Gillian was visiting the pediatrician with her 9-month old son Max who was suffering from a reoccurrence of coughing. Since his birth Max had had several colds, one almost monthly. One 'cold' had been so severe he was diagnosed with pneumonia and hospitalized for 5 days. The pediatrician at this point was growing concerned. Max did not attend daycare and had no siblings at home. His exposure to contagions was limited yet he continued to get severe respiratory infections. He also was not thriving as he should be. His mother reported that he had an excellent appetite but he was not gaining weight as expected. During the visit Max had a bowel movement and the doctor noted that his stools appeared greasy. The doctor asked his mother if Max's skin ever tasted salty. Gillian said it did, but she assumed that was normal. They live in a warm climate and she assumed Max was just sweating a lot. The doctor checked Max's records and noted that Max was born at home and no newborn screening was done. He performed the newborn blood spot test (Guthrie Card) which identifies 7 possible genetic defects. The test came back later that week indicating that Max potentially had cystic fibrosis. The pediatrician scheduled Max for a sweat test at the regional medical center which confirmed the preliminary diagnosis of cystic fibrosis.

Cystic fibrosis (CF) is one more common inherited genetic syndromes with 1 baby in every 2-3000 live births having CF. It is an autosomal recessive trait, meaning that an individual would have to inherit the defective CF gene from both parents to have the disease. There are almost 2,000 known genetic mutations in the gene, however diagnostic tests only identify the most common forms. Most individuals with CF are diagnosed by age 2. There are about 30,000 people living with CF in the United States today. While many CF sufferers live into their 50s and 60s the median age for survival is only 33.4 years with patients succumbing to chronic lung infections.

Patients with CF suffer from chronic respiratory infections and have impaired digestive absorption because of thick mucus accumulation in these tissues. How does this mutation cause so many problems and what does it have to do with cell structure? CF mutations affect the folding of a protein called the cystic fibrosis transmembrane conductance regulator (CFTR). The normal version of this protein is embedded in the plasma membrane of cells and acts as a chloride channel, i.e., it allows chloride to pass out of the cell. When chloride moves out of the cell, sodium ions typically follow. Where salt goes, water follows. Water following the movement of sodium and chloride thins the body's normally secreted mucus and allows the body to move the mucus naturally. Mucus is secreted by tissues in many organs to protect and lubricate the exposed surface of the tissue. In healthy individuals, the mucus is secreted in one spot and then swept away. Microbes trapped in the mucus are destroyed by stomach acid or other body mechanisms. When the cell has a defective CFTR protein water cannot move sufficiently in response to the movement of chloride. As a result, the mucus produced by CF patients has a thick, and viscous consistency and doesn't clear the tissues in a timely manner. Mucus is an excellent medium for the growth of microbes. The viscosity and slow movement of mucus in CF patients gives microbes time in which grow. The growth of microbes in CF patient mucus is the root of their health care issues. The thick mucus becomes infected leading to scarring of tissues and possibly sepsis. In addition to the respiratory tract, mucus is produced throughout the digestive, excretory and reproductive tracts.

The impact of cystic fibrosis on patient health varies from person to person. There are a number of treatments available to help patients. Some medications in use today include CFTR modulators, digestive enzymes to assist with food breakdown and nutrient absorption, mucolytics to breakdown mucus and antibiotics to prevent and treat lung infections. Lung transplants are also performed when a

patient’s lung function has been damaged by chronic infections, however there is a long waiting list for transplants and transplant surgery always has risks.