Cystic Fibrosis
Cystic Fibrosis (CF) is an autosomal recessive disease affecting pulmonary, gastrointestinal, and exocrine gland function.

CF is the most common life shortening genetic disease in the Caucasian population.

Mutations in the gene that encodes for the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) protein cause CF.
Daily Schedule of a Child with CF

* Get up in the morning and before breakfast
  
  Inhaled bronchodilator (5-10 minutes)
  
  Inhaled mucus modulator (pulmozyme or hypertonic saline) (10-20 minutes)
  
  Airway clearance (20-30 minutes)
  
  Inhaled antibiotic (TOBI/Cayston) (7-20 minutes)

Breakfast (Vitamins and enzymes (possible acid blockade))

Lunch (enzymes)

ACT (before lunch or after school)

Dinner (enzymes/ acid blockade)

After dinner ACT

+ Liver disease, ++++Diabetes, +Anxiety/Depression

2.5 to 4.5 hours at a minimum of daily therapy
Daily Costs of Cystic Fibrosis

- Monetary
- Time (patient and family)
- Resources
- Quality of life

![Chart showing annual medical costs for Pediatric Group (n=439) and Adult Group (n=625).]
What does a new parent think of when the doctor says Cystic Fibrosis?

Initially they only feel:
Fear
Anxiety
Dread
Panic
Sadness
Rage
CF clinic care
- Comprehensive Multidisciplinary Care
- Quality Improvement Program
- Community Advocacy/Education

CF basic science research
- Small molecules that activate and correct CFTR
- Gene editing / Stem cells/repairing the damage
- Calming the inflammatory response
- Examining Pseudomonas aeruginosa
- Examining the microbiome

CF clinical research
- Clinical Trial Network: Therapeutic Development Network (TDN)
- Industry and PI initiated studies
- QI Program
- Community and patient education and advocacy

Diagram:
- BEDSIDE
- BENCH
Quarterly visits (at a minimum) monitoring growth, pulmonary function, liver function, screening for CFRD, Anxiety and depression

Multidisciplinary approach (nutrition, social work, nursing, respiratory therapy, physical therapy, psychology, pharmacology, research coordination, medical treatment, patient and family advisory board)

Treatment Guidelines: developed by multidisciplinary teams of experts and are based on a nationwide data that is collected from all accredited CF Centers. The guidelines are evidence based (Inpatient and Outpatient practice guidelines and age specific) Annual Labs and Routine Microbiology Surveillance

Education: families, patients, schools, communities, work places, coaches....
Model of care

- Weekly meetings: Review and discuss patients
- Biweekly Center meetings: Review age specific treatment/care guidelines
- Patient reviews: Review problems since last visit, goals from last visit
- Biweekly QI meetings
- Monthly Patient and Family Advisory Boards
- Quarterly educational meetings
- Annual Retreats
Cystic fibrosis: median survival age, 1940-2007

Median survival age (years)

Year


Nutritional Needs were Re-evaluated

In 2016 > 40 yrs

- anti-Staphylococcus antibiotics
- anti-Pseudomonas antibiotics
- airway clearance
- inhaled antibiotics
- inhaled mucolytics

0 5 10 15 20 25 30 35


2010

37.8
Drug Development: Approach to Restoring CFTR Function

*Potentiators:* Increase the flow of ions through CFTR present at the cell surface

*Correctors:* Increase the cellular processing and delivery of CFTR proteins to the cell surface
Demonstrated that restoring CFTR function results in dramatic improvement in clinical outcomes for individuals with CF.

Gave us the ability to further understand the clinical benefits of restoring CFTR function.
Effect of Decreased Rate of Decline in FEV$_1$

- **G551D With Ivacaftor**
- **F508del/F508del**

---

Lung Transplant

**FEV1 % Predicted**

**Age in Years**

- 6
- 16
- 26
- 36
- 46
- 56
- 66
- 76
- 86
Restoring CFTR Function Reduces Hospitalizations

Rowe, Heltshe,…Ramsey et al. AJRCCM 2014

- Improvement in weight/BMI
- Altered the microbiology

* p<0.001
Towards “Personalized” CF Care: Genomics and Proteomics

- Genomics
  - Microarrays

- Proteomics

= 

- Modifier Genes
- Pathways of Injury
- Individual Variation
- New Treatments
- Individual Treatment Approaches
Areas of Need

- Transition from Pediatric to Adult Care
- Insurance needs (underinsured) for medications, hospitalizations, and medical devices
- Genetic counseling support for newborn screening programs
- Advocacy for job security for patients and for families
- Ability to be seen at an accredited CF Center
- Mental Health Providers skilled in Chronic Illness
Questions