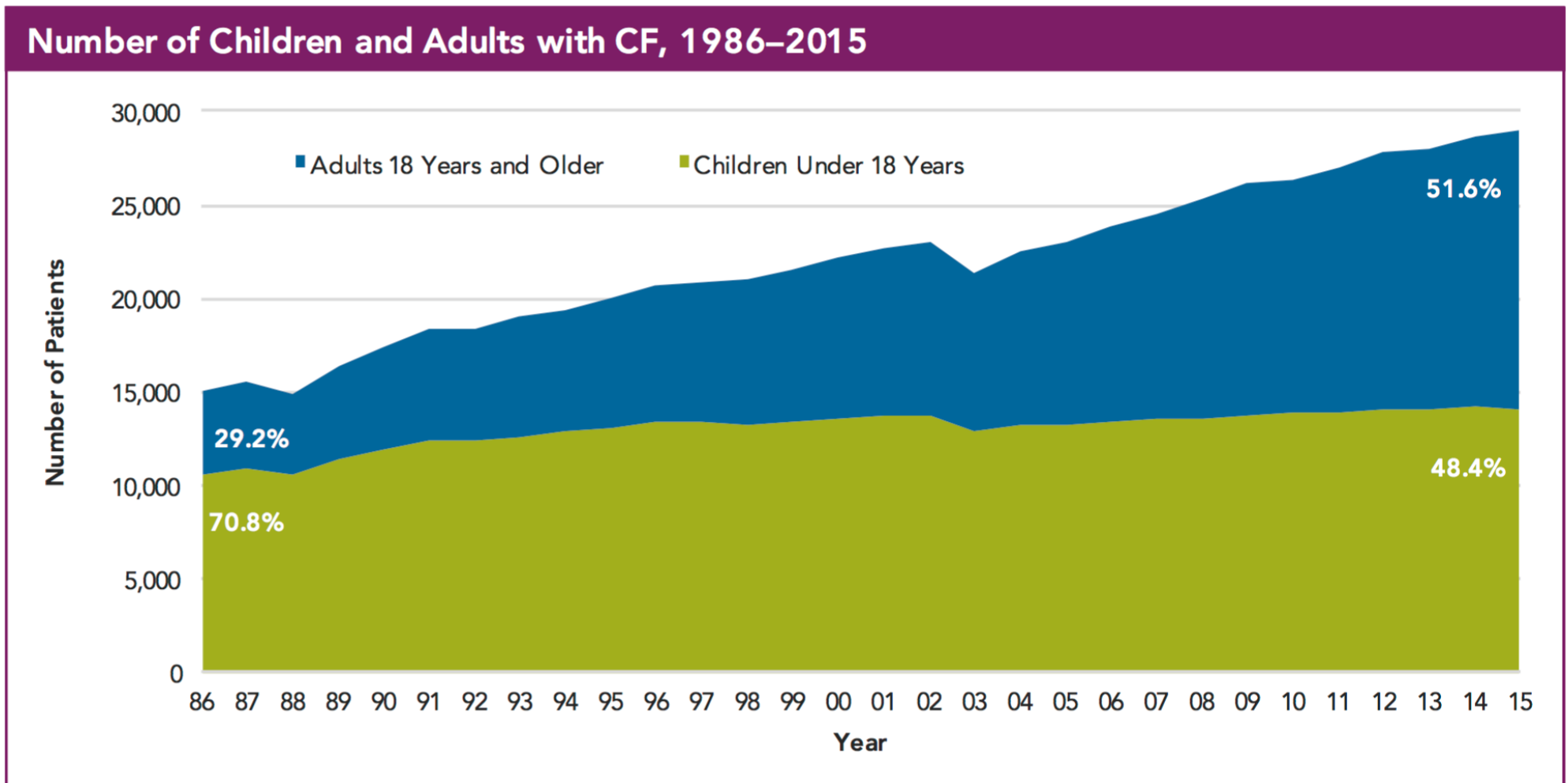


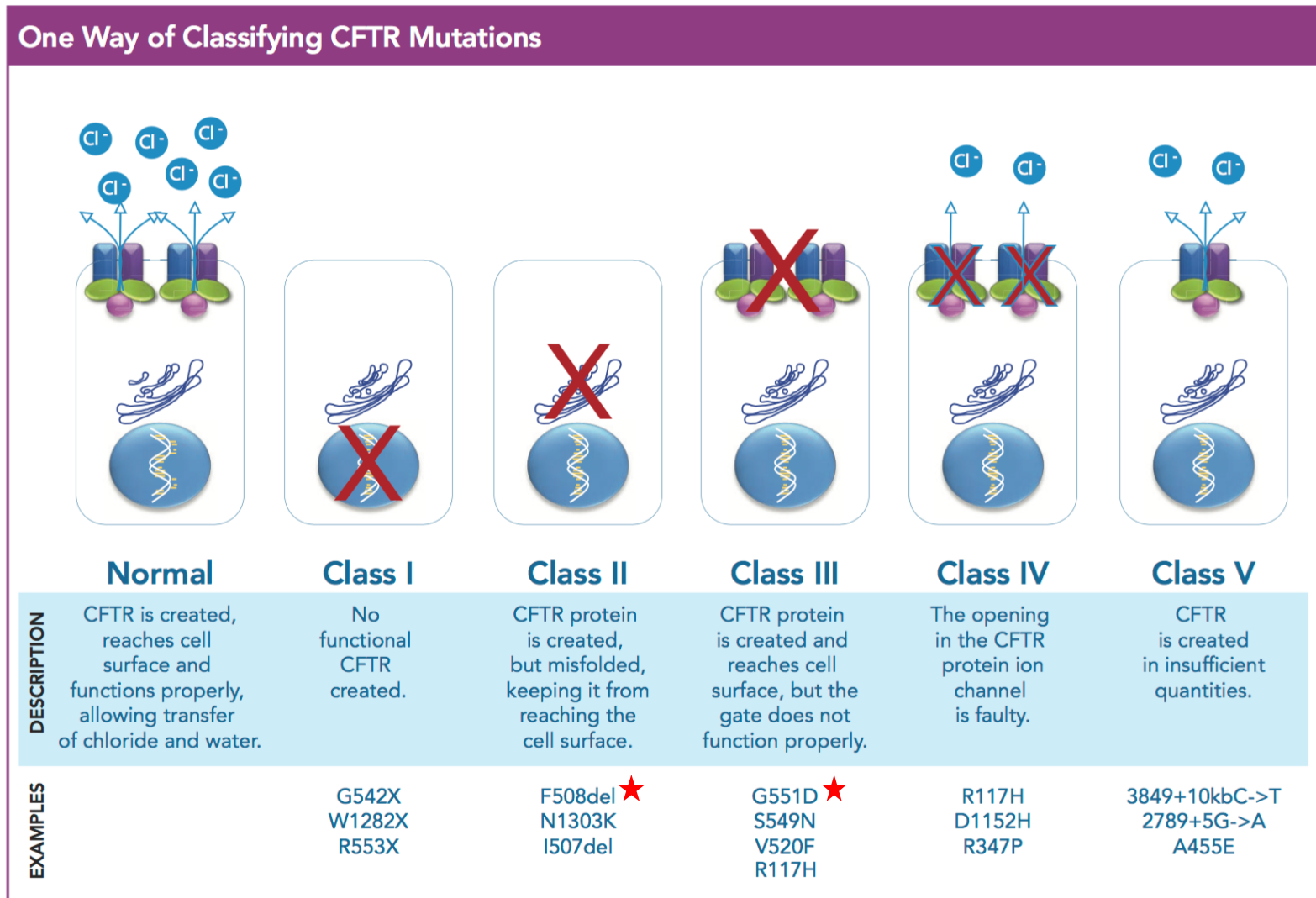
Why it's increasingly important for adult pulmonologists to learn about CF



Median predicted survival 41-42 years

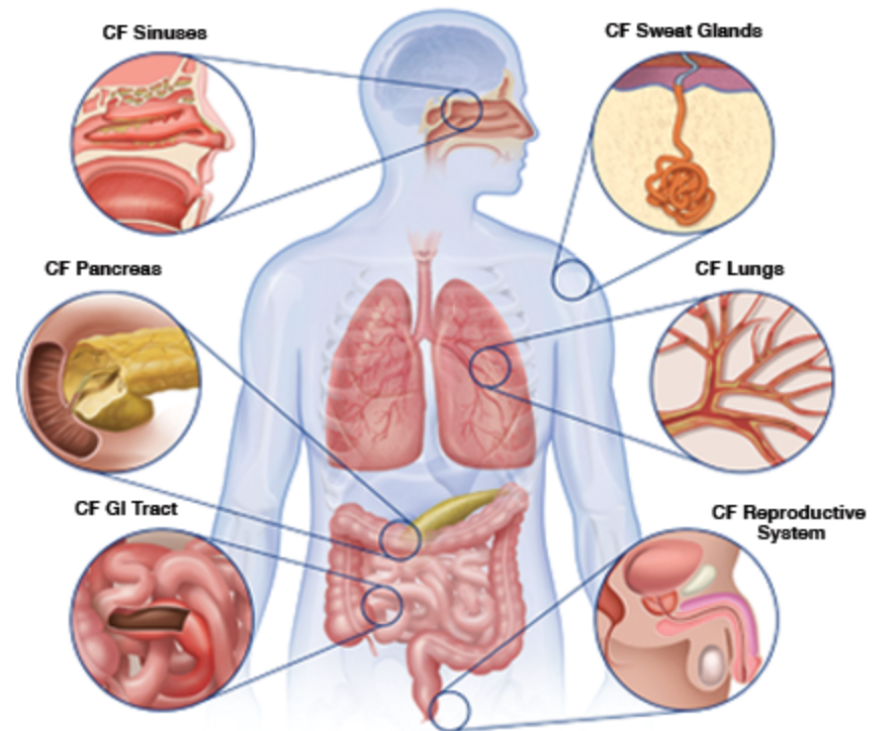
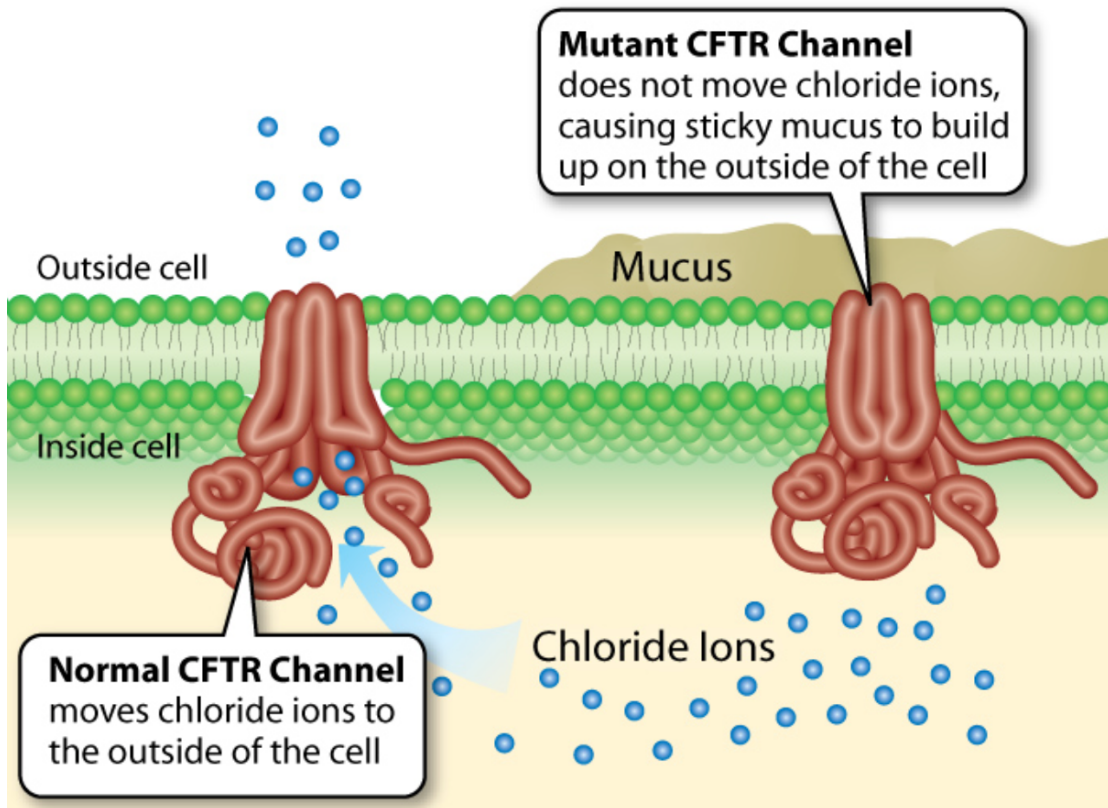
The Basic Defect in CF

(even more important because of the development of CFTR modulators)



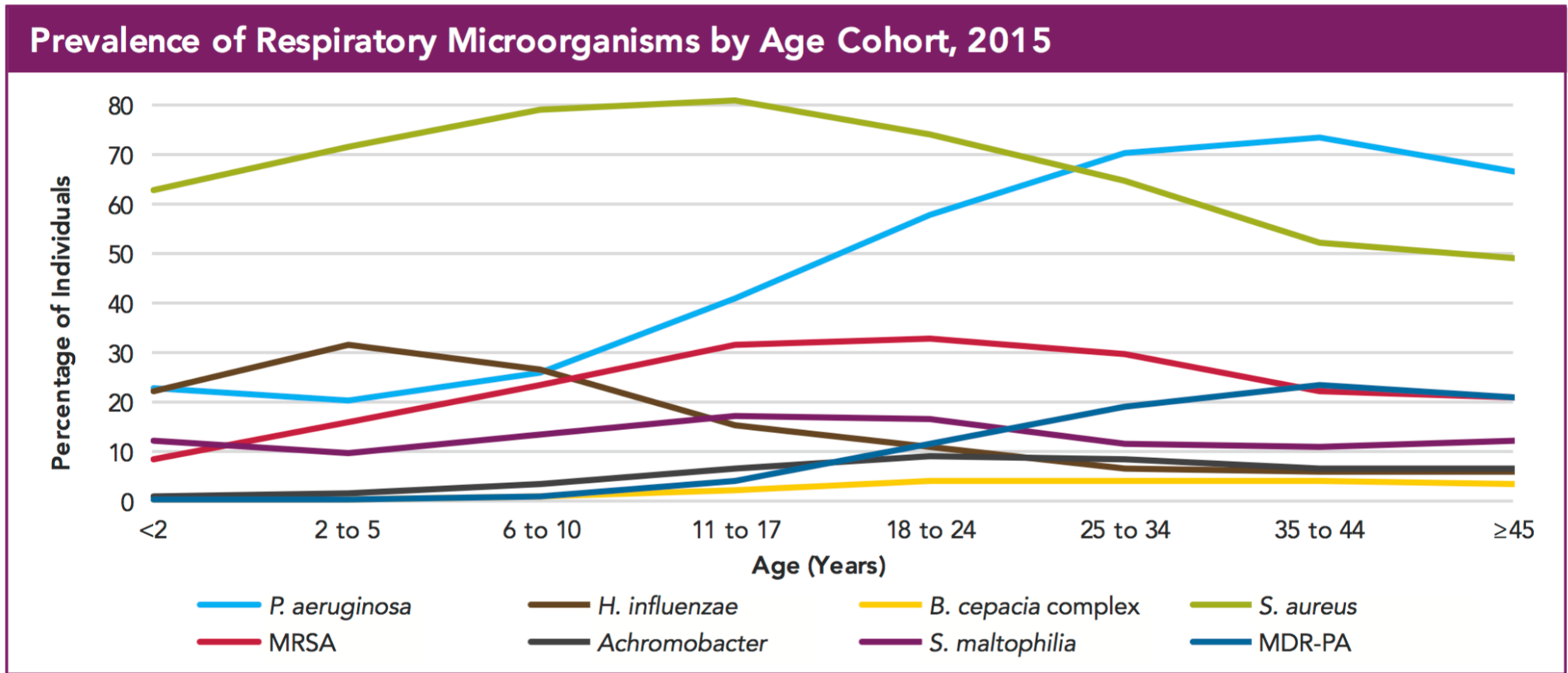
Autosomal recessive disorder, CFTR gene located on chromosome 7

Pathophysiology



obstruction -> infection -> inflammation -> destruction

Common bacterial organisms



Steno – Bactrim TOC, then Levaquin or Ceftazidime

Burkholderia – contraindication to transplant in most centers

Diagnosis in adults

Table 2—Phenotypic Features Consistent With a Diagnosis of Cystic Fibrosis

Chronic sinopulmonary disease manifested by:

Persistent colonization/infection with typical CF pathogens including *Staphylococcus aureus*, nontypeable *Haemophilus influenzae*, mucoid and nonmucoid *P aeruginosa*, and *B cepacia*

Chronic cough and sputum production

Persistent chest radiograph abnormalities (eg, bronchiectasis, atelectasis, infiltrates, and hyperinflation)

Airway obstruction manifested by wheezing and air trapping

Nasal polyps, and radiograph or CT scan abnormalities of the paranasal sinuses

Digital clubbing

GI and nutritional abnormalities, including:

Intestinal: DIOS and rectal prolapse

Pancreatic: pancreatic insufficiency and recurrent pancreatitis

Hepatic: chronic hepatic disease manifested by clinical or histologic evidence of focal biliary cirrhosis or multilobular cirrhosis

Nutritional: failure to thrive (protein-calorie malnutrition), hypoproteinemia and edema, and complications secondary to fat-soluble vitamin deficiency

Salt loss syndromes: acute salt depletion and chronic metabolic alkalosis

Male urogenital abnormalities resulting in obstructive azoospermia

- Sweat test: < 40 normal, 40 – 60 borderline, > 60 abnormal
- CFTR mutation analysis (most common genes) or full sequencing
- Nasal PD, semen analysis

Chronic respiratory regimen

- Airway clearance and exercise
- Bronchodilators (albuterol), don't routinely use anticholinergics
- Inhaled antibiotics – TOBI, Cayston FDA-approved (Colistin, Fortaz, Vanc, Meropenem)
- Macrolides (avoid in NTM)
- DNase or Pulmozyme (once daily, mild disease)
- Hypertonic saline 3 – 7% (twice daily, mild disease)
- CFTR modulators
 - ivacaftor – one copy of G551D
 - Ivacaftor/lumacaftor - homozygous dF508

Chronic nutrition regimen

- Pancreatic enzyme supplements (500-2500 U lipase/kg/meal)
- PPI for acid suppression (enhances PERT or GERD)
- Vitamin supplementation (A, D, E, K), iron, zinc
- Evaluate yearly for CF Related Diabetes (OGTT)
- Evaluate for CF Related Bone Disease (DEXA)
- Evaluate yearly for liver disease (LFTs +/- RUQ US)

CF Pulmonary Exacerbations

- DDX includes (among many things) exacerbation of chronic bacterial growth, acquisition of new bacteria, ABPA, NTM, pneumothorax, sinus disease, reflux
- Mild to moderate – oral quinolone
- Moderate to severe – IV, use 2 anti-pseudomonals
- Aminoglycoside dosing – 10 mg/kg/day tobra Q24
- Consider continuous infusion of Beta lactams
- Consider short course of oral steroids
- Airway clearance, Oxygen supplementation, NIV

Complications

- Respiratory:
 - pneumothorax - consider if patient is future transplant candidate, try to avoid VATS/pleurodesis
 - massive hemoptysis – consider embolization
- Infectious:
 - ABPA – check IgE, aspergillus antigen, treat with steroids, antifungals
 - NTM – AFB sputum annually, CT Chest, ATS/IDSA criteria
- GI:
 - DIOS – gastrograffin enema
 - pancreatitis, intusseption, colon CA

CFTR modulators

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**A CFTR Potentiator in Patients
with Cystic Fibrosis and the *G551D* Mutation**

Bonnie W. Ramsey, M.D., Jane Davies, M.D., M.B., Ch.B., N. Gerard McElvaney, M.D., Elizabeth Tullis, M.D.,
Scott C. Bell, M.B., B.S., M.D., Pavel Dřevínek, M.D., Matthias Griesse, M.D., Edward F. McKone, M.D.,
Claire E. Wainwright, M.D., M.B., B.S., Michael W. Konstan, M.D., Richard Moss, M.D., Felix Ratjen, M.D., Ph.D.,
Isabelle Sermet-Gaudelus, M.D., Ph.D., Steven M. Rowe, M.D., M.S.P.H., Qunming Dong, Ph.D., Sally Rodriguez, M.S.,
Karl Yen, M.D., Claudia Ordoñez, M.D., and J. Stuart Elborn, M.D., for the VX08-770-102 Study Group*

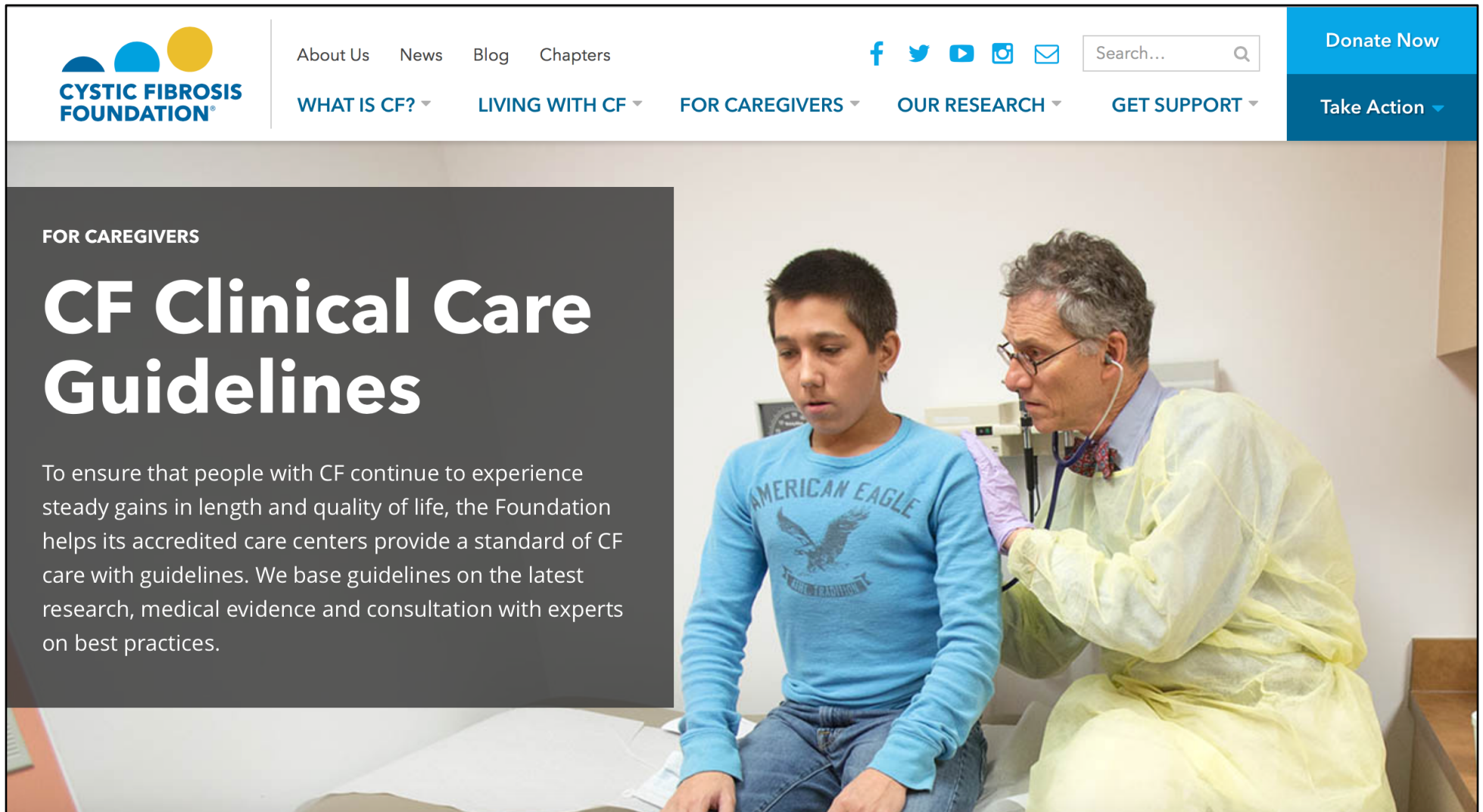
ORIGINAL ARTICLE

**Lumacaftor–Ivacaftor in Patients with Cystic
Fibrosis Homozygous for Phe508del *CFTR***

C.E. Wainwright, J.S. Elborn, B.W. Ramsey, G. Marigowda, X. Huang, M. Cipolli,
C. Colombo, J.C. Davies, K. De Boeck, P.A. Flume, M.W. Konstan, S.A. McColley,
K. McCoy, E.F. McKone, A. Munck, F. Ratjen, S.M. Rowe, D. Waltz,
and M.P. Boyle, for the TRAFFIC and TRANSPORT Study Groups*

Clinical Care Guidelines

(basically review articles)



The image is a screenshot of the Cystic Fibrosis Foundation website. At the top left is the logo, which consists of three overlapping circles in blue, yellow, and blue, with the text "CYSTIC FIBROSIS FOUNDATION" below it. To the right of the logo is a navigation menu with links for "About Us", "News", "Blog", and "Chapters". Further right are social media icons for Facebook, Twitter, YouTube, Instagram, and Email. A search bar with the placeholder text "Search..." and a magnifying glass icon is also present. On the far right of the top navigation bar are two buttons: "Donate Now" and "Take Action". Below the navigation bar is a horizontal menu with dropdown arrows for "WHAT IS CF?", "LIVING WITH CF", "FOR CAREGIVERS", "OUR RESEARCH", and "GET SUPPORT". The main content area features a dark grey box on the left with the text "FOR CAREGIVERS" in white, followed by the large white heading "CF Clinical Care Guidelines". Below this heading is a paragraph of text: "To ensure that people with CF continue to experience steady gains in length and quality of life, the Foundation helps its accredited care centers provide a standard of CF care with guidelines. We base guidelines on the latest research, medical evidence and consultation with experts on best practices." To the right of this text is a photograph of a doctor in a yellow protective gown and purple gloves using a stethoscope to examine a young boy's chest. The boy is wearing a blue long-sleeved shirt with an "AMERICAN EAGLE" logo.

FOR CAREGIVERS

CF Clinical Care Guidelines

To ensure that people with CF continue to experience steady gains in length and quality of life, the Foundation helps its accredited care centers provide a standard of CF care with guidelines. We base guidelines on the latest research, medical evidence and consultation with experts on best practices.