

THE PEDIATRIC JOURNEY



Introducing the New Cystic Fibrosis Clinic at El Paso Children's Hospital

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PEDIATRIC PULMONOLOGIST



We are excited to announce the opening of the new Cystic Fibrosis (CF) Clinic at El Paso Children's Hospital. This clinic is dedicated to providing comprehensive, multidisciplinary care to children with CF.

What is Cystic Fibrosis?

Cystic fibrosis (CF) is an autosomal recessive genetic disorder that affects the respiratory, digestive, and reproductive systems. It is caused by a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which is responsible for producing a protein that regulates the movement of chloride and bicarbonate, as well as downregulating the epithelial sodium channel (ENaC).

The prevalence of CF is approximately 1 in 2,500 to 3,500 live births in Caucasians. In Hispanics, the prevalence is 1 in 7,500 to 10,000. CF prevalence is lower in other populations, such as African Americans (1 in 17,000 live births) and Asians (1 in 31,000 live births).

In people with CF, the CFTR protein is either missing or does not work properly, leading to a buildup of thick mucus in the lungs. This mucus can obstruct airways and create an environment that is conducive to infection and inflammation, allowing damage and scarring of the lungs over time. CF can also affect other organ systems. In the digestive system, CF causes problems with nutrient absorption and digestion such as pancreatitis. CF affects the endocrine system by placing patients at risk for developing CFTR-related diabetes and can affect the reproductive system by leading to fertility issues.

The symptoms of CF can vary depending on the severity of the disease and the organs affected. Common symptoms include:

- Persistent coughing, wheezing, or shortness of breath
- Frequent lung infections, such as pneumonia or bronchitis
- Difficulty breathing or exercising
- Thick, sticky mucus production
- Digestive problems, such as poor weight gain, abdominal pain, and diarrhea
- Fertility problems

Traditionally, the treatment for CF consists of managing the symptoms and slowing the progression of the disease. Treatment typically involves a combination of inhaled medications like short acting beta agonists, hypertonic saline, dornase alpha and antibiotics, airway clearance techniques, nutritional support, and other supportive therapies.

However, a new and exciting therapy is dramatically improving the life expectancy of patients with CF. CFTR modulators have shown to improve lung function, reduce the frequency of pulmonary exacerbations, and improve quality of life in people with CF. However, not all people with CF are eligible for CFTR modulator therapy, as it depends on the specific CFTR mutation they have. It is important to discuss treatment options with a healthcare provider who specializes in CF.

How to Diagnose Cystic Fibrosis

The diagnosis of cystic fibrosis typically involves a combination of clinical evaluation, genetic testing, and laboratory testing. Here is an overview of the diagnostic process for CF:

1. Clinical Evaluation:

- Infants with positive CF newborn screening results
- Infants with symptoms suggestive of CF (e.g. chronic cough, recurrent respiratory infections, failure to thrive, or meconium ileus)
- Older children and adults with symptoms suggestive of CF (e.g., male infertility, chronic respiratory infections, or chronic sinusitis)
- Siblings of a patient with confirmed CF, if the diagnosis cannot be established based on genetic testing



CEO MESSAGE

What a great time to be a part of the El Paso Children's Hospital community! Over the past 11 years, El Paso Children's Hospital has grown in its expertise and capabilities with the development of several pediatric multi-specialty clinics, including one of our newest, the Cystic Fibrosis Clinic. This specialty clinic is led by Clinic Director Dr. Endy A. Dominguez Silveyra and Co-Director Dr. Denease Francis, along with an outstanding, dedicated multidisciplinary team.

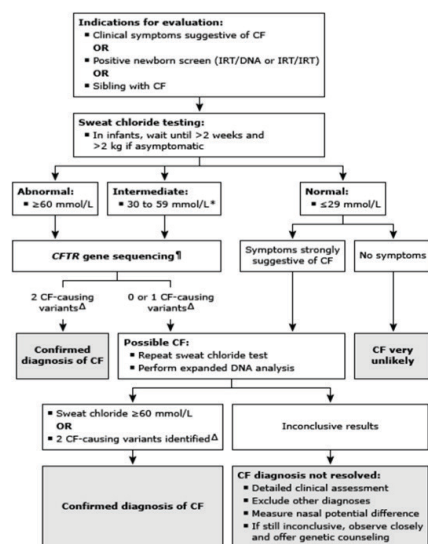
This clinic marks our 14th specialty clinic to better serve the community. Our continuous growth, along with the commitment to our HEART values, ensures a bright future for El Paso Children's Hospital and the entire region. Thank you for always going above and beyond to deliver the highest quality care to our patients.

2. Sweat Chloride Testing:

Sweat chloride concentration	Result	Interpretation
<29 mmol/L	Normal	CF very unlikely
30 to 59 mmol/L	Intermediate	Possible CF, additional testing recommended*
≥60 mmol/L	Abnormal	Diagnosis of CF, if confirmed by a second test and if clinical symptoms consistent with CF are present

3. Genetic Testing: CFTR sequencing should be performed in individuals with high sweat chloride test results and any uncertainty in the diagnosis, including:

- Patients with intermediate sweat chloride results (in addition to repeat sweat chloride testing)
- Patients with confirmed or suspected CF, if the genotype is not yet known. In these patients, the gene sequencing confirms the diagnosis, and the specific CFTR mutation has important implications for treatment and prognosis
- Patients with normal sweat chloride results with a strong clinical suspicion for CF



CF, cystic fibrosis; IRT, immunoreactive trypsinogen; DNA, genetic screen for pathogenic CF transmembrane conductance regulator (CFTR) gene variants.
* For asymptomatic infants with intermediate sweat chloride results, sweat chloride testing should be repeated at 1 to 2 months of age.
† For infants with sweat chloride ≥60 mmol/L, CFTR sequencing is not mandatory if the newborn screen included DNA testing and definitively identified 2 CF-causing variants.
Δ "CF-causing variants" denotes variants in the CFTR gene that are known to cause disease, using classifications identified in the CFTR project.

What Does the Cystic Fibrosis Clinic at El Paso Children's Hospital Offer?

CF is a complex and challenging disease that requires specialized care and management. The new Cystic Fibrosis Clinic at El Paso Children's Hospital is staffed by a team of experts who are dedicated to providing the best possible care and support for patients and their families. Our team includes pediatric pulmonologists, pediatric gastroenterologists, pediatric endocrinologists, pediatric pathologists, dietitians, respiratory therapists, social workers, physical and occupational therapists, and other healthcare professionals who have extensive experience in the management of CF.

At the CF clinic, patients will have access to a wide range of services and treatments, including:

- Comprehensive evaluation and diagnosis (sweat chloride test)
- Customized treatment plans tailored to the individual patient's needs
- Medication management and monitoring
- Nutritional assessment and support
- Pulmonary function testing and other diagnostic procedures
- Airway clearance techniques and breathing exercises
- Social and emotional support for patients and their families
- Coordination of care with other healthcare providers

Our team takes a multidisciplinary approach to care, working closely with patients and their families to develop individualized treatment plans that address their unique needs and goals. We also work closely with community pediatricians

and other healthcare providers to ensure that our patients receive the best possible care and support.

We understand that living with CF can be challenging, and we are committed to providing our patients with the best possible care and support. The new Cystic Fibrosis Clinic at El Paso Children's Hospital is a testament to our commitment to improving the lives of people with CF and their families.

How Can You Access the Cystic Fibrosis Clinic at El Paso Children's Hospital?

If you have a patient who you think may benefit from the services offered, please don't hesitate to refer them to us. The CF clinic will work with each patient to develop a personalized treatment plan based on their unique needs and circumstances.

Conclusion

The establishment of a new cystic fibrosis clinic is an exciting development for the CF community in El Paso, Texas and the borderland region. By providing specialized care, the clinic has the potential to greatly improve patient outcomes and quality of life. Currently we are working to achieve certification through the Cystic Fibrosis Foundation to become an accredited CF Care Center. We look forward to seeing the positive impact that this clinic will have on the CF community in the years to come.



PICKY EATERS

Brianna Garza, MD
PEDIATRICS

Picky eating can be a source of anxiety for parents and children alike. With so much guidance from peers and social media, it can be hard to filter out which advice is appropriate for you and your family. The good news is that the best advice is sometimes the most simple! Here are a few tips:

1. Try to be consistent with dinner times and make sure that the whole family is involved with minimal distractions - no TV or cell phones. This is a great time to model healthy eating habits with your child by taking a bite of that vegetable, too! Plus, when we are less distracted and more thoughtful about eating, it's less likely that we will overeat.
2. Get kids involved with what shows up on their plate! As we head into spring, it is an opportune time to start a vegetable garden. Also, give your child a chance to help prepare some of the food in the kitchen. Kids who are involved with that vegetable, from seed to dinner plate, are more likely to try it when given the opportunity!
3. Don't make trying new foods so high stakes. We definitely want kids to try new things, but let them know that trying and figuring out what they like is the most important part. Praise them for trying and encourage them to try it again, possibly prepared in a different way the next time.
4. Give kids options, but not too many. When preparing dinner, give kids 2 acceptable options to choose from. The main course can be decided by you, but you can give your child a few choices for a side dish. For example: "We are having chicken tonight. Should we have peas or broccoli with that?"

I hope this helps guide your family to healthier eating habits and a more productive dinner time.



GASTRO INTESTINAL MANIFESTATIONS IN CYSTIC FIBROSIS

Denease Francis, MD
PEDIATRIC GASTROENTEROLOGY, HEPATOLOGY, AND NUTRITION

Cystic fibrosis (CF) is an autosomal recessive disorder caused by mutations in the cystic fibrosis transmembrane regulator (CFTR). While primarily a pulmonary disease, cystic fibrosis affects multiple organ systems including the gastrointestinal (GI) system. This can manifest in several ways including gastroesophageal reflux disease, meconium ileus, distal intestinal obstruction syndrome, and CF-related liver disease. The most commonly affected organ in the GI system is the pancreas.

Normally acinar cells secrete proteins, digestive enzymes, into the pancreatic duct lumen. CFTR is located on the apical surface of the ductal cells and permits the movement of anions and fluid into the duct membrane which allows these digestive enzymes to remain soluble in an alkaline solution. In cystic fibrosis, the secretion of chloride, sodium, and bicarbonate into the duct lumen is impaired however; the acinar cells continue to secrete proteins leading to a hyperconcentrated acidic fluid in the duct lumen. This viscous fluid obstructs the ducts and acini leading to epithelial injury, inflammatory changes, destruction of the acini, and eventually fibrosis. This process begins in utero and progresses after birth. Approximately 85-90% of patients with cystic fibrosis will be pancreatic insufficient with 60% being insufficient at the time of diagnosis. Up to 30% of patients who are pancreatic sufficient initially will eventually progress to insufficiency.

Exocrine pancreatic insufficiency (EPI) is closely related to CF genotype with patients having milder mutations more likely to be pancreatic sufficient whereas those with more severe mutations will be insufficient. It is thought to be irreversible though there is some evidence to suggest that use of CFTR modulators early in diagnosis may prevent progression to or even reverse EPI. EPI occurs when pancreatic colipase/lipase secretion is less than 1-2% of normal levels. It results in maldigestion and malabsorption of carbohydrates, fats, and proteins. Clinically these patients will present

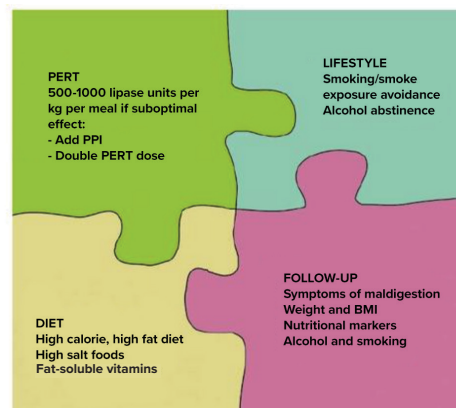
with poor weight gain, growth failure, abdominal bloating, steatorrhea, and edema. They may also have evidence of fat-soluble vitamin (A, D, E, and K) deficiency. In patients with CF, exocrine pancreatic function is assessed at diagnosis and yearly.

The most commonly used method for diagnosis of EPI is the fecal elastase. In CF patients with severe insufficiency (elastase $<100 \mu\text{g}$), fecal elastase has a sensitivity of 100%. For patients with mild insufficiency who do not require enzymes (elastase = $100 - 200 \mu\text{g}$) its utility is more limited. Elastase is not affected by use of pancreatic enzyme replacement therapy (PERT) though watery stools may produce false negative results. Another method of assessing for EPI is the coefficient of fat absorption/excretion though this is not commonly done due to its tedious nature. Patients are placed on a high fat diet for 5 days with stool collected over the last 3 days. A detailed diet record is maintained during the test which is used to calculate the amount of fat consumed. The amount of fat in the stool is then calculated. For infants, fecal fat $>15\%$ of intake is considered abnormal. For adults, fecal fat $>7\%$ of intake is abnormal. The coefficient of fat excretion will indicate the presence of fat in the stool only but not the etiology.

The mainstay for management of EPI is use of pancreatic enzyme replacement therapy. Derived from porcine pancreas, it contains variable amounts of lipase, protease, and amylase. Capsules are available in various formulas and dosing is based on the lipase units. Generally, dosing starts at 1000 lipase units/kg/meal in patients under age 4 and 500 lipase units/kg/meal in older children and adults. Recommended maximum dose is 2500 lipase units/kg/meal or 10000 lipase units/kg. Some patients may require higher dosing though caution should be used as excessive enzyme use has been associated with fibrosing colonopathy. Enzymes are deactivated in an acid environment thus acid suppression with

proton pump inhibitors may be used to improve enzyme effectiveness in patients with continued steatorrhea despite adequate dosing. Enzymes are administered at the beginning of a meal. Relizorb is a newly available delivery system for CF patients with EPI who receive supplemental nutrition via a gastrostomy tube. It consists of a cartridge filled with beads containing lipase which is connected to the feeding line. Use of Relizorb does not replace PERT but is rather used in conjunction. It's important to note that in addition to PERT, CF patients with EPI benefit from adequate nutritional support including supplements and vitamins (A, D, E, and K).

In summary, patients with cystic fibrosis are likely to have exocrine pancreatic insufficiency. Testing for EPI should be done at diagnosis and yearly. Management of EPI is best achieved via a multidisciplinary approach including a pulmonologist, gastroenterologists, dietitians, and social workers.



Lindkvist B. Diagnosis and treatment of pancreatic exocrine insufficiency. *World Journal of Gastroenterology*. 2013 Nov;19(42):7258-7266. DOI: 10.3748/wjg.v19.i42.7258. PMID: 24259956; PMCID: PMC3831207.



CMO CORNER

Jeffrey Schuster, MD
CHIEF MEDICAL OFFICER AT EL PASO CHILDREN'S HOSPITAL

PATIENT RIGHTS AND RESPONSIBILITIES

It is important that health care providers remember that all of our patients have rights. In our hospital, that usually means the parents have the right to be an active participant in the care of the patient. Parents have the right to accept, partially accept, or reject a treatment plan. They are able to change providers as they wish and even discharge themselves from the hospital. If a situation becomes concerning, please obtain help from the Quality or Compliance teams.

Families must be able to comprehend the treatment plan for the patient. We have certified translators for Spanish, and we can use telecommunications for other languages. Families can waive their right to a certified translator if they feel that the communication with the provider is sufficient.

Patients or parents also have responsibilities. They must be active participants in the care of the patient and provide all the information they have about the patient's history. This could include current medications, alternative medications, non-traditional therapies, and vitamins.

Patients and families must also treat all hospital staff, other patients, and visitors with courtesy and respect. They must obey hospital rules and safety regulations. The patient and family are responsible for the outcome if they do not follow the care, treatment, and services plan for the patient.

TTUHSC Department of Pediatrics Grand Rounds

4/5/2023 • Dr. Jonathan Chiao • Peds ENT

4/19/2023 • Dr. Nalinda Charnsangavej

5/3/2023 • Dr. Gilbert Handal

5/17/2023 • Dr. Karen Skjei • Peds Neuro

6/7/2023 • Dr. Harry Wilson • History of Medicine

6/21/2023 • Dr. Abraham Lopez • Neuropsychologist

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