



Cystic Fibrosis

What is CF?

CF is the most common life-shortening autosomal recessive disorder in the US

- Affects about 33,000 people in the US and 70,000 worldwide
- 1 in 31 people are carriers

cff.org

What is CF?

Now diagnosed primarily by newborn screen

- Some diagnoses are made in childhood and adulthood
- Confirmed through sweat test, genetic test and clinical evaluation

cff.org

What is CF?

Caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene

- More than 2000 mutations (cfr2.org)
- Loss of CFTR function affects sodium and chloride ion channels, altering the flow of salt and fluids in and out of cells

DESCRIPTION	Normal	Class I	Class II	Class III	Class IV	Class V
CFTR normally transports sodium and chloride ions out of the cell. In the normal CFTR, the channel is always open, allowing for the flow of salt and fluids.		Class I mutations are associated with a defective CFTR protein that is not produced in the cell.	Class II mutations are associated with a defective CFTR protein that is produced but does not function properly.	Class III mutations are associated with a defective CFTR protein that is produced but does not function properly.	Class IV mutations are associated with a defective CFTR protein that is produced but does not function properly.	Class V mutations are associated with a defective CFTR protein that is produced but does not function properly.
PREVALENCE		22%	88%	6%	6%	5%
MUTATION		G542X, W1282L, R1156G	F508del, S1231R, N1303K, R1172G, R1174G, R1175G, R1176G, R1177G, R1178G, R1179G, R1180G, R1181G, R1182G, R1183G, R1184G, R1185G, R1186G, R1187G, R1188G, R1189G, R1190G, R1191G, R1192G, R1193G, R1194G, R1195G, R1196G, R1197G, R1198G, R1199G, R1200G, R1201G, R1202G, R1203G, R1204G, R1205G, R1206G, R1207G, R1208G, R1209G, R1210G, R1211G, R1212G, R1213G, R1214G, R1215G, R1216G, R1217G, R1218G, R1219G, R1220G, R1221G, R1222G, R1223G, R1224G, R1225G, R1226G, R1227G, R1228G, R1229G, R1230G, R1231G, R1232G, R1233G, R1234G, R1235G, R1236G, R1237G, R1238G, R1239G, R1240G, R1241G, R1242G, R1243G, R1244G, R1245G, R1246G, R1247G, R1248G, R1249G, R1250G, R1251G, R1252G, R1253G, R1254G, R1255G, R1256G, R1257G, R1258G, R1259G, R1260G, R1261G, R1262G, R1263G, R1264G, R1265G, R1266G, R1267G, R1268G, R1269G, R1270G, R1271G, R1272G, R1273G, R1274G, R1275G, R1276G, R1277G, R1278G, R1279G, R1280G, R1281G, R1282G, R1283G, R1284G, R1285G, R1286G, R1287G, R1288G, R1289G, R1290G, R1291G, R1292G, R1293G, R1294G, R1295G, R1296G, R1297G, R1298G, R1299G, R1300G, R1301G, R1302G, R1303G, R1304G, R1305G, R1306G, R1307G, R1308G, R1309G, R1310G, R1311G, R1312G, R1313G, R1314G, R1315G, R1316G, R1317G, R1318G, R1319G, R1320G, R1321G, R1322G, R1323G, R1324G, R1325G, R1326G, R1327G, R1328G, R1329G, R1330G, R1331G, R1332G, R1333G, R1334G, R1335G, R1336G, R1337G, R1338G, R1339G, R1340G, R1341G, R1342G, R1343G, R1344G, R1345G, R1346G, R1347G, R1348G, R1349G, R1350G, R1351G, R1352G, R1353G, R1354G, R1355G, R1356G, R1357G, R1358G, R1359G, R1360G, R1361G, R1362G, R1363G, R1364G, R1365G, R1366G, R1367G, R1368G, R1369G, R1370G, R1371G, R1372G, R1373G, R1374G, R1375G, R1376G, R1377G, R1378G, R1379G, R1380G, R1381G, R1382G, R1383G, R1384G, R1385G, R1386G, R1387G, R1388G, R1389G, R1390G, R1391G, R1392G, R1393G, R1394G, R1395G, R1396G, R1397G, R1398G, R1399G, R1400G, R1401G, R1402G, R1403G, R1404G, R1405G, R1406G, R1407G, R1408G, R1409G, R1410G, R1411G, R1412G, R1413G, R1414G, R1415G, 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What is CF?

Thick, sticky mucus builds up and blocks the ducts of the exocrine glands, including the lungs and pancreas

- Mucus build up causes chronic infection, inflammation, and damage

Symptoms of CF

People with CF can have a variety of symptoms, including:

- Very salty-tasting skin
- Persistent coughing, at times with phlegm
- Frequent lung infections including pneumonia or bronchitis
- Wheezing or shortness of breath
- Poor growth or weight gain in spite of a good appetite
- Frequent greasy, bulky stools or difficulty with bowel movements
- Male infertility

MANIFESTATIONS OF CYSTIC FIBROSIS

NOSE/NASE

- Nasal mucus
- Crusting

STOMACH

- Gas/bloating
- Constipation

LIVER

- Ascites (swelling)
- Biliary liver

INTESTINES

- Abdominal pain
- Abnormal gut bacteria
- Poor growth
- Poorly digested stool
- Frequent constipation
- Diarrhea
- Inflammation of the intestines
- Inflammation of the colon

HEART

- Right ventricular hypertrophy
- Pulmonary artery stenosis

LUNGS

- Chronic cough
- Wheezing
- Shortness of breath
- Frequent lung infections
- Sputum production
- Clubbing (swelling of the ends of fingers)
- Hemoptysis (coughing up blood)
- Chronic respiratory failure

SPLEEN

- Hypertrophy

PANCREAS

- Pancreatitis
- Male infertility
- Exocrine insufficiency
- Exocrine insufficiency (pancreatic insufficiency)

GALLBLADDER

- Biliary cirrhosis
- Gallstones
- Cholelithiasis

REPRODUCTION

- Infertility (especially in males)
- Abnormal sperm
- Abnormal eggs
- Abnormal embryos

SKIN

- Very salty-tasting skin
- Persistent coughing
- Wheezing or shortness of breath

Case #1

During a conversation, a mother lets you know that her child is a carrier of the CF gene. You should:

- Make sure that you have a treatment plan for him.
- Do nothing ✓
- Learn more about CF and guidelines
- Make sure you keep him away from other children with CF in your school

People with CF do best when:

- They have highly individualized treatment
- They can balance their treatments with daily life
- Their care plans address the whole person—physical, mental and social





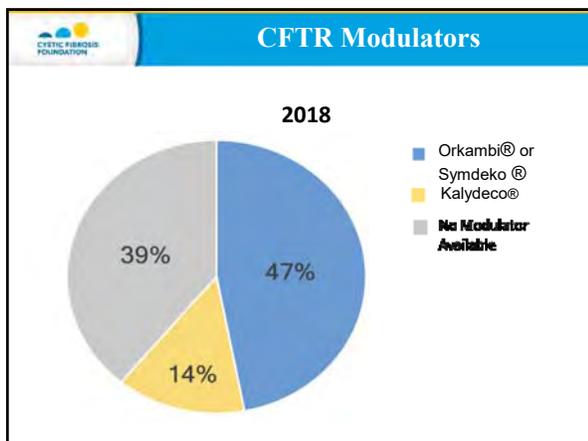


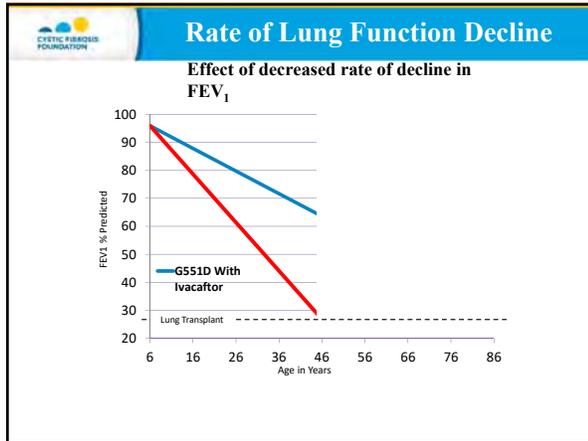
COORDINATING

Treatments and Therapies



Highly effective CFTR Modulators Will Completely Change CF Care





-
- Next Generation CFTR Modulator Combinations**
Phase 2 Trial Results
- **Patients with one copy of F508 del**
 - 10%+ absolute improvement in FEV₁, compared to placebo
 - 30+ mmol/L drop in sweat chloride
 - Remarkable improvement in quality of life
 - **Patients with two copies of F508del**
 - 9% absolute improvement in FEV₁, compared to Tezacaftor and Ivacaftor
 - 30+ mmol/L drop in sweat chloride
 - **No safety concerns noted**

CYSTIC FIBROSIS FOUNDATION

CF is leading the medical field in "personalized" medicine



Work on "Rare Mutations"

- There are over 1000 CFTR mutations that 5 or less people in the world have
- We can't do standard clinical trials to make sure everyone who would benefit will receive drug



Even with CFTR Modulators, a whole generation will have to deal with lung disease



In 10 years, over 50% of patients in the U.S. will still need symptomatic therapies despite potent new CFTR therapies



You can maximize your student's overall learning experience, while helping to maintain his or her health.



Airway Clearance

To be performed 2-3 times per day

- Bronchodilator
- Mucus thinners
 - Hypertonic saline
 - Pulmozyme® (Dornase Alfa)
- Airway clearance technique
 - Manual chest physical therapy
 - High-frequency chest wall oscillation (vest)
 - Positive expiratory pressure (PEP) or oscillating PEP
 - Autogenic drainage
- Inhaled antibiotics
- Inhaled corticosteroids



Cough at school

- Chronic
- Moves mucus out of large airway
- Not contagious
- Self conscious



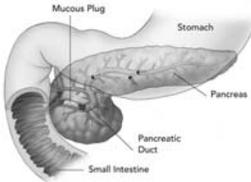
In school accommodation:

- Nebulization treatment
- Airway clearance
 - Child can and should bring school work with them during the required treatment.
- Water, tissues, hand sanitizer at desk



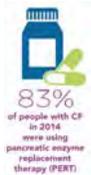
Nutrition

- Increase caloric requirement
- Obstruction of digestive system
 - Require vitamins
 - High-calorie, high-fat foods
- CF-related diabetes
- Gastroesophageal reflux
- Oral enzymes
 - Steatorrhea
 - Abdominal pain



Enzymes

- Aid in digestion of carbohydrates, protein and fats
- Brand necessary
- Individualized by dosage and brand
 - Lipase (digest fat)
 - Protease (digest protein)
 - Amylase (digest carbohydrate)
- To be given just before eating (meals and snacks)
- Room temperature



 **In school accommodation:**

- Enzymes
- Caloric requirement of child with CF
- Bathroom usage
 - Freedom
 - Privacy
 - Odor



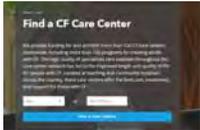
 **Pulmonary Exacerbation**

- Chronic bacterial infections
- Acute exacerbation

Staphylococcus aureus (Staph)
Pseudomonas (P. aeruginosa)
Methicillin-Resistant *Staphylococcus aureus* (MRSA)
Burkholderia cepacia (*B. cepacia*)
Nontuberculous Mycobacteria (NTM)

 **In school accommodation:**

- Partner with the child's care center

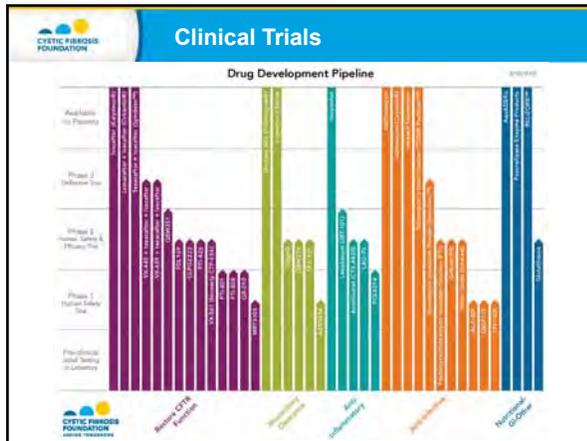


- Individualized Education Plans (IEPs)



IEP/504 plans

- Time needed for medication or therapy
- Adjust school rules to allow child to self administer enzymes
- Have a plan when child is hospitalized or home sick
- Audio or video tapes of class when missed
- Access to bathroom in nurses office
- Don't limit bathroom or water
- School medical emergency plan

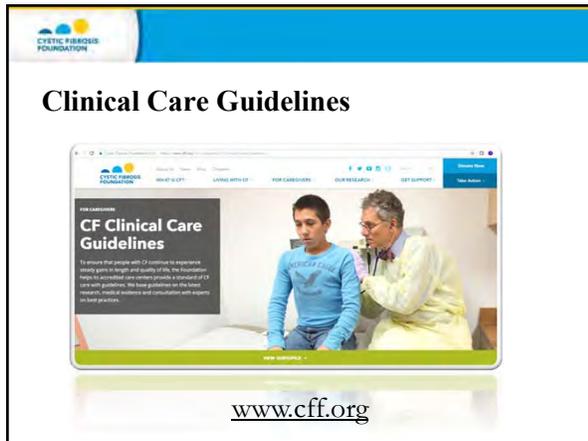


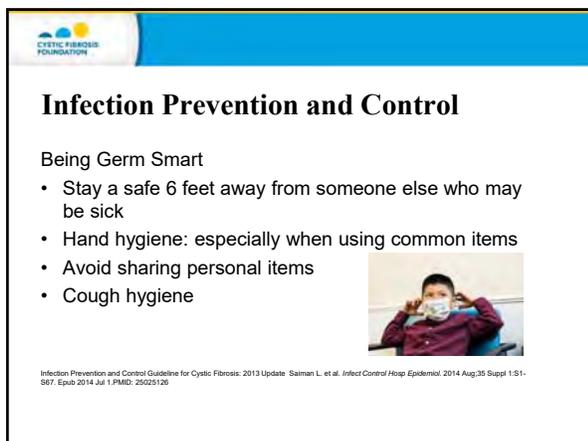
Case #2

Sam is in 5th grade and constantly forgets to come to the office for his enzymes. You should:

- See if you can work out a new plan ✓
- Do nothing
- Give them to him after he eats lunch
- Recommend that he take his enzymes before coming to school.







When there is more than one child with CF in your school

- Encourage disclosure
- Minimize the time in the same area (6 ft rule)
 - Separate classrooms
 - Separate bathrooms and water fountains
 - Schedule common areas (gym, cafeteria) at different times
 - Assign lunch tables, lockers as far away from each other as possible
 - Make sure not at nurses office at the same time
 - Make sure teachers know to notify nurse when sending student with CF to the nurses office

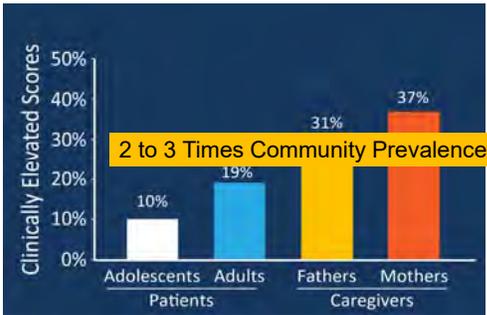


Resources@cff.org

Mental Health



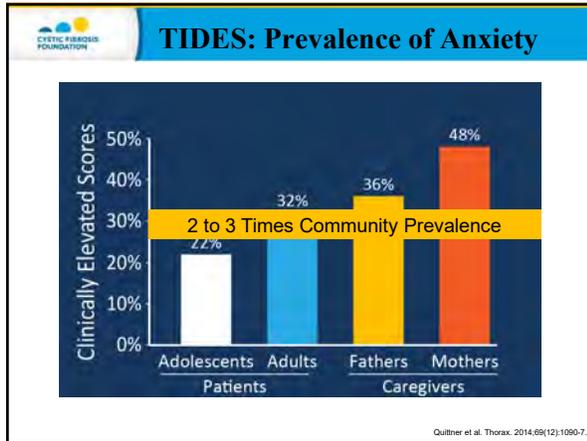
TIDES: Prevalence of Depression

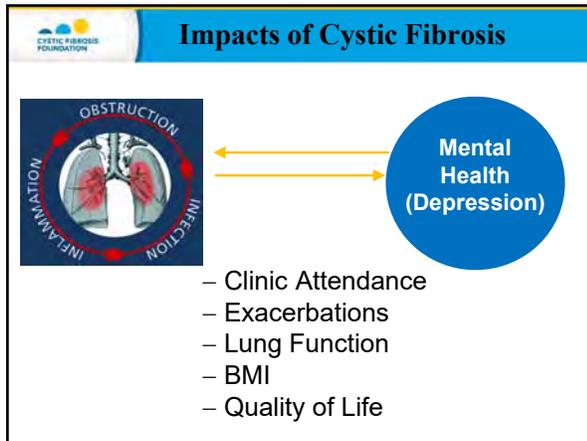


Group	Clinically Elevated Scores
Adolescents	10%
Adults	19%
Fathers	31%
Mothers	37%

2 to 3 Times Community Prevalence

Quilmer et al. Thorax. 2014;69(12):1090-7.





CF related diabetes (CFRD)

- Not type I or type II
- Like type I
 - Insulin deficient
 - Due to scarring of pancreas
- Like type II
 - Insulin resistant
 - Could be due to inflammation, infection, steroid usage

Moran, A. et al. Diabetes Care 2010. Dec. 33(12):2697-2708

 **CF related diabetes (CFRD)**

- Not treated with diet and exercise
- Not treated with orals
- Do not limit calories to 1200!
- May have times when don't need insulin

Moran, A. et al. Diabetes Care 2010. Dec. 33(12):2697-2708

 **Symptoms CFRD**

- Polyuria
- Polydipsia
- Lethargy
- **Weight loss**
- Ketoacidosis is not usually associated with CFRD

Annual screening: OGTT beginning at age 10

 **Case #3**

You have 2 children in your school with cystic fibrosis.
You should:

- a) Screen them for anxiety and depression
- b) Review documents on cff.org
- c) Make sure their lockers are far apart
- d) B and C 
- e) All of the above



BELOGING



CYSTIC FIBROSIS FOUNDATION

Mission

To cure CF and provide all people with the disease the opportunity to lead full, productive lives by funding research and drug development, promoting individualized treatment and ensuring access to high-quality, specialized care.



<https://www.cff.org/About-Us/About-the-Cystic-Fibrosis-Foundation/Our-Mission/>



Accredited Care Centers

- Comprised of pediatric and adult care programs and affiliate programs
- 126 centers in the United States
- 285 Programs:
 - 123 pediatric
 - 110 adult
 - 52 affiliates



<https://www.cff.org/Care-Centers/Find-a-CF-Care-Center/>

Treatment Burden

Therapy	Time per treatment	Frequency (minimum)	Total time spent daily
Inhaled bronchodilators (such as β ₂ -adrenergic agonists, anticholinergics)	5 minutes	2–3 times/day	10–15 minutes
Inhaled mucolytics such as <ul style="list-style-type: none"> • hypertonic saline • dornase alfa (Pulmozyme) 	20 minutes 5 minutes	2 times/day 1 time/day	40 minutes 5 minutes
Inhaled antibiotics such as <ul style="list-style-type: none"> • tobramycin (TOBI), colistimethate (Coly-Mycin M) • aztreonam (Cayston) 	20 minutes 3 minutes	2 times/day 3 times/day	40 minutes 9 minutes
Airway clearance ^a	20–30 minutes	2 times/day	40–60 minutes

Lomas, P, Fowler, S., AJN, August 2010 • Volume 110 • Issue 8 • p 30-37



CF and School

For Teachers

Learn how to collaborate and partner with parents to create a supportive environment for students with CF. Includes a checklist for teachers and a list of resources.

Questions? Call 1.800.FIGHT.CF

 **Case #4**

You now have a better understanding of challenges of cystic fibrosis. When you have a child with CF in your school you should:

- a) Encourage the parents to connect you with their care team.
- b) Review documents on cff.org
- c) Feel free to call 1.800.FIGHT.CF with questions
- d) All of the above ✓

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