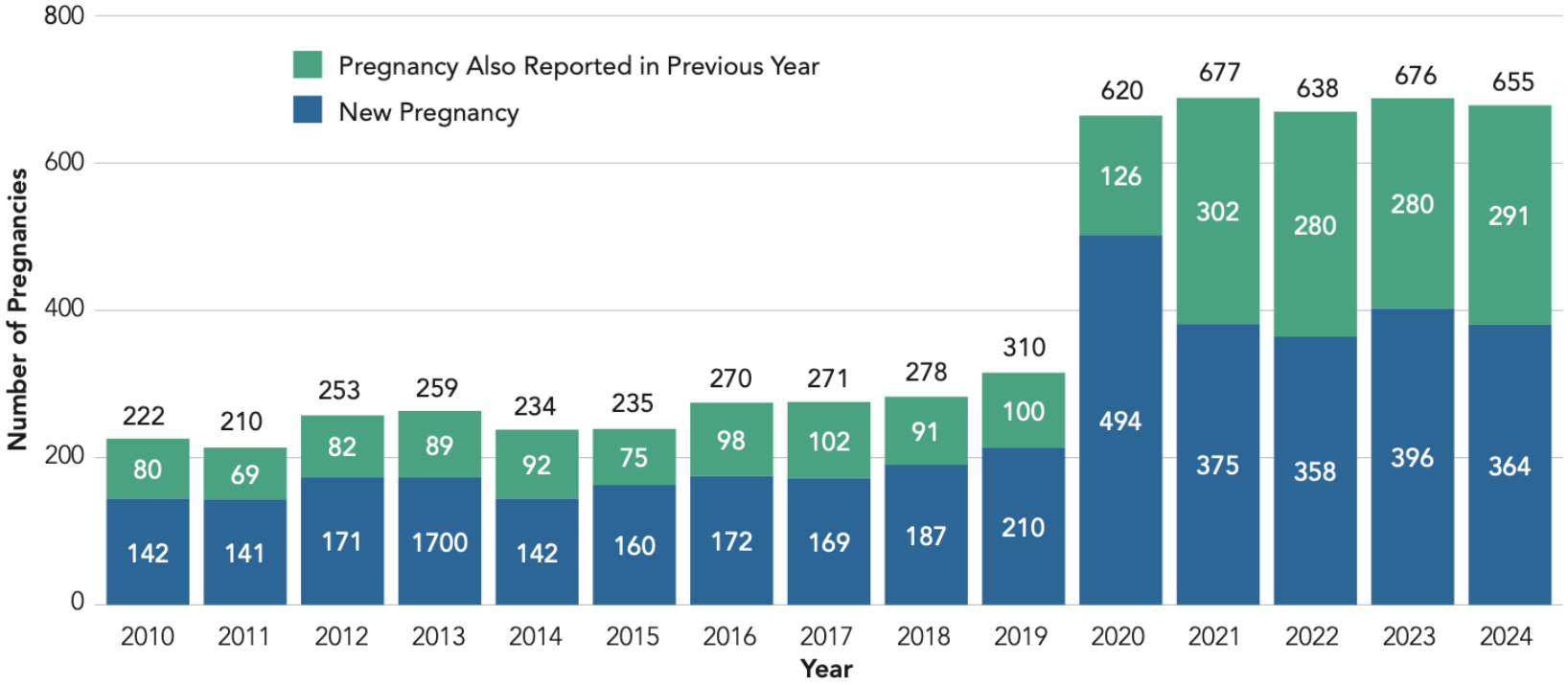
2023
- Blood pressure
- Lipid panel
- Insulin pump use
- Non-insulin therapies
- More causes of death: Cardiovascular, cancer-related

2019
- Colorectal cancer screening



CFF. Accessed April 27, 2026. https://www.cff.org/sites/default/files/2025-05/2024-Patient-Registry-Highlights-Report.pdf.

Number of CF Pregnancies by Year (Does Not Include Fathers with CF)



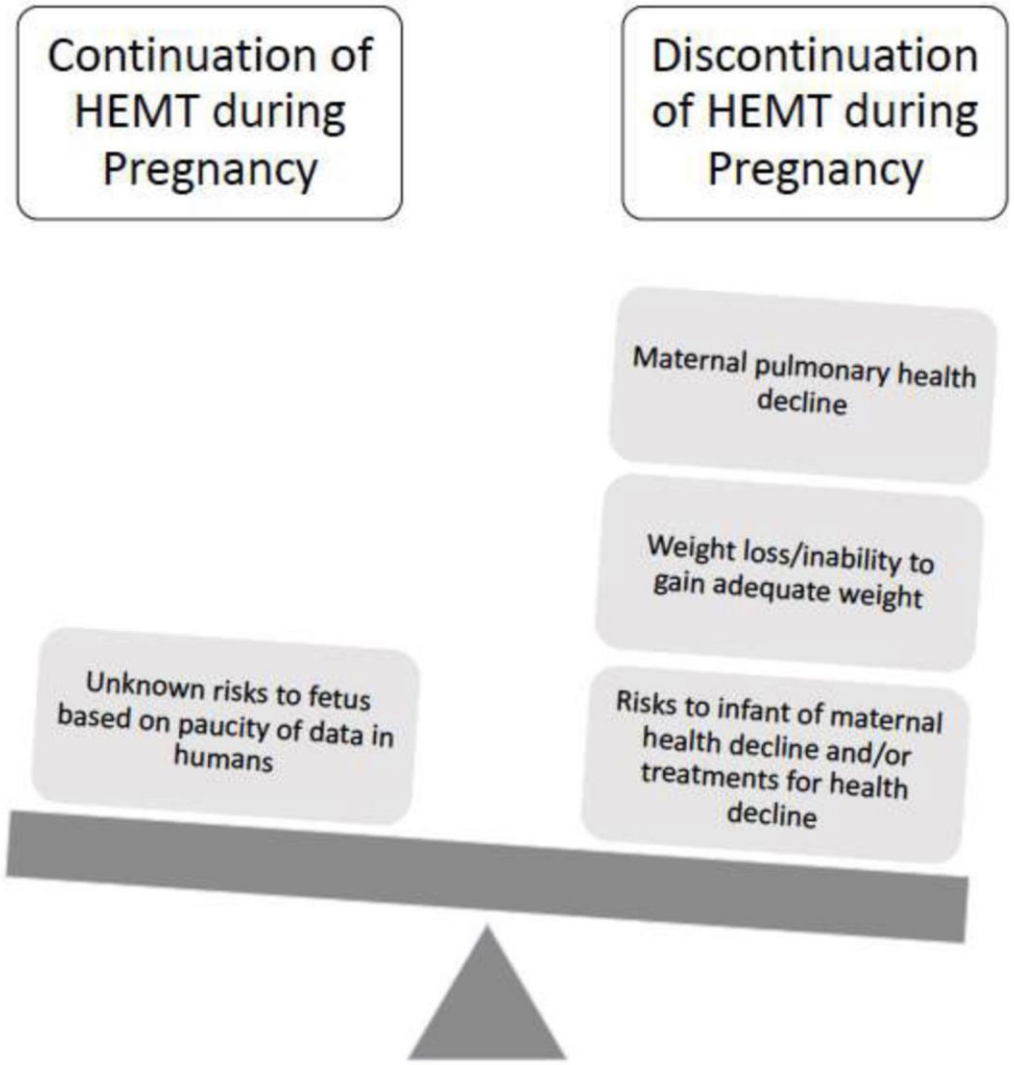
- 35% infertility prior to modulators; now…
  - Thin cervical mucus
  - Improved cervical and uterine pH
  - Improved BMI
  - Improvement in delay of puberty and anovulation

BMI = body-mass index.

CFF. Accessed April 27, 2026. https://www.cff.org/media/38406/download?inline. Taylor-Cousar JL. *J Clin Med*. 2020;9(9):2706. Kazmerski TM, et al. *Pediatr Pulmonol*. 2022;57(Suppl 1):S75-S88.

Practical Updates in Primary Care

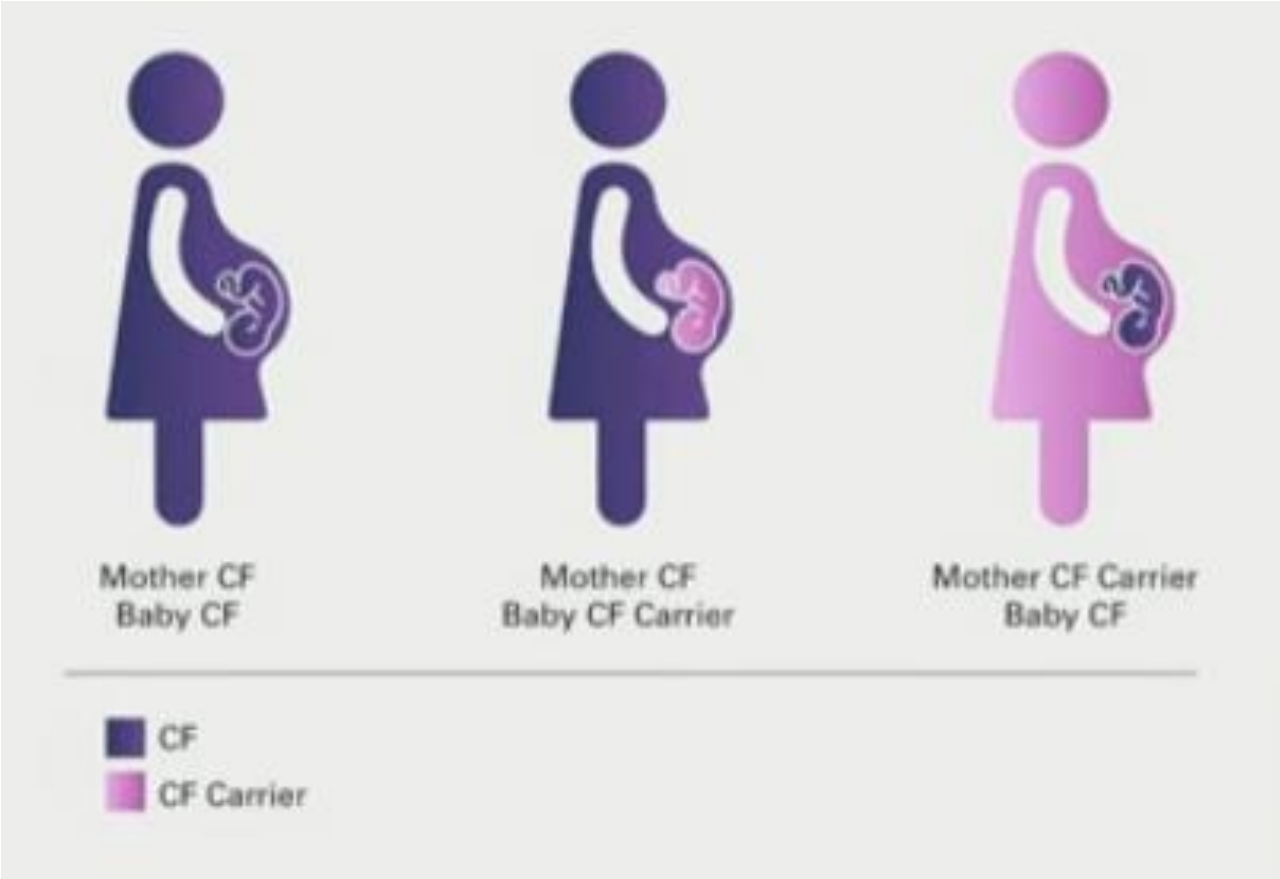
CFTR Modulators and CF Pregnancies



HEMT = highly effective modulator treatment.

Ramos KJ, et al. *J Cyst Fibros*. 2021;20(3):381-387.

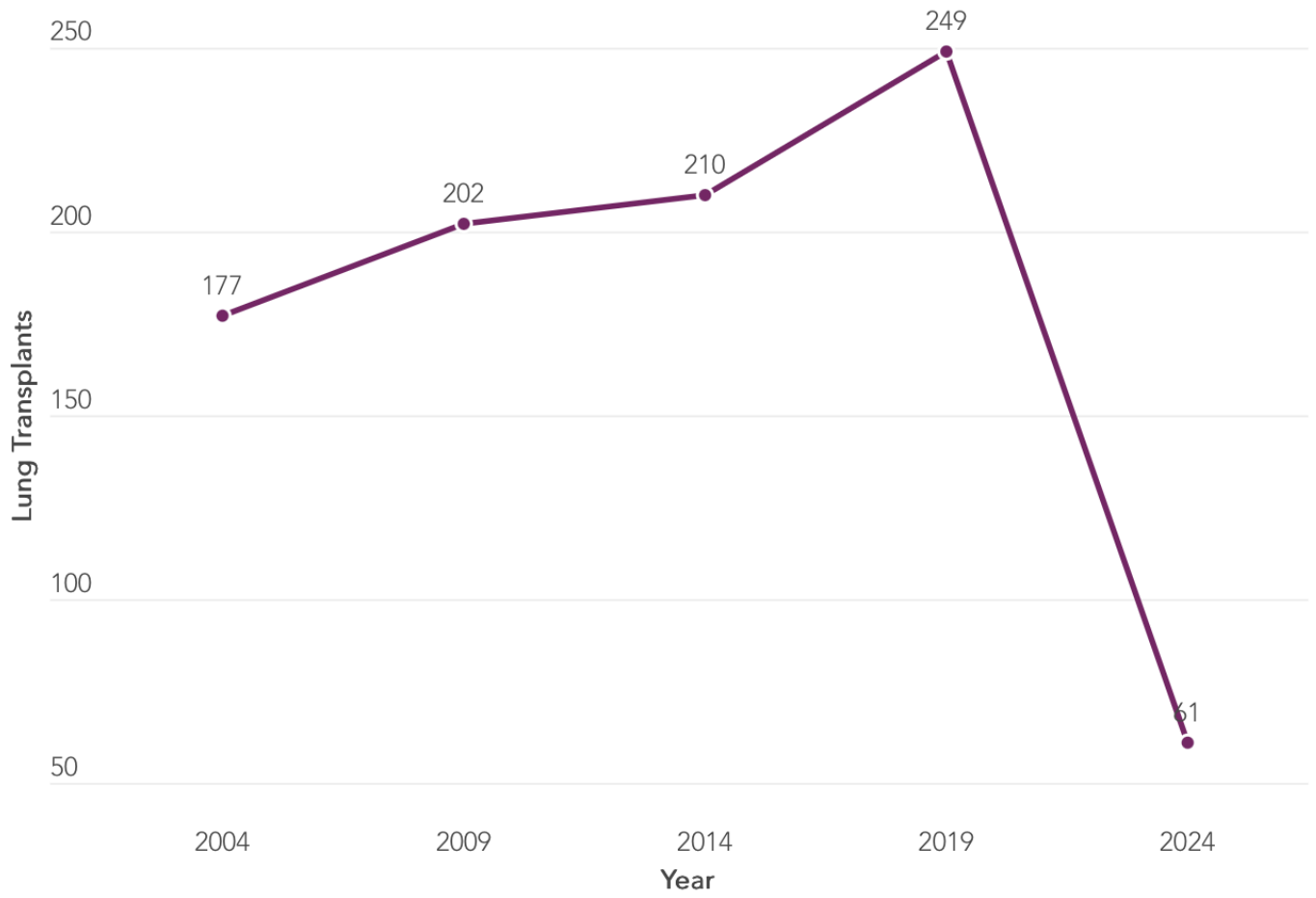
What's Even Earlier than Newborn Screening: Non-Invasive Prenatal Testing (NIPT)



Courtesy of Jain R, Taylor-Cousar J.

Practical Updates in Primary Care

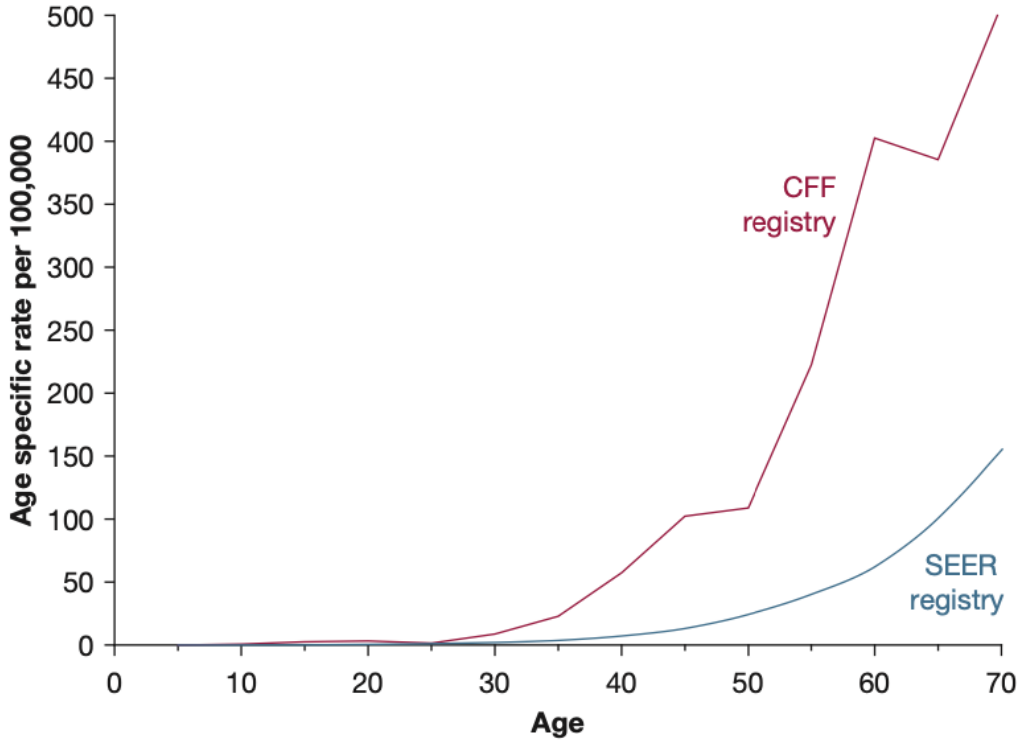
Lung Transplants by Year



CFF [www.cff.org]. Last updated 2025. https://www.cff.org/medical-professionals/2024-patient-registry-highlights.

Practical Updates in Primary Care

Increased Risk of GI Tract Cancer in CF

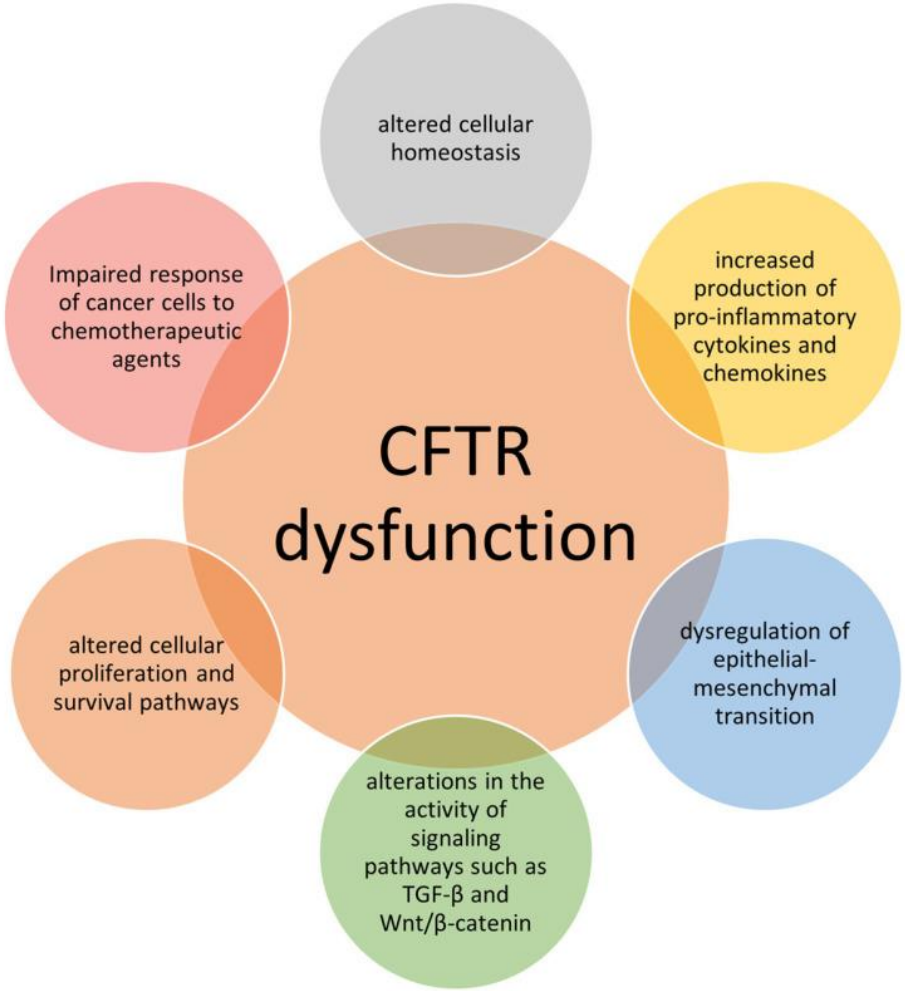


- Higher incidence of GERD, inflammatory bowel disease, diabetes
- Impaired mucosal barrier function, bowel microbiome alteration, inflammation (bowel obstruction), abnormal immune response
- Nutrition
  - High-fat and low-fiber diet
  - Vitamin D deficiency
- CFTR-related
  - Associated with class I-III mutations
  - Risk associated with higher sweat Cl-
  - CF carriers at higher risk for CRC, stomach, and other GI-related cancers
- CFTR gene acts as a tumor suppressor gene
  - Does GI cancer in CF behave differently than non-CF?

SEER = National Cancer Institute (NCI) Surveillance, Epidemiology, and End Results Program; GERD = gastroesophageal reflux disease; CRC = colorectal cancer; GI = gastrointestinal.

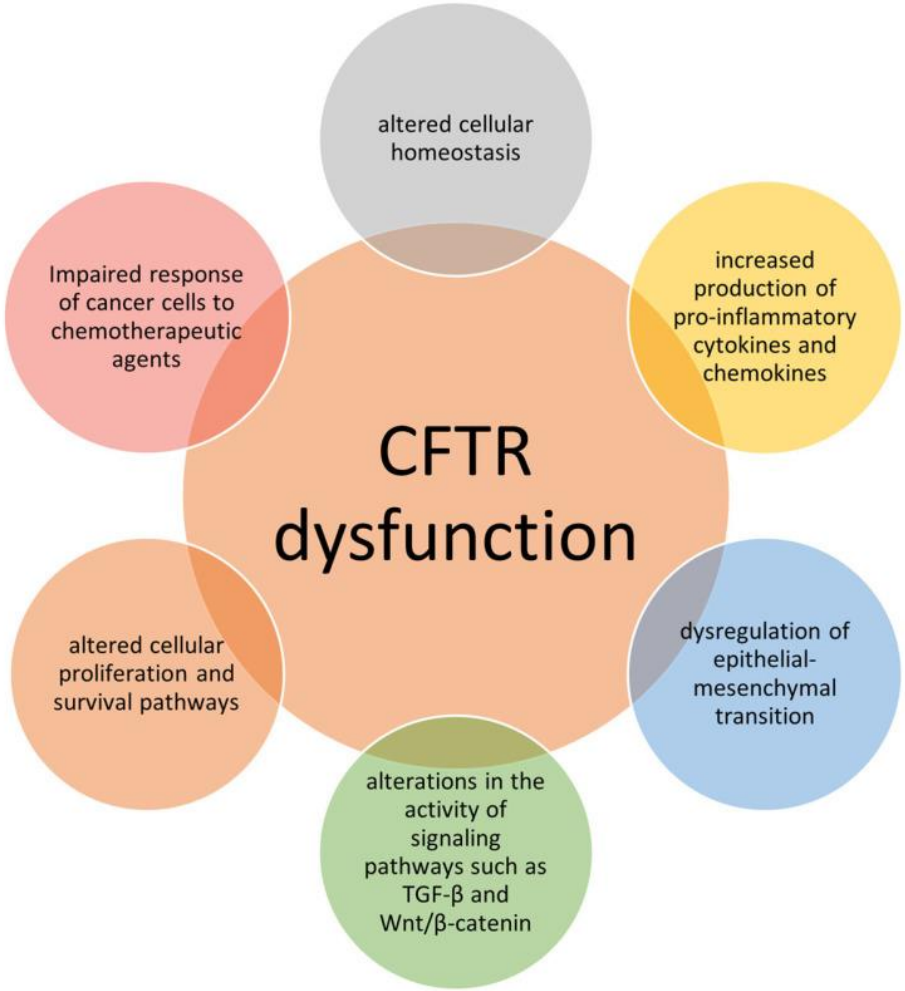
Maisonneuve P, Lowenfels AB. *Chest*. 2022;161(2):356-364. Than BLN, et al. *Oncogene*. 2016;35(32):4179-4187. Liu C, et al. *Cancer Manag Res*. 2020;12:4261-4270.

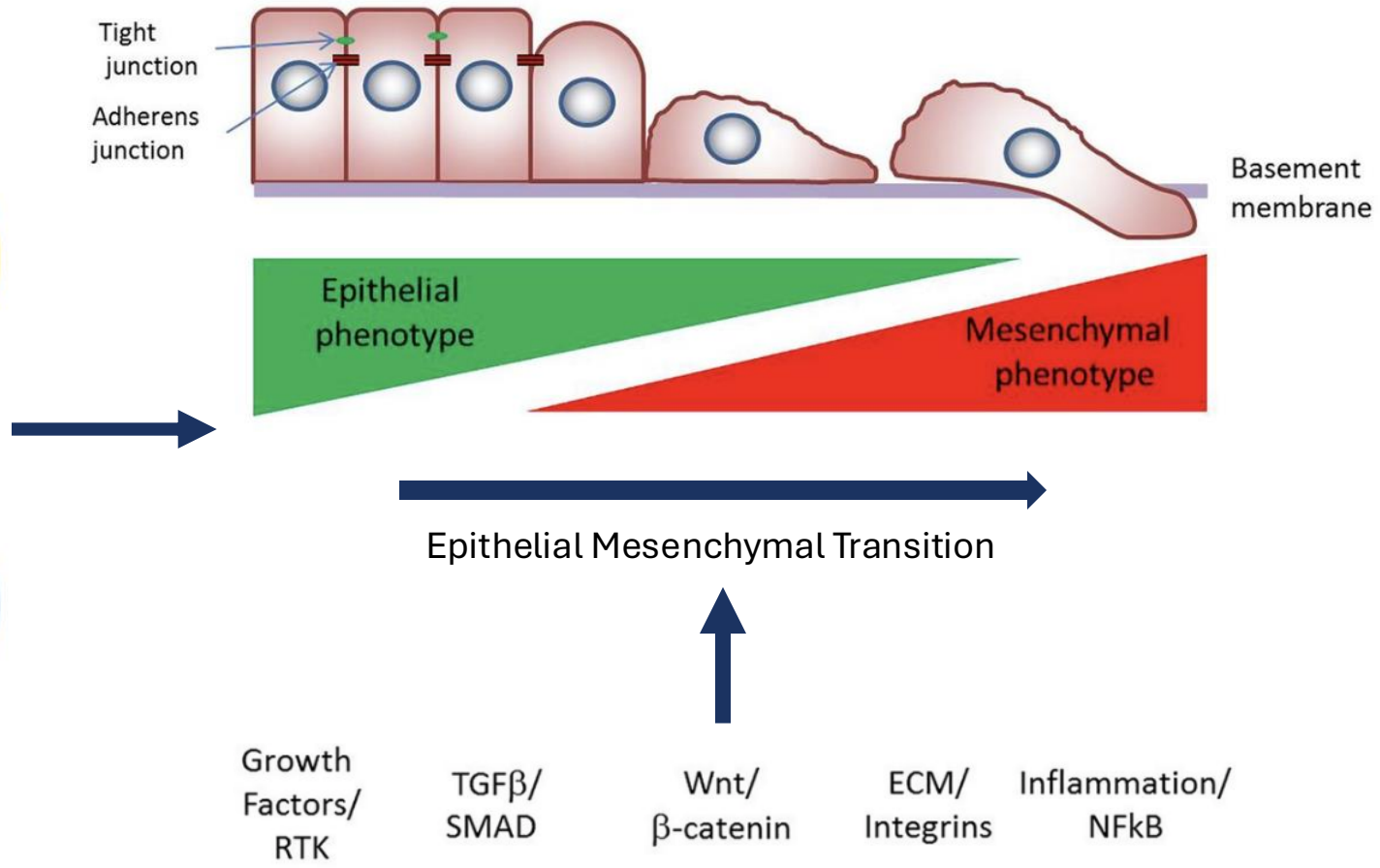
CFTR Is a Tumor Suppressor Gene!



Parisi GF, et al. *Cancers (Basel)*. 2023;15(17):4244. Bartis D, et al. *Thorax*. 2014;69(8):760-765.

CFTR Is a Tumor Suppressor Gene!



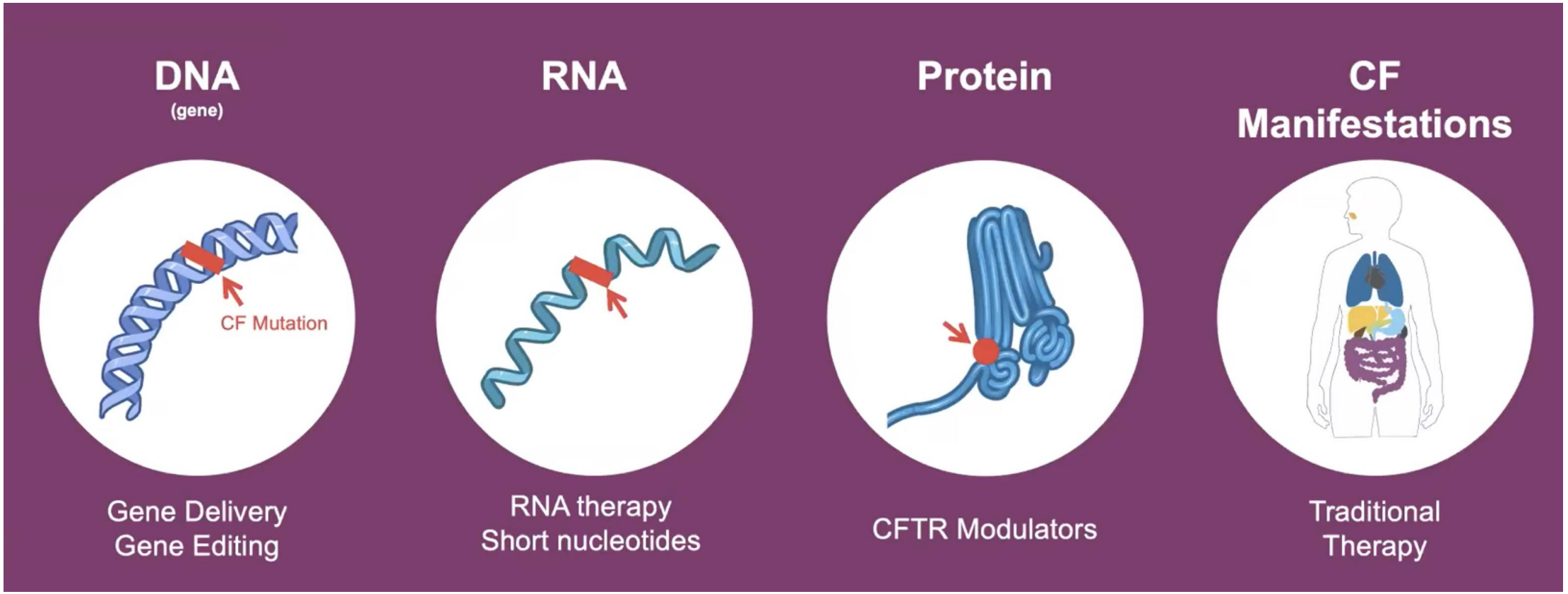


ECM = extracellular matrix.

Parisi GF, et al. *Cancers (Basel)*. 2023;15(17):4244. Bartis D, et al. *Thorax*. 2014;69(8):760-765.

Practical Updates in Primary Care

Therapeutic Approach to CF



Courtesy of CFF.

Practical Updates in Primary Care

Multidisciplinary Approaches

Established Model of Care for pwCF: Multidisciplinary Team

Yankaskas JR, et al. *Chest*. 2004;125(1 Suppl):1S-39S. Smyth AR, et al. *J Cyst Fibros*. 2014;13(Suppl 1):S23-S42. Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.

Practical Updates in Primary Care

Management of CF

- Maintain airway clearance
- Chronic suppressive therapy and early treatment of exacerbations
- Adequate nutrition and exercise
- Psychosocial support
- Diagnose and treat complications
- Assess transplant referral/advanced care planning

Yankaskas JR, et al. *Chest*. 2004;125(1 Suppl):1S-39S. Smyth AR, et al. *J Cyst Fibros*. 2014;13(Suppl 1):S23-S42. Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.

Practical Updates in Primary Care

Management of CF

CF Foundation Guidelines

- Quarterly visits to CF Care Center
  - Monitor pulmonary status (FEV_1)
  - Review medical regimen
  - Identify specific medical or psychosocial issues
- Annually
  - Comprehensive evaluation by all team members
  - Depression and anxiety screening
  - Labs: vitamins A, E, D, LFTs, OGTT, A1C, IgE
  - Vaccinations

LFTs = liver function tests; OGTT = oral glucose tolerance test.

Yankaskas JR, et al. *Chest*. 2004;125(1 Suppl):1S-39S. Smyth AR, et al. *J Cyst Fibros*. 2014;13(Suppl 1):S23-S42. Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.

Management of CF

CF Foundation Guidelines

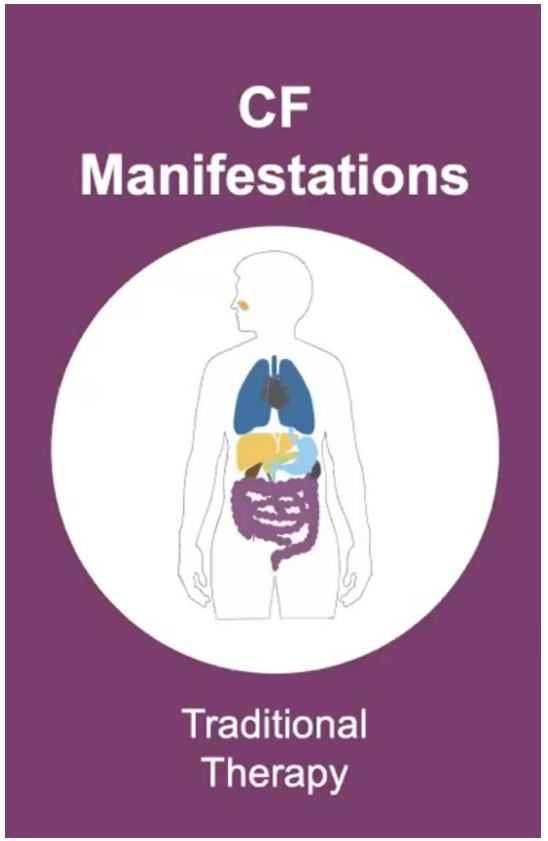
- Routinely
  - Respiratory specimen cultures with annual surveillance for mycobacterial growth (AFB culture)
  - BMI assessment
  - Nutritional assessment
  - Exercise assessment
  - Discussion of disease trajectory

AFB = acid-fast bacilli.

Yankaskas JR, et al. *Chest*. 2004;125(1 Suppl):1S-39S. Smyth AR, et al. *J Cyst Fibros*. 2014;13(Suppl 1):S23-S42. Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.

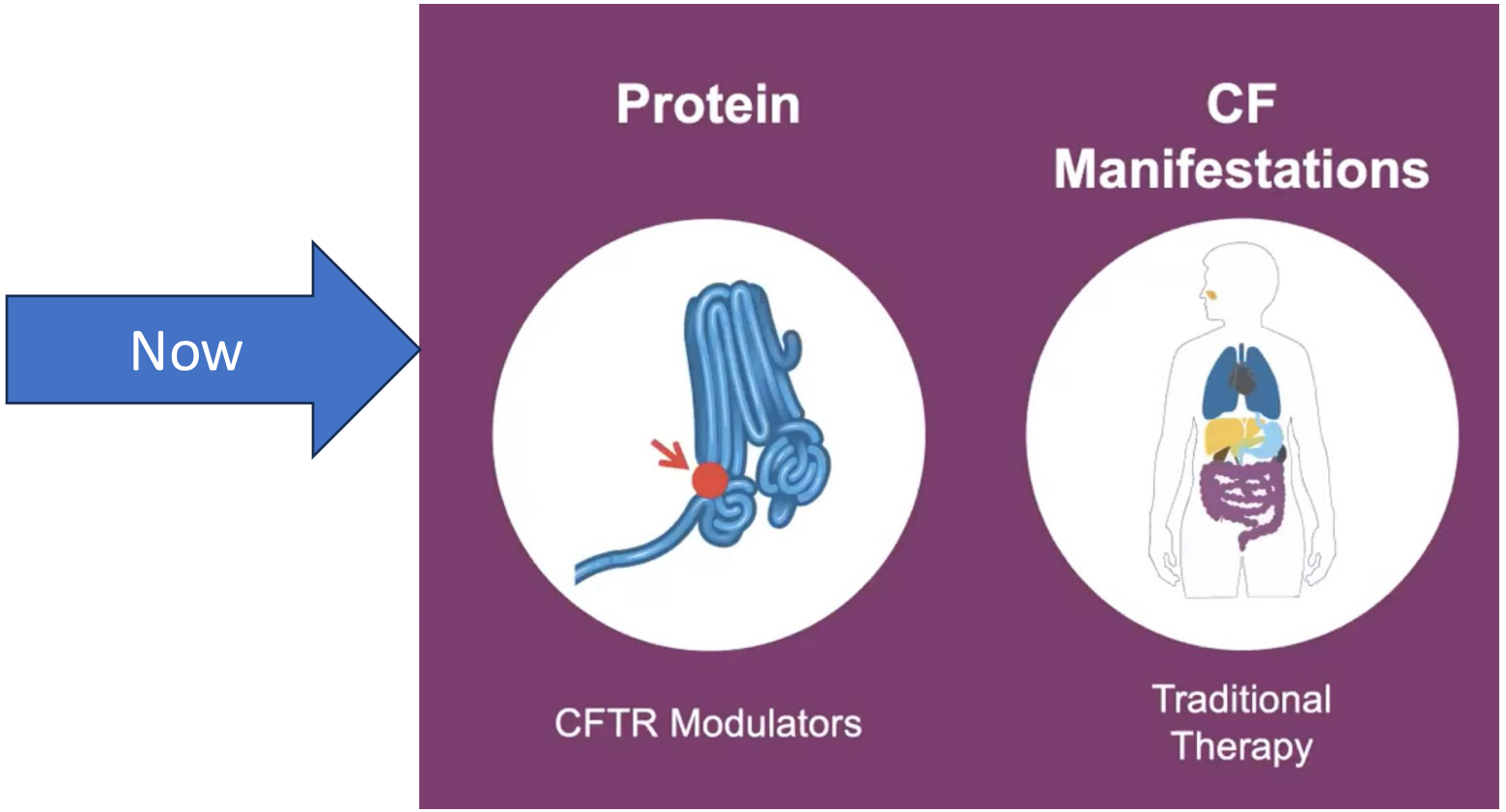
Management of CF

Then



Courtesy of CFF.

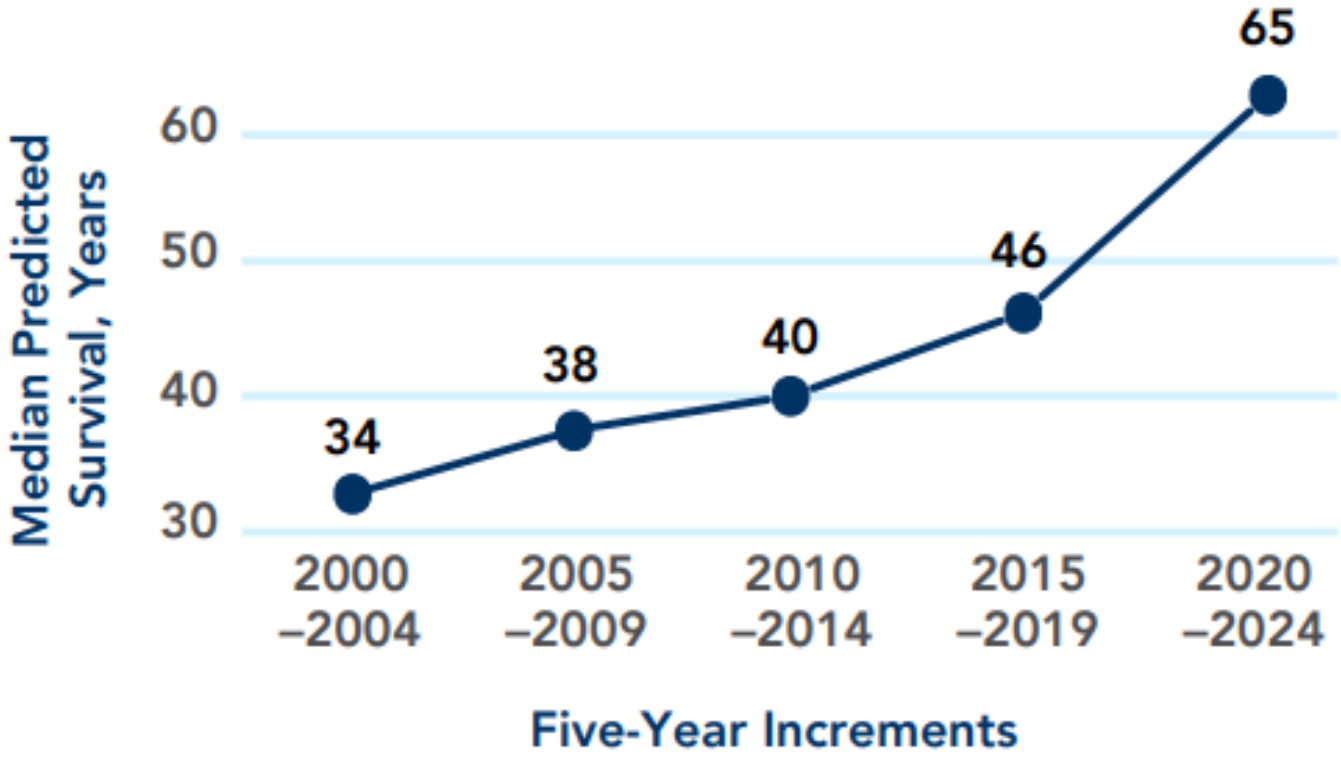
New Management of CF



Courtesy of CFF.

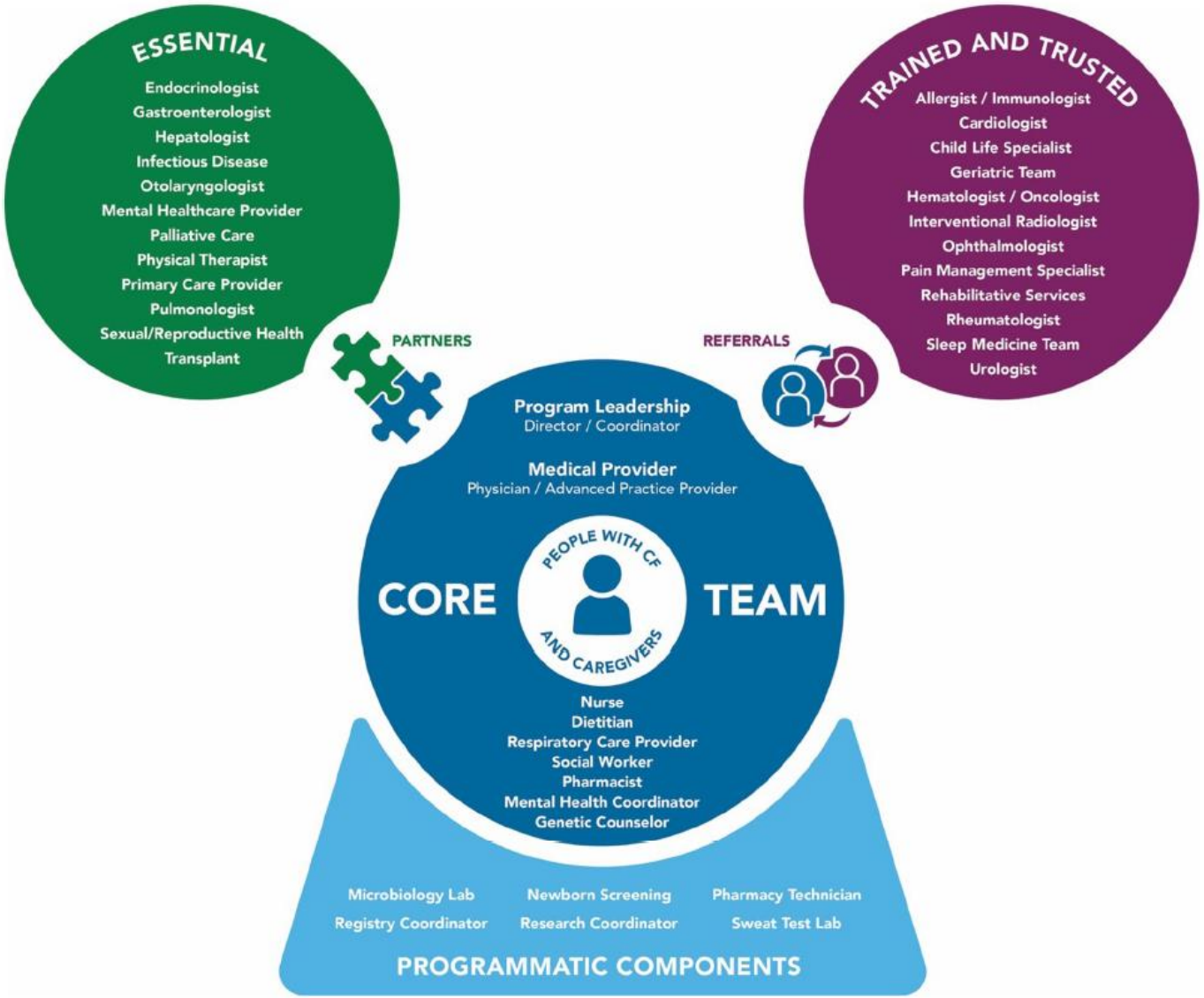
CF Foundation Patient Registry

People with CF are living longer



CFF. Accessed April 28, 2026. https://www.cff.org/sites/default/files/2025-05/2024-Patient-Registry-Highlights-Report.pdf.

Updated CF Care Team



Brown RF, et al. *J Cyst Fibros*. 2024;23(6):1045-1054.

Practical Updates in Primary Care

Updated CF Care Team

Brown RF, et al. *J Cyst Fibros*. 2024;23(6):1045-1054.

CF Foundation Patient Registry

More Adult-Focused Outcomes

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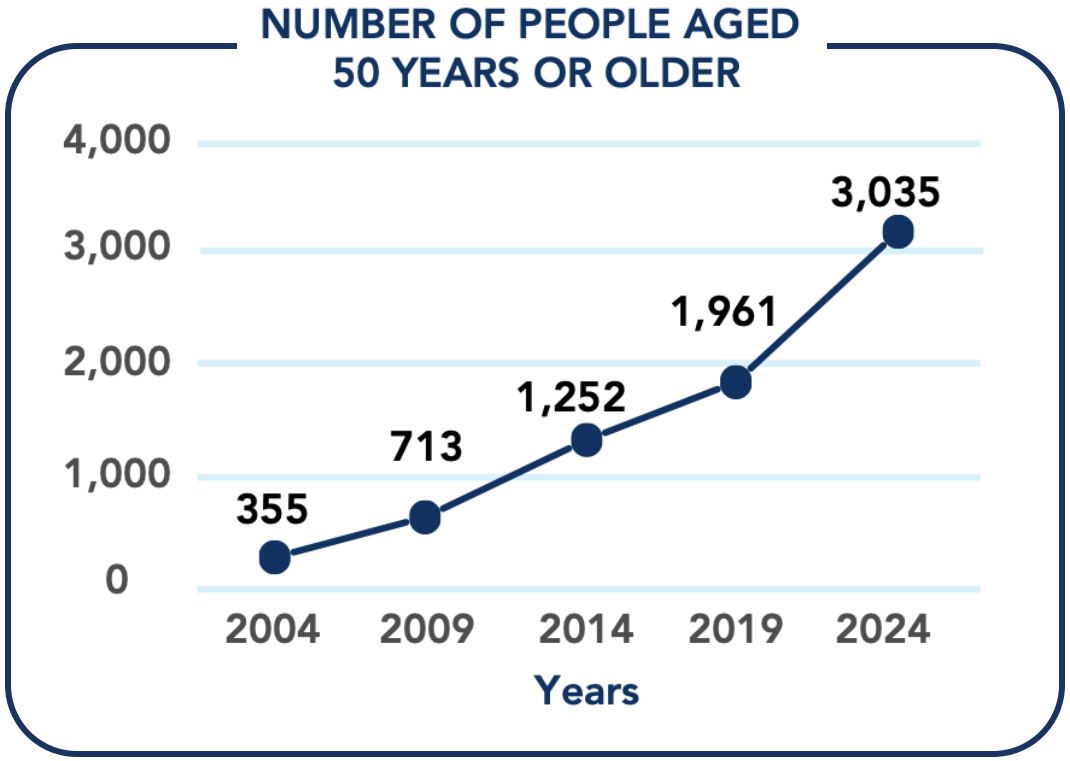
- Blood pressure
- Lipid panel
- Insulin pump use
- Non-insulin therapies
- More causes of death: Cardiovascular, cancer-related

2019
- Colorectal cancer screening



CFF. Accessed April 27, 2026. https://www.cff.org/sites/default/files/2025-05/2024-Patient-Registry-Highlights-Report.pdf.

Management of CF

Lung imaging

- Chest CT now recommended over routine CXR

Liver evaluation

- Monitoring of hepatic enzymes
- Imaging—ultrasound or MRI
- Evaluate for steatosis and fibrosis

CT = computed tomography; CXR = chest X-ray; MRI = magnetic resonance imaging.

Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.

Practical Updates in Primary Care

Management of CF

Annual screening for diabetes

- Oral glucose tolerance test

CF-related diabetes

- Screening for microvascular complications
  - Dilated retinal exams
  - Diabetic foot exam
  - Urine microalbumin to creatinine ratio

Auth R, et al. *J Prim Care Community Health*. 2023;14:21501319231173811.

Practical Updates in Primary Care

Management of CF

Increased risk for colon cancer

- 5- to 10-fold
- Screening should begin at age 40 for most
- Transplant recipients start at age 30
- Repeated at 5-year intervals or sooner

Bone health

- DEXA at age 18
- Repeat every 1-5 years based on results

DEXA = dual-energy X-ray absorptiometry.

Auth R, et al. *J Prim Care Community Health*. 2023;14:21501319231173811.

Management of CF

Family Planning

Menopause

Cancer

Cardiovascular

Renal/Urologic

Auth R, et al. *J Prim Care Community Health*. 2023;14:21501319231173811.

Practical Updates in Primary Care

Adapting the CF Care Model

- With the advent of highly effective CFTR modulators, pwCF are living longer and having fewer hospitalizations, fewer sick visits
- Do they still need to be seen as frequently at CF centers?

- A large national survey showed both CF care providers and pwCF endorsed
  - Decreasing visits to CF Care Center for those with stable health who are taking a CFTR modulator
  - Increasing integration of telehealth
  - Adopting remote care activities

Tran QT, et al. *J Cyst Fibros*. 2024;23(6):1066-1071. Brown RF, et al. *J Cyst Fibros*. 2024;23(6):1045-1054.

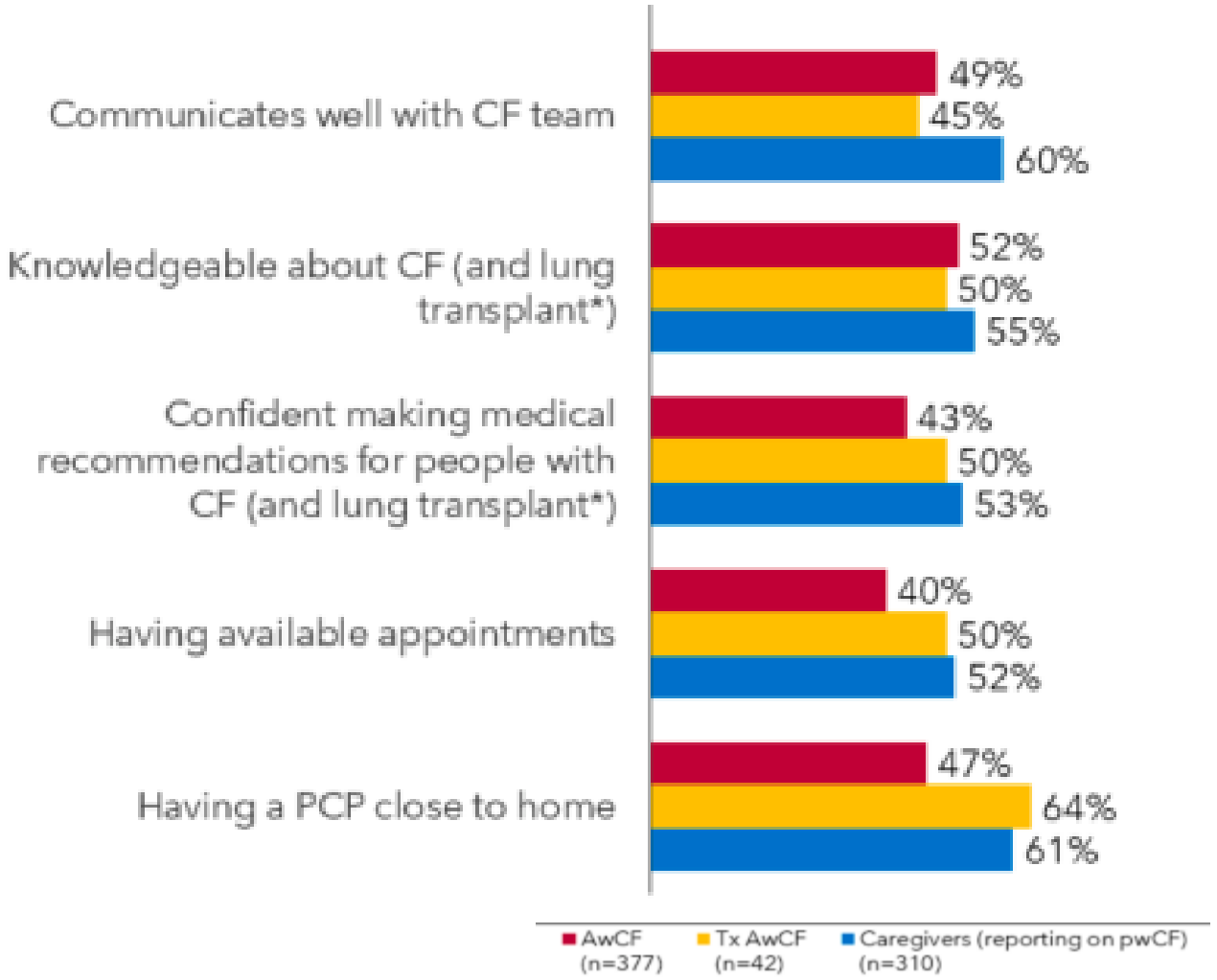
Adapting the CF Care Model

- Most pwCF or caregivers reported currently seeing PCP distinct from CF care team
  - Only 12% felt PCP not necessary

- In contrast, 72% of CF care team members felt that pwCF thought they didn't need PCP as CF team met all care needs
  - Most CF providers underestimated percentage of patients with PCP

Brown RF, et al. *J Cyst Fibros*. 2024;23(6):1045-1054.

Most Important Factors for pwCF When Considering PCP



Tran QT, et al. *J Cyst Fibros*. 2024;23(6):1066-1071. **Goetz DM, et al. *J Cyst Fibros*. 2024;23(6):1055-1065.**

What Do pwCF Want in Their Providers?

Fig. 1. Primary dimensions of Trust derived from analyses of CF Foundation Experience of Care (XoC) Survey.

Van Gorp S, et al. *J Cyst Fibros*. 2024;23(6):1035-1037.

Key Learning Points

- Pre-symptomatic newborn screening and CFTR modulator therapy have brought about a new era for patients with CF
- Increasing lifespans will require a shift in focus toward preventive care and screening (eg, CF-related diabetes, colorectal cancer)
- PCPs should be familiar with the unique screening guidelines and treatment regimens employed by CF centers
- PCPs should work collaboratively with CF centers, monitoring patients with CF closely for nutrition/growth and other CF-related complications