Focused Clinical Multi-Disciplinary ISP Proposal
Cystic Fibrosis

Description:

Cystic fibrosis is a complex inherited disease that affects tens of thousands of people in the United States. In the past, cystic fibrosis (CF) was known as a fatal disease of childhood, but medical advances have significantly improved the life expectancy and the quality of life of CF patients. Now, many people with CF live well into their adulthood, and some even into their sixties. Proper treatment and management of CF can really make a difference in a patient’s life. Therefore, medical education and research regarding CF is a vital and relevant pursuit.

Cystic fibrosis is truly a complex, multi-system disease. The Cystic Fibrosis Transmembrane Regulator (CFTR) protein that is affected by the genetic mutation is involved in many different parts of the body. The CFTR protein codes for a channel in the cell membrane that allows chloride to leave epithelial cells. The pathophysiology linking CFTR mutations to clinical CF is incompletely understood. A prominent theory suggests, however, that when the CFTR does not function properly, not enough chloride leaves the cell, and too much sodium is absorbed. Thus, water is not absorbed normally, creating a thick mucus. This mucus plugs ducts and tubes in organs that express the CFTR protein, causing a number of medical problems. CF affects the respiratory system, the digestive system, the sweat glands, and the reproductive organs. Therefore, care for the CF patient must be multidisciplinary, involving the attention of several medical specialties such as pulmonologists, gastroenterologists, geneticists, and other specialists like endocrinologists, fertility specialists, and gynecologists. Care for the CF patient requires a multidisciplinary team, consisting of physicians, nurses, nutritionists/dieticians, physical/respiratory therapists, and social workers. All of these people play a significant role in the well being of a cystic fibrosis patient.

Pulmonologists are often the most involved physicians in the care of CF patients, since 85% of CF patients will die from respiratory failure. The challenge that pulmonologists face is that lung disease in CF is progressive. Infections (often with resistant bacteria and fungi) and inflammation take their toll on the airway and can lead to weeks of hospitalization and debilitating pulmonary exacerbations. It is the job of the pulmonologist to ensure that the patients are on the most effective antibiotic regimen as well as on the appropriate prophylaxis. They also have to treat complications such as bronchiectasis, pneumothorax, and hemoptysis. Respiratory failure is the most common cause of CF-related death in patients. Lung transplants may be an option for patient whose lung function has deteriorated to this point. This introduces a whole new set of medical issues.

Gastroenterologists often treat patients for symptoms associated with pancreatic enzyme insufficiency, such as malnutrition, malabsorption, or abdominal pain. They also
face more serious problems such as meconium ileus, distal intestinal obstructive syndrome, rectal prolapse, and liver disease. They provide care by treating infections and obstructions, maintaining pancreatic enzyme doses, and preventing malnutrition.

As cystic fibrosis patients get older, the roles of reproductive endocrinologists, fertility specialists, and OB/Gyn doctors increases. Males are most often infertile and females are less likely to become pregnant. There is a great deal of counseling involved in this area of patient care. In addition to genetic counseling, patients may choose to go through in-vitro fertilization or other assisted reproductive therapies.

Cystic fibrosis patients have other medical problems such as sinusitis and nasal polyps. Many patients develop diabetes at a young age, possibly as a consequence of pancreatic damage.

Genetics also plays an important role in CF. Since CF is an autosomal recessive genetic disorder, genetic counseling and genetic testing have become very important in families with CF. There are over 1,000 different CF mutations identified, with more than 87 commonly testable mutations. Different genotypes are also related to varying levels of disease. Applied genetics has become quite important in CF. Carrier screening and prenatal diagnosis (amniocentesis, chorionic villus sampling, and fetal ultrasound screening) are now available, as well as preimplantation genetic diagnosis. Gene therapy is also a hope for future disease treatment.

In addition to the physicians, the CF team is an immense wealth of support and knowledge for the patients and their families. Respiratory and physical therapists play a key role in maintaining lung function in CF patients. Patients get regular pulmonary function tests, chest physical therapy, bronchial drainage, and aerosol treatments. Therapists can also help implement an exercise program, as well as teach the families how to give treatments at home. Nutritionists and dieticians educate patients and families on the importance of nutrition in a CF patient. They supply patients with information on high caloric supplements, correct use of enzymes, and supplemental vitamins. Social workers help patients and their families deal with the social, psychological, financial, and emotional burdens of CF. They are often intricately involved in the lives of CF patients and can help with everything from parental support to the psychological and emotional issues of end of life care. Nurses are often the link between all the other disciplines. They coordinate with families and can be a great source of education and support.

Cystic fibrosis is further complicated in that treatment greatly differs as the patient grows older. An adult with CF does not have the same medical problems or concerns as an eight year old or an infant with CF. Therefore, the care of cystic fibrosis patients requires a varied approach depending on whether the patient is an adult or a child.

**Definition:**

Cystic fibrosis is an ideal disease to study for the FCM-ISP because it encompasses many different disciplines as described above. My overall goal for this project is to become an expert in caring for a patient with cystic fibrosis. As a future pediatrician, having an enhanced knowledge about cystic fibrosis will not only benefit my practice, but also my patients.

Specifically, I have several objectives for this project:
1. Learn about the management and treatment of medical problems and complications faced by CF patients, mostly focusing on the respiratory and gastrointestinal systems.
2. Become well acquainted with diagnostic procedures as well as genetic testing and genetic counseling.
3. Work closely with the CF Team and learn about how their roles change as the patients grow older and their disease progresses.
4. More specifically, I would like to learn how to perform and analyze pulmonary function tests, do chest physical therapy, understand the basics of respiratory therapy, determine nutritional requirements for a patient, and how to properly prevent malnutrition.
5. Get involved with the social work and psychological aspects of CF.
6. Understand what it means to have CF. Learn about the questions and concerns of patients as well as their fears.

Though I expect to meet all of these goals, I am sure that my goals and expectations will change and become more focused as I acquire more knowledge. I plan to be flexible throughout the following year, in order to be able to meet any new goals.

**Plan and Methods:**

I will be attending the Pediatric Cystic Fibrosis Clinic and the Adult Cystic Fibrosis Clinic regularly throughout my designated ISP months. I will also attend these clinics whenever possible throughout my fourth year. The Pediatric CF Clinic occurs twice a week at Children’s Hospital on Tuesday and Thursday mornings and is run by Dr. Pian. The Adult CF Clinic is held Wednesday evenings at Perlman Clinic and is run by Dr. Conrad. In both clinics, I will not only be working with the physicians, but I will also be working with the respiratory therapists, dieticians, social workers, and nursing staff that specialize in the care of CF patients. I will learn how to assess the pulmonary function of the patients, determine dietary and social needs of the patients, as well as how to medically manage and treat CF patients on an outpatient basis.

I will also follow hospitalized CF patients with Dr. Pian, Dr. Conrad, and Dr. Dohil throughout my two designated ISP months and whenever possible during my fourth year. This will give me exposure to the acute care of a CF patient in distress. Inpatient management of a struggling CF patient varies greatly from outpatient management of a relatively healthy CF patient. I will also be working with the dieticians, social workers, physical therapists, and respiratory therapists in the hospital.

Additionally, Dr. Pian, Dr. Conrad, and Dr. Dohil will select between 5-10 of their own cystic fibrosis patients from different age groups, in varying stages of the disease, who consent to work with me. Throughout my fourth year, I will work closely with these patients and their families. I will go to their doctor appointments with them. If they are hospitalized, I will follow them throughout their hospital stay. I will learn as much from their experiences as possible. Therefore, I will get a better sense for what continuity of care means for a cystic fibrosis patient. This will also give me a chance to become close with patients who have CF and to better understand what it is like to be a CF patient,
whether he is a child hospitalized with Pseudomonas or young adult awaiting a lung transplant.

In the two months that I will strictly dedicate to the ISP, I will also do literature research to learn about the latest developments with cystic fibrosis – ranging from gene therapy to lung transplantation. I will also work closely with the adult CF social worker Amy Brown at Perlman Clinic, who is involved with all of the non-medical issues that CF patients have to face. I plan to arrange to visit both a genetics clinic where genetic testing is done, and a hospice to experience end of life care for CF patients.

I am in the process of getting Human Subjects Approval for this project, in order to be able to be involved with patients in the manner described above.

**Synthesis:**

My goal for this project is to become proficient in the overall care of CF patients, not just one aspect of their care. Therefore, I will combine all of the knowledge that I gain from each of the disciples and apply it, every time I work with a patient who has CF.

**Summary:**

Upon completion of the project, I will prepare a written summary of my experiences throughout the year. There will be two parts to this summary. The first section will contain an overview of all of the medical and clinical knowledge that I have gained throughout the year from going to clinics, rounding on inpatients, following specific patients over time, and doing literature reviews. It will represent the knowledge that I will carry into my residency/career as to how to treat and manage a CF patient from all of the different aspects described above. The second half of the proposal will focus on what it means to be involved in the care of a CF patient. There is a great amount of emotional investment required by all those who care for CF patients, which clearly includes their families. This portion of the summary will include personal experiences with patients, their families, and staff.

**Evaluation:**

Besides the final written summary, Dr. Pian, Dr. Conrad, and Dr. Dohil will evaluate the success of the project based on my participation and performance over the course of the year. They will help guide me towards my goals throughout my learning process. Since they are all experts in cystic fibrosis, they will be able to judge if I have met these goals and if I have learned enough to be able to care for a CF patient and their family in my future career as a pediatrician. Their evaluations will be included with my final summary and turned into the ISP Committee.
References:

