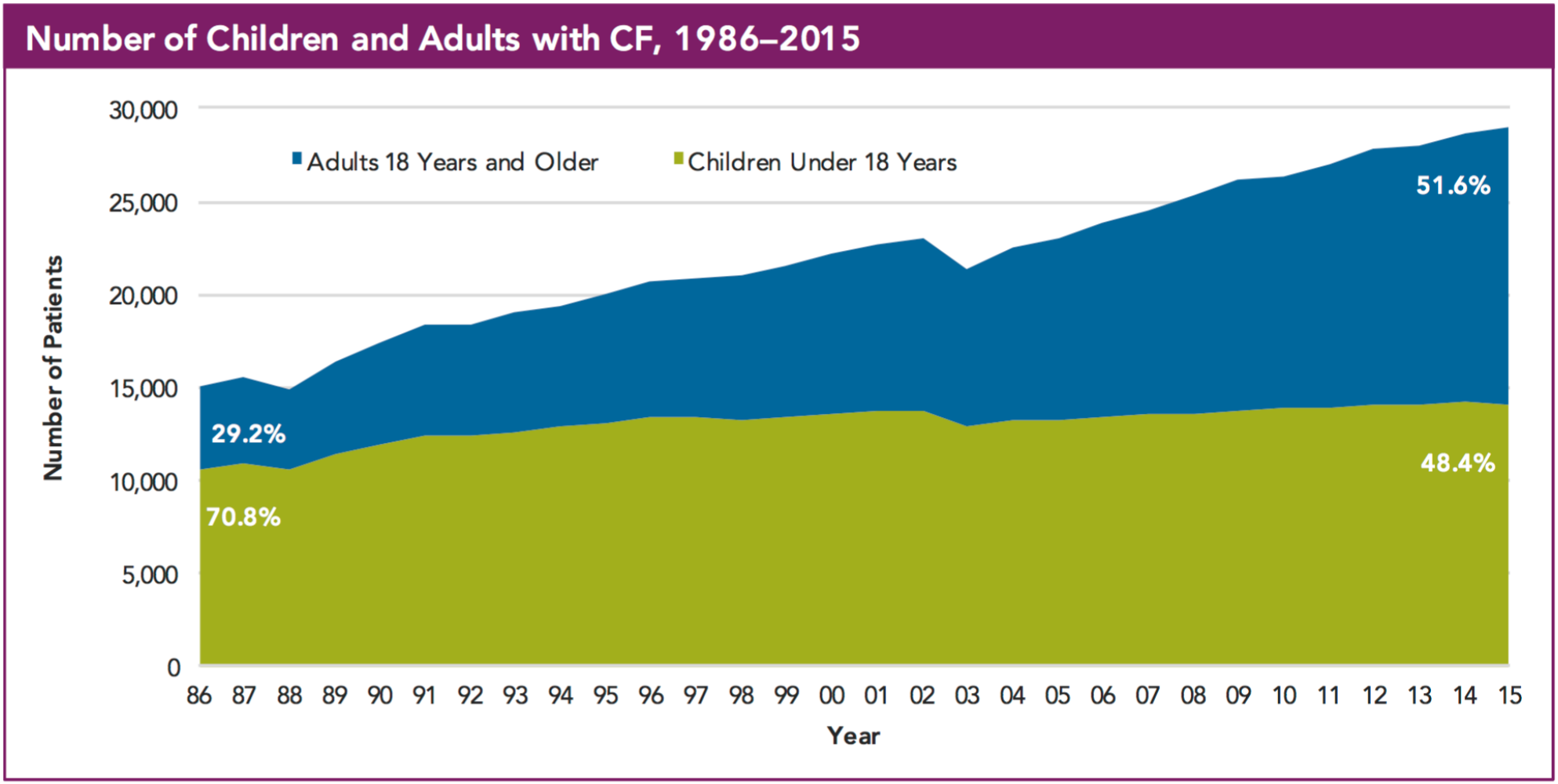
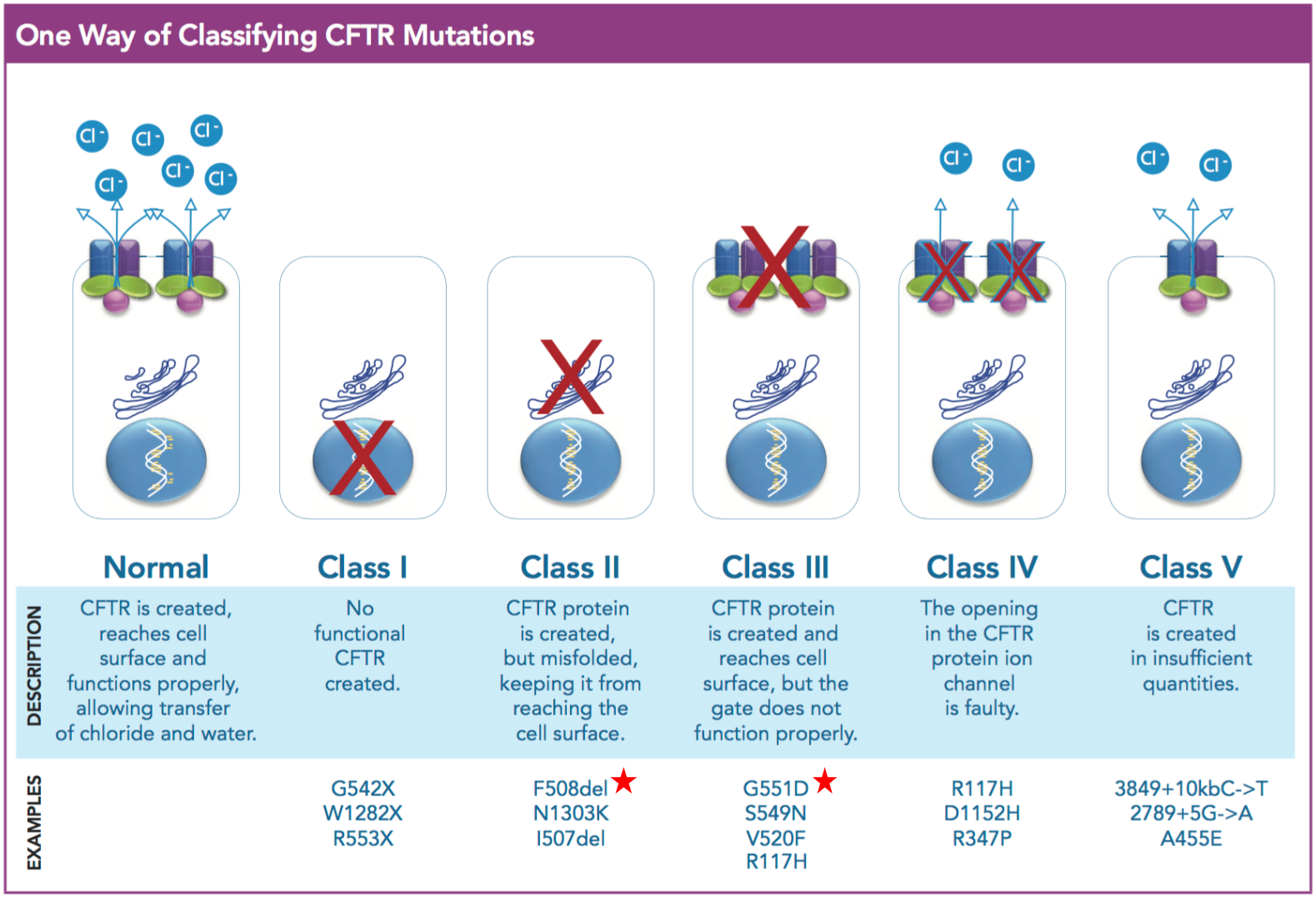
Why it's increasingly important for adult pulmonologists to learn about CF



Median predicted survival 41-42 years

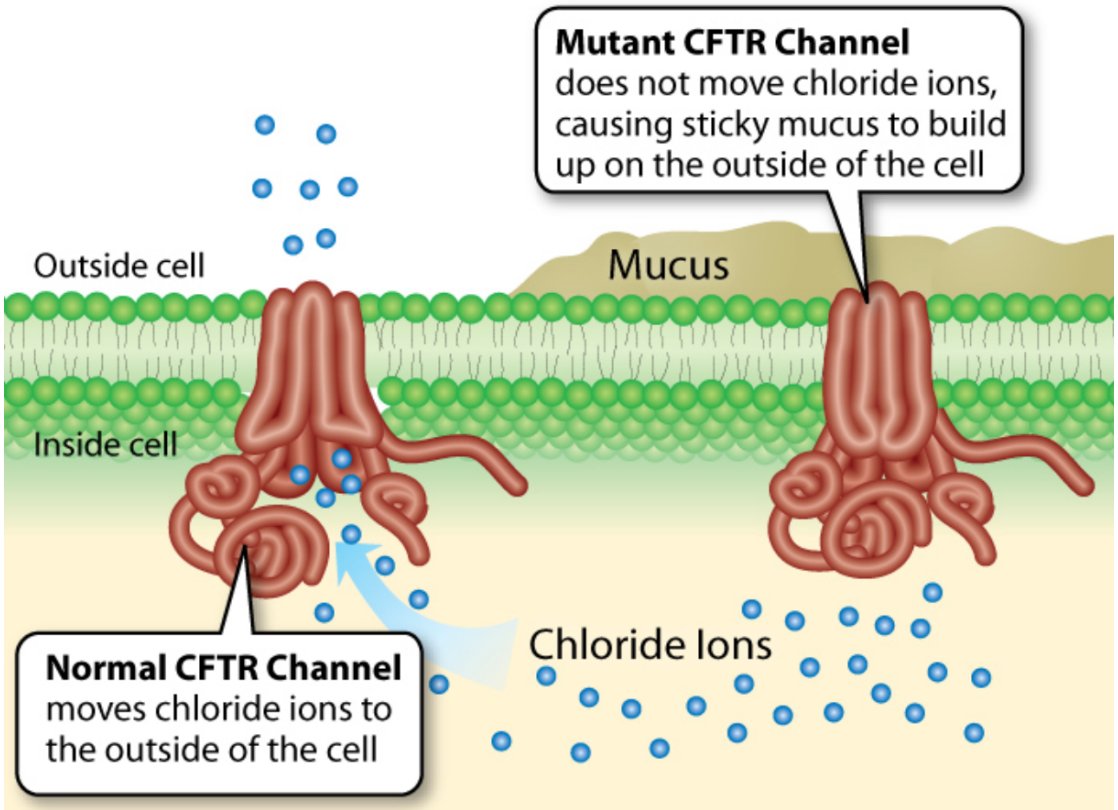
The Basic Defect in CF

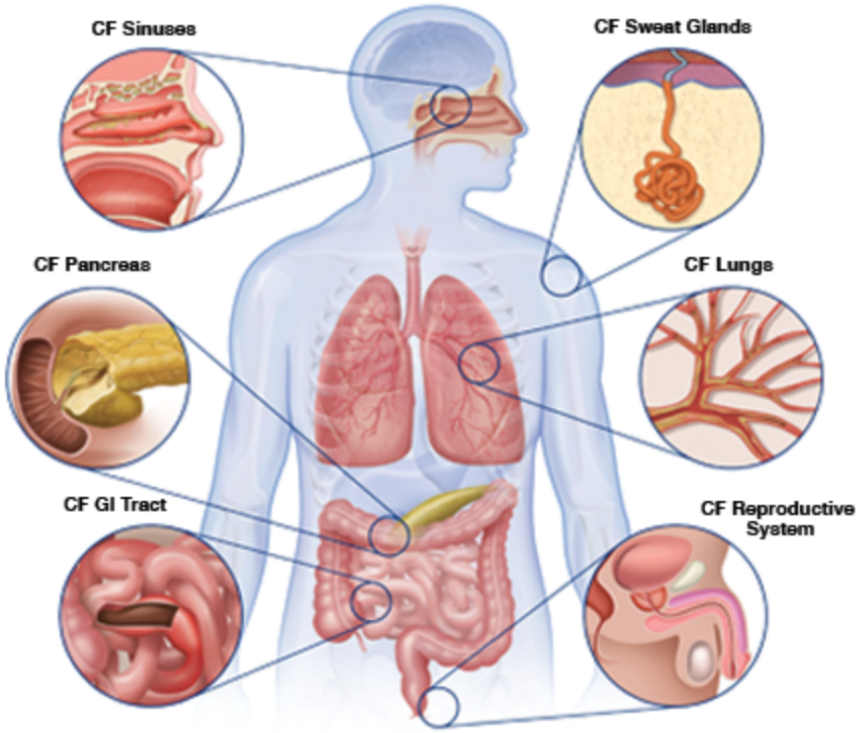
(even more important because of the development of CFTR modulators)



Autosomal recessive disorder, CFTR gene located on chromosome 7

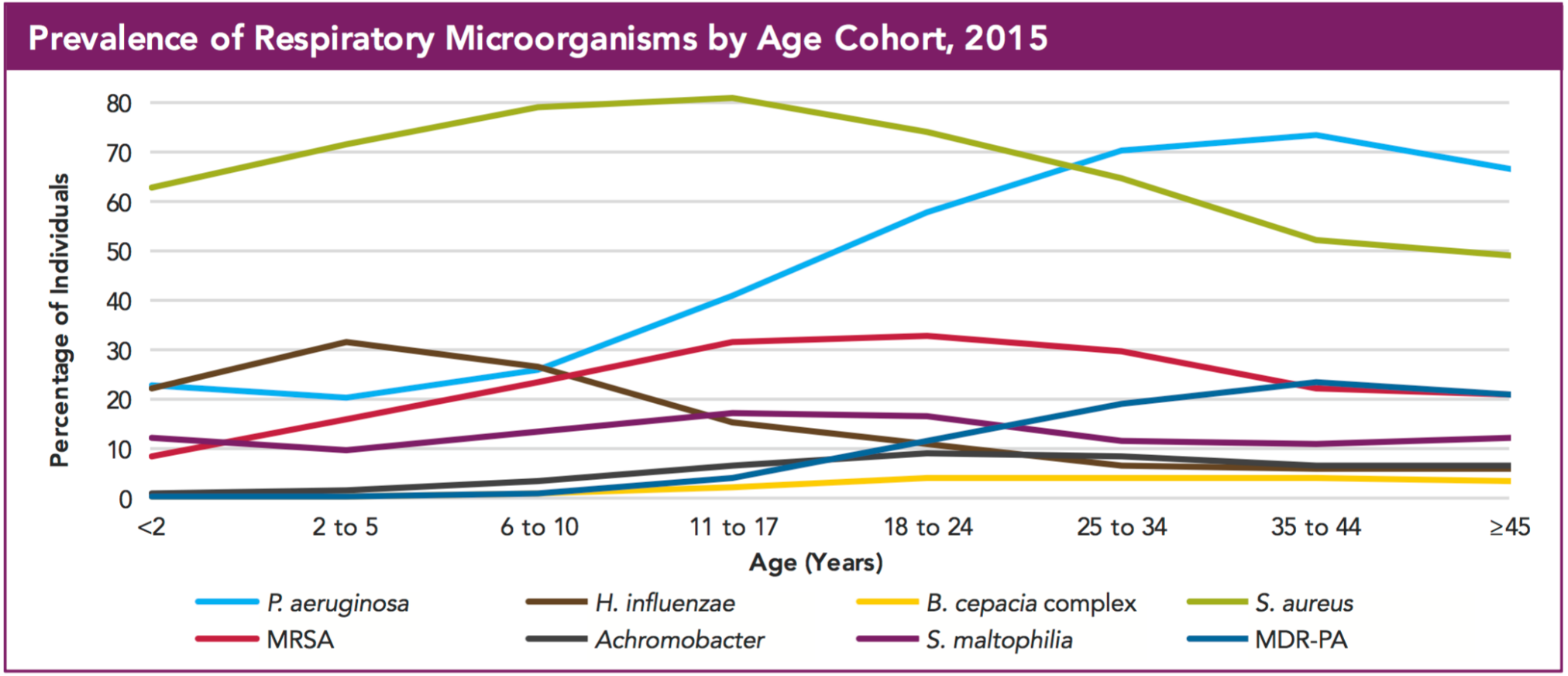
Pathophysiology





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, intusseception, colon CA

CFTR modulators

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

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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)