Type: Research Paper

Subject: Pathophysiology

Subject area: Nursing

Education Level: Undergraduate/College

Length: 5 pages

Referencing style: APA

Preferred English: US English

Spacing Option: Double

Title: Cystic fibrosis

Instructions: topic of paper cystic fibrosis

Focus: please pay attention to instruction attatched

Structure: please us format provided and pay attention to instruction attached

Pathophysiology of Cystic Fibrosis

Student's Name

Department & College Name

Course Number and Name

Professor's Name

Due Date

Description of Pathology

Cystic fibrosis is a progressive genetic disorder that affects both the lungs and digestive systems. It affects cells that produce mucus, sweat and digestive juices causes these fluids to be thick and sticky plugging up tubes, ducts and passageways. Cystic fibrosis symptoms vary from one patient to the other because of underlying comorbidities and other health factors. They include; coughs, lung infections, inability to gain weight and fatty stools.

Scientifically, CF is caused by a mutation within the CF transmembrane conductor regulator (CFTR) gene. This specific gene is important as it regulated the movement of chloride and sodium ions across the epithelial cell membranes. Without the chloride to attract water to the cell surface, the mucus in various respiratory and digestive organs become thick and sticky. Estimates show that close to 40,000 children and adults live with cystic fibrosis in the United States alone (Cystic fibrosis mutation database). Averagely, 105,000 people get diagnosed with Cystic fibrosis across different racial and ethnic groups. According to the Cystic Fibrosis Mutation Database (http://www.genet.sickkids.on.ca/cftr/) more than 1,400 people have identified affected with Cystic Fibrosis. Its prevalence therefore keeps on increasing especially in children. According to the above database, prevalence varies but

approximately 1in every 25 Caucasian Europeans are carriers of the CF gene with an approximation of 1 child in every 2500 births having CF (Cystic fibrosis mutation database).

Normal anatomy of the major body system effected

Cystic fibrosis is caused by a defective gene affecting many organ systems. while damage to lungs and pancreas are most critical, CF also involve other organs greatly affecting the lives of people affected. From anatomical standpoint, without the necessary chloride and sodium ions to attract water the mucus within the bronchioles which play an important part within respiration dehydrate the bronchioles causing obstruction by thickening mucus adhering to the walls of the airways (Ren et al., 2018). Small intestines are affected by CF through distal obstruction (meconium ileus). Both pancreatic duct and bile ducts are affected by obstruction. CF also impact the male reproductive organs by obstructing van deferens. Additionally, in female CF causes cervical canal obstruction by thickening of mucus within the reproductive organ. Musculoskeletal systems are also impacted by CF causing inspiratory muscle atrophy, weakening of anti-gravity muscles conditions such as gastrocnemius and kyphosis of the spine which often results in neck and back pain.

Therefore, lack of chloride shift from organ to organ by the CFTR protein leads to accumulation and increase of viscosity of mucus providing a hide out of bacteria from body's immune system. However, with CF infection, there is a paradoxical increase in sodium and chloride uptake causing increased water reabsorption causing thickening and dehydration of mucus. The abnormal chloride movement out of the cell leads to dehydration of mucus, pancreas, biliary secretion and other secretions. Exocrine glands are important in both respiratory and digestive organs (Ren et al., 2018). They are the organs responsible for excreting mucus and sweat glands. In lungs in causes suppurative ling disease in younger Caucasian populations.

With a depleted volume of airway surface liquid, respiratory system led to abnormal mu-conciliary clearance causing bronchiectasis, lung damage and other bacterial infections. The CFTR gene causes mucus to be sticky and thick abnormally blocking ducts towards the pancreases and the important digestive pancreatic enzymes is blocked from reaching the small intestine and performing normal digestive functions (Ren et al., 2018). Water scientifically improves biological diffusion processes within the cell membranes. CF affects the CF transmembrane conductor regulator (CFTR) gene which is important as it regulated the movement of chloride and sodium ions across the epithelial cell membranes or digestive organs.

Normal physiology of the major body system effected

Within the respiratory tract, the CTFR gene encodes the CTFR protein which acts as a bridge to a chloride channel that is present within the epithelial tissues. Within the process of diffusion and osmosis, chloride is driven against its concentration to create a balance. CFTR is present in the airway's epithelial cells and submucosal glands. CF therefore obstruct causing defective results within the disruption of chloride ion movement and further affects sodium reabsorption reducing the amount of water secretions (Ren et al., 2018). All these reduce the amount of airway surface liquids. With reduced airway surface liquids, immunological functions cannot be complete impeding mucus clearance. With such an alteration to lungs, bacteria growth with biofilm mode of growth attaches to the host immune system contributing to future damage.

The pancreas is also affected by because the impacts of CF cause a permanent damage to the exocrine pancreases rendering people with CF to have insufficient pancreatic juice within their body, this can cause CF-related diabetes mellitus. Within the gastrointestinal tract, viscous mucus cause bowel obstruction in-utero causing meconium ileus. It can also

cause cholestasis in biliary tree in-utero which results into neonatal jaundice (Quittner et al., 2016). If untreated, it can result to distal intestinal obstruction syndrome and other liver conditions. Studies show that 98% of men with CF are infertile, Why? Because of the congenital absence of the vas deferens.

Mechanism of Pathophysiology

While from above we have noted that cystic fibrosis affects cells that produce mucus, sweat and digestive juices making these secretion pug the tubes, ducts and passageways within respiratory and digestive organs. The pathology of CF is merited by a mutation within the CF transmembrane conductance regulator gene. They pathophysiology therefore works by decreasing mu-cociliary clearance altering sodium ion transport and chloride giving an opportunity for bacteria colonization within the respiratory tracts and digestive systems. According to Quittner et al. (2016), the inability of shift of these ions causes bacterial infections of *Pseudomonas, Haemophilus influenza and Staphylococcus aureus* which is normally caused by obstruction.

Prevention

CF pathophysiological structures indicate that it resides within a gene making it difficult to prevent. CF therefore at this point is not possible to prevent. Most studies indicate that most babies have CF caused by genetic transference from their parents (Ratjen et al., 2015). The disease therefore is already present in organs such as pancrease and the liver but will start to show signs after birth. Gene editing or therapy may be the future of cystic fibrosis but for now, it is a grey area which needs a lot of research. Since both parents must have the CF gene to pass it to their child, future might edit these specific genes preventing CF occurrence.

Treatment

Before treatment patients are diagnosed. In children blood samples are tested for higher levels of chemicals called immunoreactive trypsinogen (IRT) which is normally released by the pancreas. Sweat test can also be used to test on CF by checking the salt content. Older people can be tested through genetic and sweat tests in the event of recurring bouts of inflamed pancreas, nasal polyps, chronic sinus, infertility and lung infections. As at now, we have no cure for cystic fibrosis but medication and therapies that can ease the symptom and reduce the disease complication improving the quality of life (Ratjen et al., 2015). Therefore, managing cystic fibrosis is complex however, the medication must be objective in prevention and controlling of infections, remove and loosen mucus from the lungs, treat and prevent intestinal blockage and provide adequate nutrition. Anti-biotics, anti-inflammatory, inhaled medications, oral pancreatic enzymes, stool softeners, acid-reducing medication among others help in reducing the severity of symptoms. other interventions include surgical procedures and therapies which include nasal and sinus surgery, oxygen therapy, non-invasive ventilation, feeding tube, bowel surgery and liver transplant.

Conclusion

While it is true that future research may have an answer towards CF through gene editing, it is important to be first proactive towards developing prevention strategies than cure. Advancements in diagnosis and management have since provided comprehensive diagnosis and delayed disease progression increasing patient survival levels.

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