An annotated table-of-contents for the

Cystic Fibrosis New Patient Binder

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Pediatric Pulmonary Division

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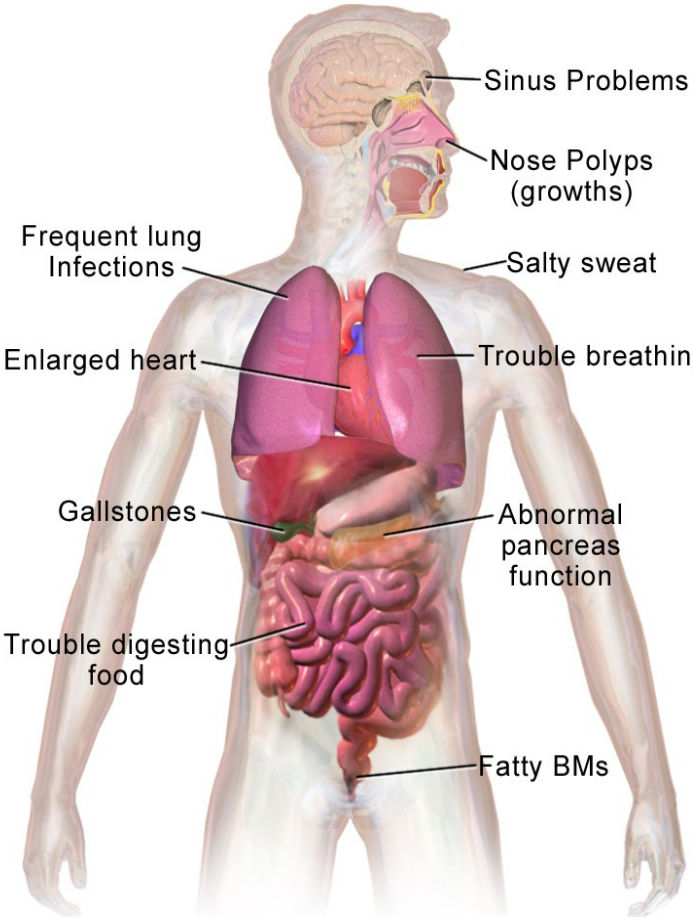
Introduction

This booklet is a summary of the key points in the detailed and comprehensive information included with the new patient binder. We know the amount of information parents of children with CF need to know is massive, and may seem daunting and overwhelming. We hope this summary will help you access and navigate the information as you need it. Each topic includes references to binder materials with more in-depth information on the topic.

Topics:

What is Cystic Fibrosis?

Cystic fibrosis (SIS-tik fi-BRO-sis), or CF, is an inherited disease of a defective gene that affects cells inside many different organs (like the lungs and pancreas), as well as cells on the surface of the body (like the skin). These cells have a protein on their surface called the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR). The CFTR gene controls how the CFTR protein is made.



Blausen.com staff (2014). "Medical gallery of Blausen Medical 2014". WikiJournal of Medicine 1 (2). DOI:10.15347/wjm/2014.010. ISSN 2002-4436. Via Wikimedia Commons

The CFTR protein controls the movement of salt and water in and out of the body's cells. In people who have CF, the defective gene causes the CFTR protein to not work well. This causes mucus in the body to become thick and sticky and also causes very salty sweat.

This glue-like mucus builds up and causes problems in many of the body's organs. CF mainly affects the lungs, pancreas, liver, intestines, sinuses, and sex organs.

How Is Cystic Fibrosis Inherited?

"Inherited" means the disease is passed from parents to children through genes. People who have CF inherit two faulty genes for the disease—one from each parent. The parents don't have the disease themselves, they are carriers for the disease.

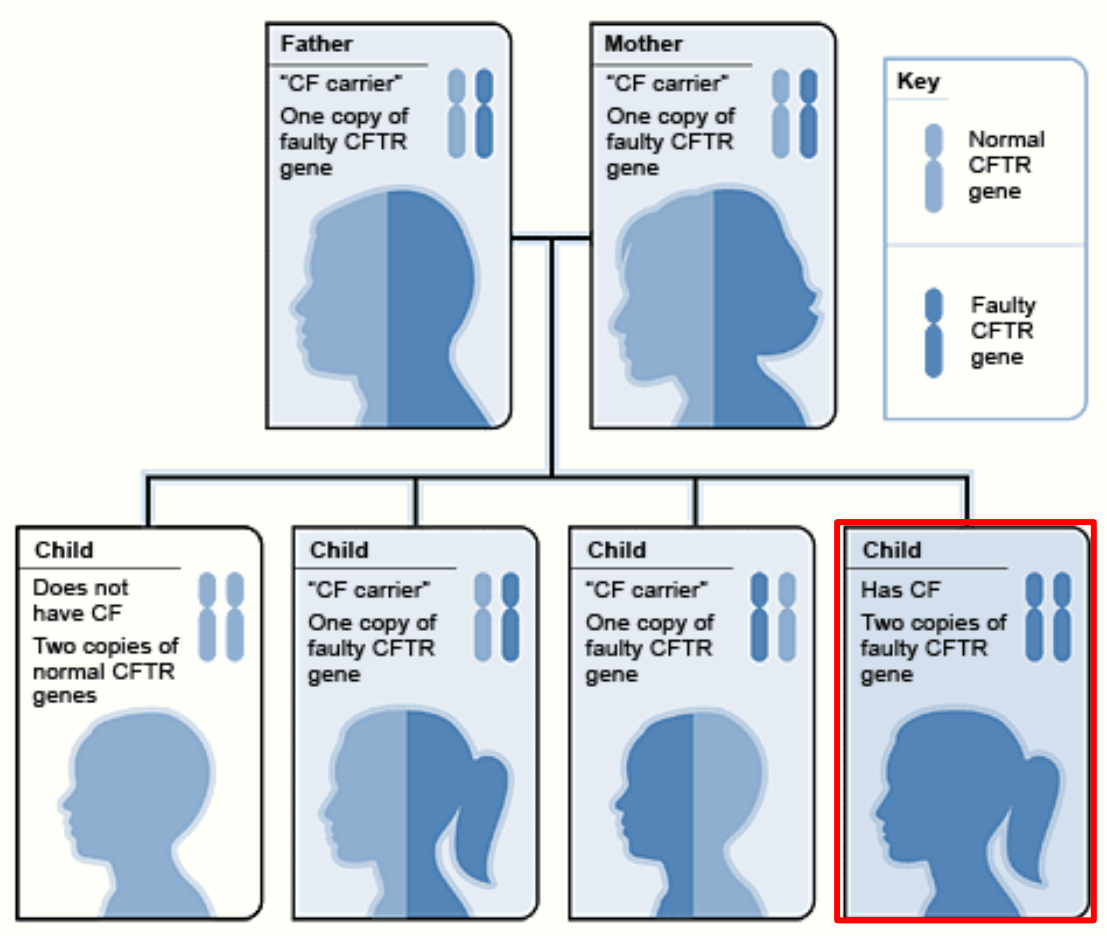
Every person inherits two CFTR genes—one from each parent. Children who inherit a faulty CFTR gene from each parent will have CF.

_____’s mutations are _____ and _____.

Children who inherit one faulty CFTR gene and one normal CFTR gene are "CF carriers." CF carriers usually have no symptoms of CF and live normal lives. However, they can pass the faulty CFTR gene to their children.

The image below shows how two parents who are both CF carriers can pass the faulty CFTR gene to their children.

Example of an Inheritance Pattern for Cystic Fibrosis



Children of parents who are carriers of the faulty CFTR gene have a:

- 1 in 4 (25%) chance to have CF.
- 1 in 2 (50%) chance to have a mutation in one of their CFTR genes, meaning they are carriers of the faulty CFTR gene, but do not have CF themselves

- 1 in 4 (25%) chance to have no mutations, meaning that both their CFTR genes are functioning. They do not have CF and are not carriers of CF.
- To learn more about your mutations, visit: http://CFTR2.org

In summary this means that each child of parents who are carriers has a 1 in 4 (25%) chance of having CF and 3 in 4 (75%) chance of not having CF.
Parents of a child who was diagnosed with CF should test all their children for CF. The child's cousins can be tested if they have symptoms of CF such as frequent illnesses, poor growth and difficulty breathing.

How does the faulty CFTR gene cause Cystic Fibrosis?

The Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene makes a protein which is found in the cells of many organs including the lungs, liver, skins and intestines (colon and bowel). This protein controls the movement of salt and water in and out of your body's cells. If the protein is defective it leads to the following:

→ Sodium chloride (salt) cannot leave the cell. Water follows salt, so this means water does not leave the cell either. Without enough water, mucus becomes thick and sticky.

→ The body does not absorb sodium chloride (salt) from sweat glands back into the skin. This is why your child's sweat is more salty.

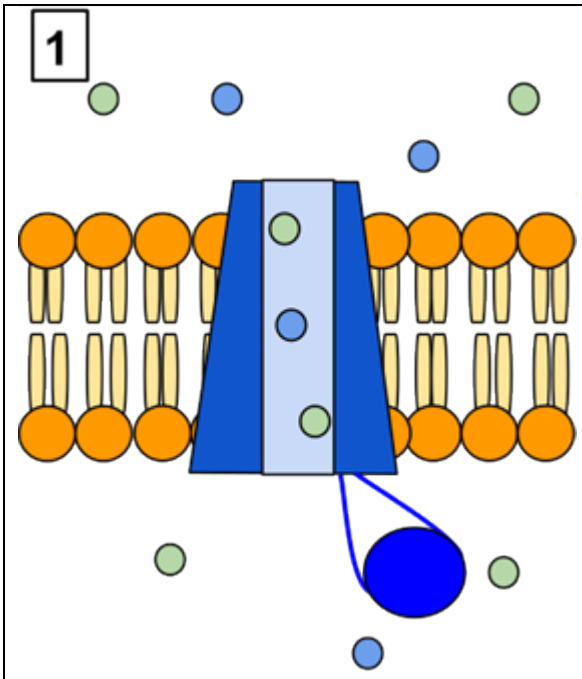
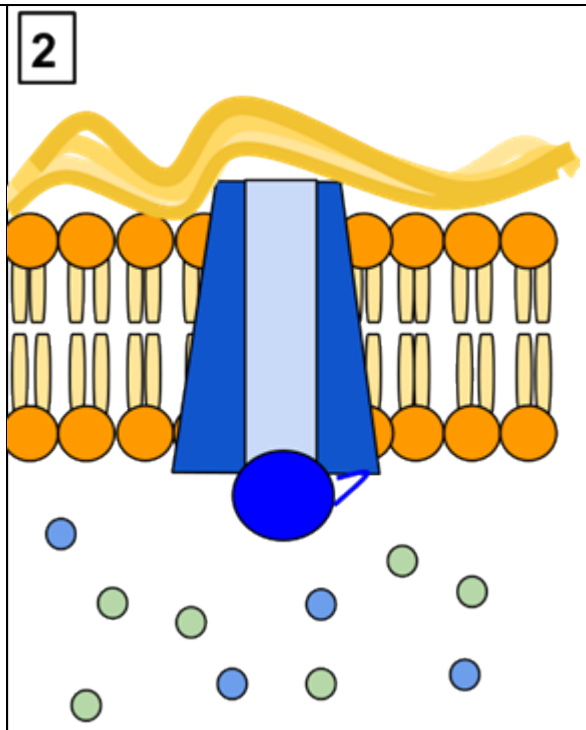
See illustration on next page to learn more about the CFTR protein

What is the treatment for CFTR gene defects?

There are some medications that can be taken by mouth that help the defective proteins work better. Your doctor will let you know if your child qualifies for one of these medications based on genetic mutations, age, and health status.

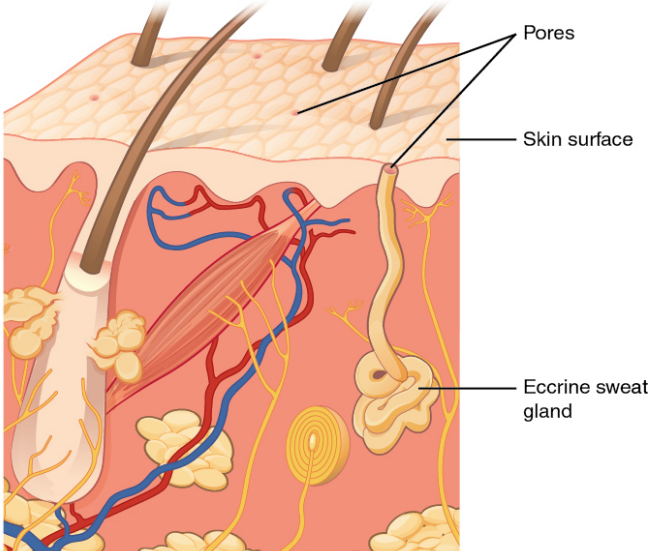
Where can I find more information about CF Genetics?

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, page 11-16
- U-M CF New Patient Binder: *Beginning CF Care,* page 1-4 and Appendix 2-4

	
Panel 1 shows the CFTR protein working correctly, salt and water molecules freely flow in and out of the cells.	Panel 2 shows the defective CFTR protein blocking the channel through which salt and water molecules flow in and out of the cell. This leads to buildup of thick mucus in the lungs.

How does CF affect the sweat glands?

Sweat glands are small tube-like structures of the skin that produce sweat. Sweat is made up of water and salt (combination of sodium and chloride) and helps your body cool down and maintain its temperature. When it's hot the sweat produced in the sweat gland travels to the surface of skin through a duct (or passage). Normally, salt and water is reabsorbed from the sweat as it travels through the sweat duct. This helps to keep your electrolytes balanced.



By OpenStax College [CC BY 3.0] via Wikimedia Commons. Access at: https://commons.wikimedia.org/wiki/File:508_Eccrine_gland.jpg

What happens in CF?

- The defective CFTR protein does not allow salt and water to reenter the cells that line the sweat duct.
- Your child will lose more salt and water than they should. This can cause dehydration and muscle cramps.
- Your child's sweat chloride test would be expected to be abnormal (high) because of the trouble reabsorbing salt.
    - Please note: the severity of CF disease cannot be predicted using the sweat chloride test results.

How to prevent dehydration and muscle cramps?

- Add salt to food and fluids. Your dietitian will help you decide how much.
- Keep fluids close by and add extra salt when it's hot and your child is active or more likely to sweat.

Where can I find more information about CF Sweat Glands?

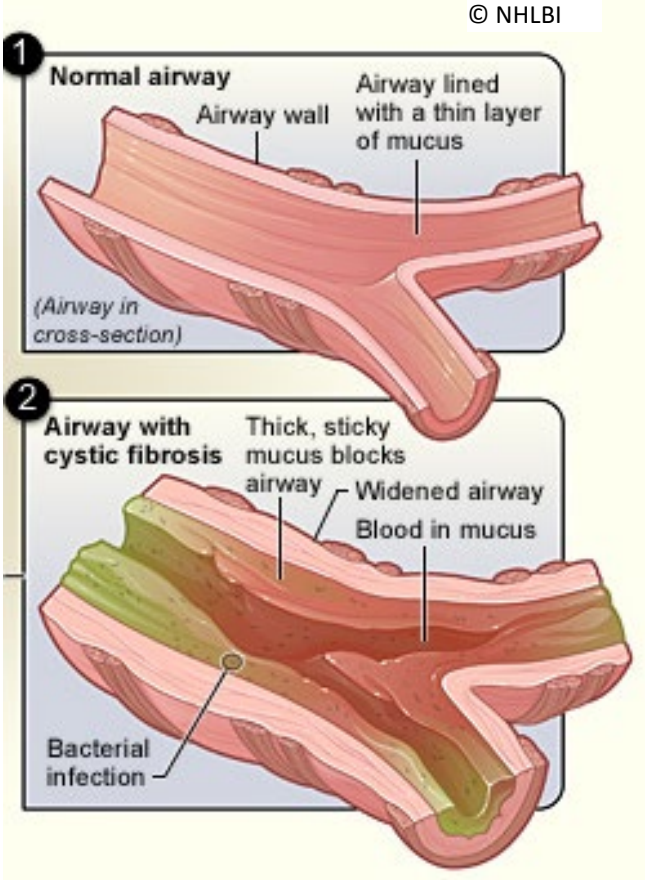
- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, page 35-36
- UM CF New Patient Binder: *Beginning CF Care,* page 15 and Appendix 6
- UM CF New Patient Binder: *Managing Nutrition & Digestive Problems,* Appendix 12

How does CF affect the lungs?

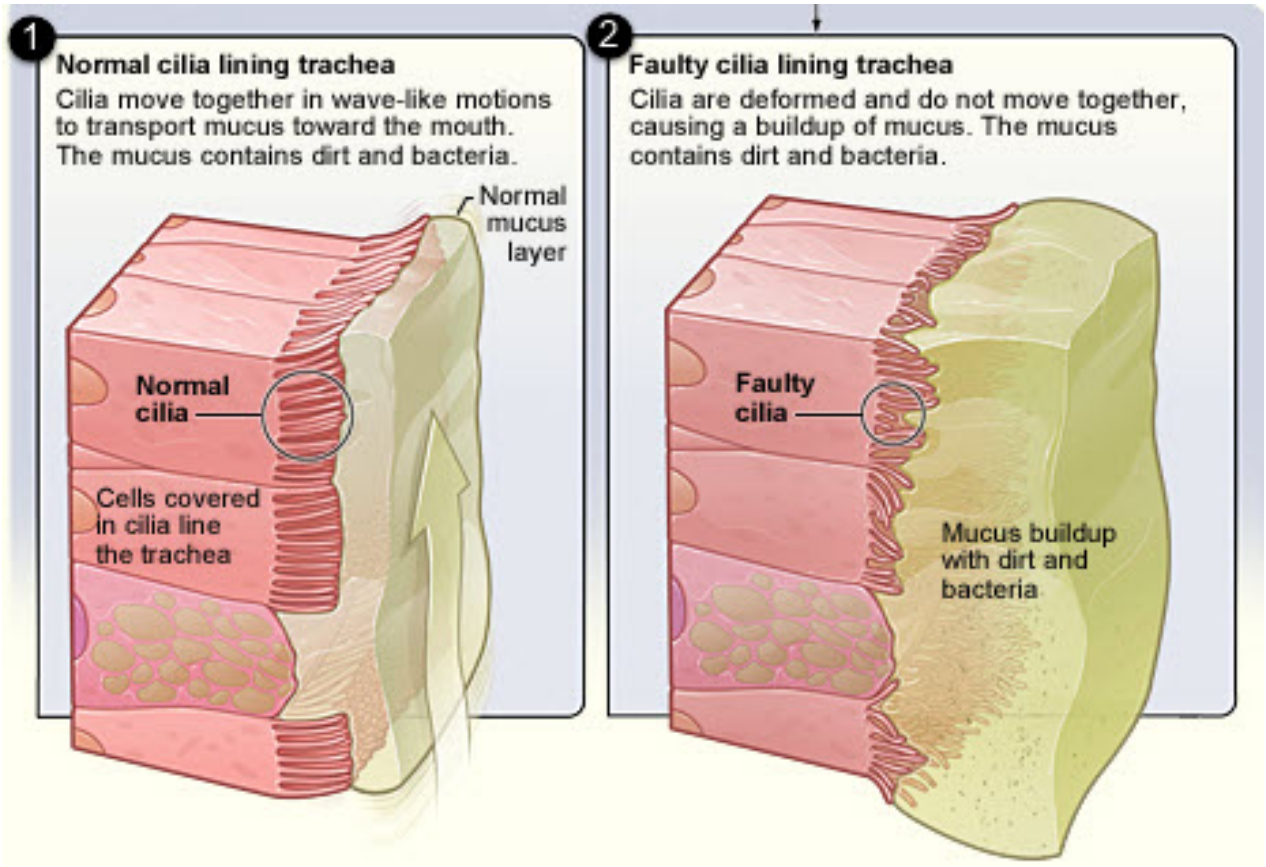
How do lungs work?

- The inside of the lungs is lined with tiny hair-like objects called cilia ("silly-ah"). Cilia move together to sweep tiny amounts of debris and bacteria out of the lungs to be swallowed.
- Mucus is thin and fluid like, being swept along with the debris and bacteria out of the lungs.

What happens in CF lungs?



- The mucus in CF lungs becomes very thick and sticky. This makes the mucus harder to move and clogs up the cilia, making it harder for them to sweep out bacteria.
- The bacteria thrive in the wet, dark, warm sticky mucus and begin to grow, causing infection. The mucus can clog up the airway and this traps air behind the mucus. That trapped air and extra mucus makes the lungs distended (wider) and fibrotic (stiff).
- The body responds by trying to fight the infection, which causes inflammation (swelling). That inflammation can make it harder to move the mucus through the airways, making more bacteria grow in the trapped mucus.
- This cycle of infection, inflammation, lung damage and clogged airways causes the majority of illnesses, lung damage, and lowered life expectancy in CF patients.



- Symptoms of CF lung infection include:
  - Increased cough (wet or dry sounding)
  - Increased sputum (coughing up mucus)
  - A cold that is lasting longer than a couple days
  - Decrease in appetite or energy.

Where to find more information about CF Lungs:

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, pages 41-42
- UM CF New Patient Binder: *Beginning CF Care,* pages 23-35 and Appendix 7-9
- UM CF New Patient Binder: *Managing Lung & Other Respiratory Problems,* pages 2, 16, 19

What is the treatment for CF lungs?

- Move the mucus! Unclog your child's cilia by moving little (or large) amounts of mucus 2 times a day.
    - Open the airway. Give inhaled albuterol first. This is a bronchodilator, which means that it opens the airways to allow mucus to move easier
    - Thin the mucus. Give salt water through a nebulizer. This is generally started when your child is a little older based on symptoms.
    - Break down the mucus. Give a mucolytic through a nebulizer. This is generally started when your child is a little older based on symptoms.
    - Clear the mucus. Do chest Physiotherapy (CPT) 2 times per day when healthy, and 4 times per day when sick. CPT shakes the mucus off of the walls in the lungs, making it easier for your child to clear out the mucus by breathing and coughing.
      Forms of CPT:
        - Manual, or by hand, Percussion and Postural Drainage (P&PD)
        - Vest device, positive pressure device
        - Breathing techniques
        - Aerobic Activity

- Decrease bacteria in the lungs.
    - Monitor bacteria in the airway with throat cultures or sputum (mucus coughed up) cultures every 3 months with your visit.
    - Give your child antibiotics as they need them. Your child will receive antibiotics by mouth, by inhaling, or directly into the bloodstream via and IV (intravenous). Your CF doctor will prescribe these when needed.

- Decrease inflammation in the lungs.
    - Give medications that can help:
        - Steroids by mouth or inhaled
        - Azithromycin by mouth

    - Follow the mucus clearing treatments and antibiotics to keep the bacteria amount low.
- Prevent infection.
    - Wash your hands!! Help your child learn the importance of hand washing.
    - Teach your child to wear a mask in the clinic and hospital hallways.
    - Work with school and daycare on germ spread and cleaning.
    - Make sure everyone in your household gets a flu shot every year.

Where can I find more information about Treatment for CF Lungs?

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, pages 44-50, 53-58, and Chapter 9 (page 101)
- UM CF New Patient Binder: *Beginning CF Care,* pages 23-35 and Appendix 7-9
- UM CF New Patient Binder: *Managing Lung & Other Respiratory Problems,* pages 2-13, 16-18, 24-30, 37-38, and pages 53-58

How does CF affect the pancreas?

How does the pancreas work?

- Your pancreas makes enzymes that break down fat, protein, and carbohydrates in the food you eat.
- Once broken down, your body can absorb the nutrients.
- Your body moves the enzymes from the pancreas into the intestines, just below the stomach.

What happens in a CF pancreas?

- Some enzymes are made, but cannot get out of the pancreas.
- Your child's food does not get broken down like it should, so does not get absorbed either.
- If not absorbed, it exits the body in your child's stool (poop).
    - Fat that is not absorbed causes large stool that is greasy, very smelly, and makes it float in the toilet. More gas is made, which has a foul smell.
    - Some vitamins need fat to be absorbed in order for them to follow. These are vitamins A, D, E, and K, which become low in CF patients.
    - Protein is needed to build muscles and body structures, and make antibodies to fight infections. When protein does not get absorbed children get very thin and their growth slows down or stops regardless of how much they eat.
        - CF people with good growth have been shown to have better lung function.

Is there any way to know if the pancreas is working?

- Yes! We can test for Pancreatic Elastase, an enzyme that should be found in stool when the pancreas is working. A higher number (more than 200) means the pancreas is making the enzyme and moving it to your child's intestines.
- You would collect a stool sample at home or in the office for this test.

What if the stool test is normal?

- That means the pancreas is working for now.
- Over time, the pancreas can start to get damaged or clogged, so we will check stool test every year.

What if the stool test is not normal?

- That means the pancreas is not working as well as it should.
- We do not need to check the stool test every year, because we know the pancreas is not working.
- We treat your child!

What is the treatment for a CF pancreas?

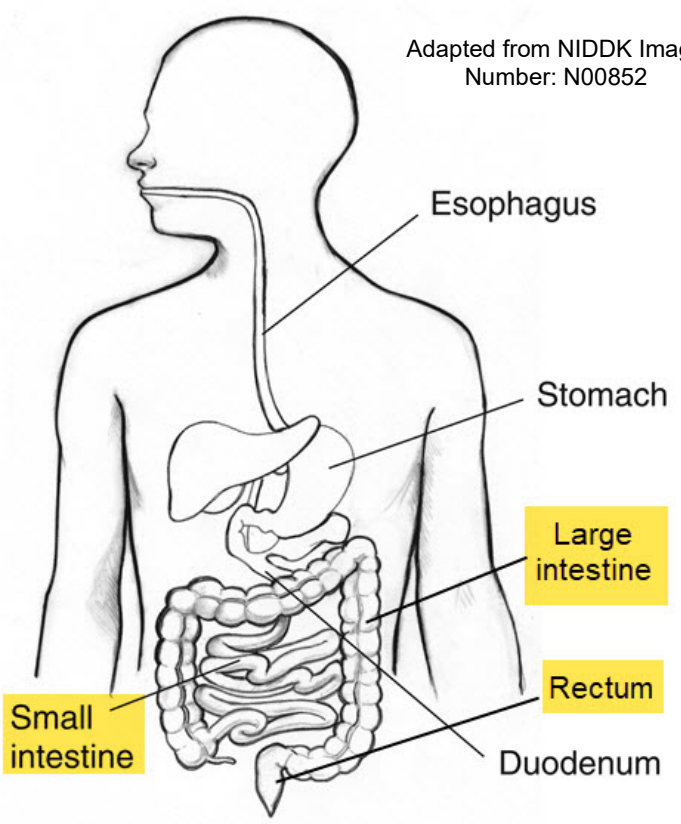
- Your child will take pancreas enzymes in pill form with every meal.
    - For infants and small children who cannot swallow pills, you will open the capsule and sprinkle the little medicine beads onto a small amount of applesauce. Give it immediately.
    - Your CF dietitian will teach you how to do this, and how to keep track of how your child is growing and absorbing nutrients.
- Your CF dietitian will see you in our clinic often to make changes to the amount of enzymes you will give. This helps to better absorb nutrients.
- Extra Calories! You will give your kid a high calorie diet to be sure they have enough food to absorb. CF kids require more calories than kids without CF.
- CF Multivitamins are designed with higher amounts of the vitamins A, D, E, and K. Your child will take this CF multivitamin and your CF doctor and dietitian will monitor vitamin levels at least yearly.

Where can I find more information about CF Pancreas:

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, pages 61-66
- UM CF New Patient Binder: *Beginning CF Care,* pages 20-22 and Appendix 7
- UM CF New Patient Binder: *Managing Nutrition & Digestive Problems,* pages 2, 7, 9-15, 19, 23-24, 28-32

How does CF effect the intestines?

The intestines are a long, continuous tube running from the stomach to the anus. Most absorption of nutrients and water happen in the intestines. The intestines include the small intestine (also called the small bowel), large intestine (also called the large bowel or colon), and rectum.



How do the intestines work?

The walls of your intestines are filled with mucus glands. These glands expel mucus, which helps make the inside of the intestines slippery. That mucus moves the stool through the intestines.

What happens in CF intestines?

- The mucus glands are still there, but the mucus gets very thick and sticky.
- Sticky mucus makes stool move slower through the intestines and can make the stool a little larger than normal, even if your child takes CF Enzyme pills.
- The large, slow moving stool can also cause constipation, or difficulty to fully empty the intestines of stool.
    - Symptoms of constipation include: abdominal pain, cramping, bloating
- The stool starts in the small intestines and moves to the large intestines through a valve called ileocecal valve. It can be difficult for large stool to pass through this valve, so it may clog up and allow only a little or nothing to pass through. If not treated this can cause an obstruction (Distal Intestinal Obstruction Syndrome or DIOS).

- Symptoms of DOIS include: severe abdominal pain, vomiting, cramping, bloating, and harder stools that happen less often than normal.
    - Meconium Ileus is an obstruction in the intestines of the first stools your baby has, when the stool is still meconium.

What is the treatment for CF intestines that get clogged with stool?

- Your CF dietitian will assess stomach symptoms and stool when you are seen in the clinic or over the phone if needed.
- Your doctor will prescribe enzymes if needed. Make sure to give your child brand Name enzymes that have not expired.
- If stools become difficult to pass, you can do some preventative treatment at home. Increase fluids and give an over the counter gentle stool softener called Miralax. If your child still has constipation, call your CF doctor, since it may lead to developing DIOS if not treated.
- In order to treat DIOS your child will be admitted to the hospital for fluids and a stool "cleanout" with medications.

Where can I find more information about CF intestines:

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, pages 66-67
- UM CF New Patient Binder: *Beginning CF Care,* pages 17-19 and Appendix 7
- UM CF New Patient Binder: *Managing Nutrition & Digestive Problems,* Appendix 2-

How does CF affect life expectancy?

What is the life expectancy of someone with CF?

- In general, the average life expectancy for someone with CF is shorter than the general population. There are many factors that influence this:
  - The specific genetic mutations
  - The bacteria that infect the lung of CF people, how their body responds to it and how much they comply with treatments
  - Other aspects of the disease, some of which we know little about (for example, other channels that influence the salt and water movement and may help protect the lung)
- As of 2019 the median life expectancy for someone with CF is 47 years; this means that half the CF people born 47 years ago are still alive today.
- The median survival has been steadily improving in the last 20 years and we expect that it will continue to increase due to advances in research and treatments.
- For babies born today, we expect their median life expectancy to be the same as in people without CF. This is why we work very hard with you to make sure people with CF do well as they get older.

Where can I find more information about CF Life Expectancy:

- UM CF New Patient Binder: *Beginning CF Care*, page 4

What are the key points to remember?

Remember our goals:

- Slow the progression of lung disease
- Prevent complications
- Keep your child healthy

How can I help my child achieve those goals?

- Bring your child to regular visits with your CF care team
    - Visits are monthly until 12 months of age, then every 3 months after 12 months of age
    - Addressing symptoms or changes early affects long term lung damage
    - Small changes early in the onset of symptoms are easier to do and more effective than doing big changes late into the onset of symptoms.
- Call between visits when your child is sick to talk with a CF nurse
- Provide good structure and guidance for your child regarding CF and treatments. They follow your lead, so help your child learn these valuable skills to help them manage their CF as adults.

What long-term goals can I plan on for my child?

Do not change the dream you have for your child because of CF!

- Expect to see them graduate to an adult CF center after high school or college.
- Expect your child to learn to start caring for their CF independently as they transition to adulthood with the help of the adult CF center.
- Set school, sports, career, and life goals together with your child, and expect that they will achieve them.

Where can I get more information or get involved with the CF community?

- **CF Foundation** (www.cff.org) this organization is involved with many activities:
    - Advocacy and fundraising to advance CF support research
    - Providing accurate CF information and education

    - Maintaining the CF Registry, which tracks data on each person with CF (this is how we get all those statistics on CF people)
- **Research**: If interested, our CF Center performs many research studies for CF and will contact you regarding studies your child qualifies for.

Will I have any help?

- YES! Your CF care team is here to help you through every step of your child's life. We encourage questions and will help you learn to manage CF.
- We are available for questions Mon-Friday 8:00-5:00 p.m. by phone, patient online portal can be used 24/7, and our on-call is available 24/7 for urgent needs as well.

Where can I find more information about CF Care Centers and the CF Foundation:

- CFF book, *An Introduction to Cystic Fibrosis: For Patients and Families*, page 88, Chapter 10 (page 115), Chapter 11 (page 125), Chapter 13 (page 135), Chapter 14 (page 143), Chapter 15 (page 151)
- UM CF New Patient Binder: *Beginning CF Care,* page 5
- UM CF New Patient Binder: *Becoming a CF Manger,* pages 12, 14, 20-24, 32
- UM CF New Patient Binder: *Working with your Child*, page 2-5