Presentation Outline and Integrated Resources

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# Introduction

Cystic Fibrosis is a common genetic disease in white children in the US and Canada, which shortens life. Cystic fibrosis is a genetic sickness that targets the secretory glands, together with the mucus and sweat glands. The disease is characterized by the accumulation of dense, gluey secretion that can cause mutilation to a lot of organs of the body. The major affected organs of this disease are lungs, pancreas, liver, intestines, sinuses, and sex organs.

## Thesis Statement

The researches have shown that the disease can be treated better if diagnosed at an early stage and even prevented by taking the precautionary measures provided by the public health sectors.

## Main Issues

The human body releases the emission called mucus that aids the lubrication of the gastral structure, generative structure, and the lining of the respirational structure accompanied by other body organs and tissues. Nevertheless in the case of this disease the mucus produced is dense and instead of the real function of lubricating that it is supposed to serve, it clogs the intestines, and the airways lining. This leads to the austere problem of breathing and lungs infection. People with this disease suffer from the digestive problems chronic coughing, wheezing, and inflammation.

**Pathophysiology and Etiology**

## Etiology (Causes)

### Gene Mutation

### Cystic Fibrosis is the result of the alteration in the CF transmembrane conductance controller gene. The gene gives orders to form the CTFR protein, a strait in the cell tissue that regulates the actions of chloride ions, in and out of cells. This process is vital for the making of secretion, perspiration, drool, and moans. Mucus is required for the correct working of numerous organs. Transmutations in the CFTR gene upset the purpose of chloride passages, averting them from controlling the movement of chloride ions and water crossways cell films. Consequently, cells that make the lining of the hallways of lungs, pancreas, and other body part yield mucus which is quite dense and gluey. This blocks the respiratory tracts and numerous channels, producing the typical signs and indications of CF.

### Risk Factors

1. Both parents have irregular CF genes and transfer that gene to their baby.
2. Environment and Lifestyle: CF patients need to take lots of calories to maintain weight, which is quite difficult for them. Alcohol consumption and smoking can worsen the disease further.
3. Age: The disease gets worse with every coming year.
4. Race: Although it arises in all the races however it is very common in the folks of North European descent.

## Pathophysiology

The complications that arise as a result of the disease vary patient to patient depending on the extent to which the mutation has occurred in the CFTR gene.

### Respiratory System Complications

1. Bronchiectasis: CF is a major reason of bronchiectasis. The airways are damaged in this condition, and the difficulty in breathing arises.
2. Chronic Infections: Due to the thick mucus in lungs the infection prolongs and the patients suffer from a sinus infection, pneumonia etc.
3. Nasal Polyps: As the inside layer of the nose is enflamed, the soft growths develop.
4. Hemoptysis: CF can cause thinning of the linings of the airways that result in patients coughing up blood.
5. Pneumothorax: In this state, the air collects in the middle of the lungs and the chest wall and causes the chest pain. Usually occurs in older people.
6. Respiratory Failure: with the passage of time the disease damages the lung tissues that they no longer work.
7. Acute Exacerbations: worsening of the respiratory system such as short breaths, and chronic coughing for weeks.

### Digestive System Complications

1. Nutritional Deficiencies: tubes that take digestive enzymes that help our body absorb proteins, fats, or fat-soluble vitamins, from the pancreas to intestines are blocked by mucus.
2. Diabetes: Insulin is produced by Pancreas. 30% of CF patients get diabetes around the age of 30.
3. Blocked Bile Duct: tube carrying bile from liver and gallbladder to the small intestine due to blockage and inflammation, leads to liver diseases.
4. Intestinal obstruction: Children and grownups with CF are more probable than the newborns to have intussusception, a disorder in which a fragment of bowels folds in on itself.
5. Distal Intestinal Obstruction Syndrome: It is complete or partial blockade of the point where small and large intestines meet.

### Reproductive system complications

1. Men: Nearly all males are sterile having CF, as the tube joining their testes and the prostate gland is stuffed with thick mucus, or sometimes totally absent.
2. Women: women are infertile than the normal females yet they have a chance to conceive and produce babies. However, the symptoms and signs of CF worsen further in pregnancy.

### Other complications

1. Osteoporosis: CF patients are more probable to have the thinning of bones.
2. Electrolyte imbalances and dehydration: Sweat of CF patients is saltier so the mineral balance in their blood gets disturbed.

# Signs, Symptoms, and Treatment

Cystic fibrosis signs and symptoms differ, contingent on the harshness of the sickness. Even in the same individual, indications may get worse or recover with the Passage of time. Some individuals might not experience indications to teens or maturity. Symptoms of CF can be categorized into 2 key groups which are as follows.

## Respiratory Signs and Symptoms

1. A continuous cough that makes dense mucus
2. Out of breath
3. Breathing difficulty
4. Workout intolerance
5. Recurrent lung contagions
6. Swollen nasal channels or a clogged nose

## Digestive Signs and Symptoms

1. Odd-smelling, fatty feces
2. Reduced weight gain and development
3. Abdominal jam, mainly in infants
4. Extreme Constipation

## Other symptoms

1. Excessive sweating
2. Your child with CF may taste "salty" when kissed

# Screening

The diagnosis or Cystic Fibrosis is a multistep procedure, and a whole screening should involve the following steps.

## Recognition techniques

### Carrier Testing for Cystic Fibrosis

Genomic testing can be used to find out if an individual brings an alteration of the CFTR gene. The test examines an individual’s DNA, which is taken from cells in a blood taster or from cells which are taken from the mouth of the person.

### Newborn Screening for CF

It is done during the initial days of the child's birth, most of the times by the healthcare provider at the hospital. The methods of screening, however, differ everywhere.

### Sweat Test

This is the painless way of testing CF which can be done at any age. If the symptoms of CF appear, a sweat test at a CF Foundation-accredited care center can help conduct screening by the measurement of salt in an individual’s perspire.

## The potential impact on health and wellness

Timely identification and treatment can:

1. Increase development
2. Support keep lungs fit
3. Lessen hospital visits
4. Enhance life-span

# Education and Prevention

The guidelines were created in 2003 which were further updated in the year 2013 for the infection prevention and control for the cystic fibrosis.

## Recommendations and application to the field of public health

1. Education: Offer instructions to every health care worker and persons having CF and their families concerning IPC performs. Educational materials should be age-appropriate and language-appropriate.
2. Audits of IPC practices: CF care teams should cooperate with their IPC groups to make conventions, specifications, and reviews to regulate application of performs for cleaning and sterilizing versatile things and tops in the health care environment.
3. Contact precautions: Every health care worker should apply Interaction Protections (i.e., wear a gown and gloves) when taking care for all people with CF, irrespective of respirational region culture outcomes.

# Resources needed

Potential cystic fibrosis treatment uses 'molecular prosthetic' for missing lung protein. (2019). ScienceDaily. Retrieved 30 May 2019, from https://www.sciencedaily.com/releases/2019/03/190313143248.htm

Science Daily magazine has published an article related to the potential treatment of Cystic Fibrosis. This treatment uses the “molecular prosthetic” for the missing lung protein. The article has put the stance that some permitted medication generally used to treat mycological contagions could also do the work of a protein strait that is absent in the lungs of individuals having cystic fibrosis, functioning as a prosthesis on the molecular level, said by the fresh study from the University of Illinois and the University of Iowa. The article says that the researches have shown that the treatments are possible for some but not all the patients, but there is no proper cure. This treatment has the power to be the 1st ever treatment to cover every kind of cystic fibrosis, irrespective of the hereditary alteration that results in the protein shortage. The article has provided the studies conducted on pigs and humans and the results are based upon the valid researches.

Moran, F., Bradley, J., & Piper, A. (2017). Non-invasive ventilation for cystic fibrosis. Cochrane Database Of Systematic Reviews. doi:10.1002/14651858.cd002769.pub5

This article aims to compare the consequence of non-intrusive airing as opposed to no non-intrusive airing in persons with cystic fibrosis for airway clearance, while sleep and the workout. They researched the Cochrane Cystic Fibrosis and Genetic Disorders Group Trials Register containing the references recognized by the comprehensive electronic database searches, related journal, and other books. The results of the study showed that this might be the easiest method and the patients of CF might prefer it over the other methods. The results do not state the evidence for the results, that the non-invasive ventilation increased the amount of the mucus coughed up, however it improved the functioning of the lungs for a short while. The results of the study are based only on the single treatment sessions and that too on a tiny number of patients. So the evidence for the long term results is limited.

Nina Avramova and Susan Scutti, C. (2019). Genetically modified virus saves teen's life, offers hope in the fight against antibiotic resistance. CNN. Retrieved 30 May 2019, from https://edition.cnn.com/2019/05/09/health/cystic-fibrosis-teen-engineered-bacteriophage-study/index.html

On May 10, this year there was a report from the renowned channel, CNN about a girl who was diagnosed with Cystic fibrosis at the very young age of 11 months only. The report tells that the girl has been suffering since the age of 8 years with an on and off infection resulted from antibiotic-resistant bacteria. MB, or Mycobacterium abscesses’, the bacterium causing Isabelle's infection, is a type of antibiotic-resistant bug that, though commonly found in nature, does not usually cause sickness in healthy people. A hospital consultant told her mom that their daughter was not going to die and that they will take the less than 1% chance. They brought in use the years ago abandoned Phage therapy. In which the natural bacteria was used to kill the bacteria of disease. Within weeks of the treatment, the girl recovered and gave new hope for the patients suffering from the same disease. The research can be further worked upon as it has hope for other patients too.

Cutting, G. (2014). Cystic fibrosis genetics: from molecular understanding to clinical application. Nature Reviews Genetics, 16(1), 45-56. doi:10.1038/nrg3849

The article was published in the year 2014 and the main claim that it proposed was that the accessibility of the human genome order and tools for questioning distinct genomes offer an unparalleled chance to use genetics in medicine. Mendelian circumstances, which are instigated by dysfunction of one single gene, proposes influential instances that exemplify how inheritances can offer understandings into the syndrome. Cystic fibrosis, one or more generally deadly autosomal receding Mendelian sicknesses, is offered in the article as an instance. Fresh advancement in clarifying sickness mechanism and reasons of phenotypic disparity, along within the growth of treatments, make evident that genetics carry on to play an important role in cystic fibrosis research 25 years after the discovery of the sickness-producing gene. The research made a claim that genes play an important role in the development of the disease.

Tabori, H., Arnold, C., Jaudszus, A., Mentzel, H., Renz, D., & Reinsch, S. et al. (2017). Abdominal symptoms in cystic fibrosis and their relation to genotype, history, clinical and laboratory findings. PLOS ONE, 12(5), e0174463. doi:10.1371/journal.pone.0174463

This study claims that abnormal systems are the hallmark for the disease called CF, nonetheless, these abnormal symptoms are misunderstood, and get little scientific devotion as compared to the respiratory manifestation. The study aims to assess and quantify the abnormal symptoms and to relate them to the laboratory constraints, medical discoveries, and medicinal account. The study was conducted on a total of 131 patients suffering from cystic fibrosis of all ages. The questionnaire was on the topics like stomach ache and non-pain indications, ailments of hunger, intake, and bowel activities along with indication-related excellence of life. The results showed the common lack of appetite and loss of taste also the abdominal pain, and the distention. The research is opening doors for further study and is providing a close understanding of the patients, and their symptoms.

# Conclusion

The disease Cystic Fibrosis though genetic, yet carries some chances of being treated, and managed better if the precautionary measurements are taken. The scientists have researched on the disease for years yet the permanent cure for the disease cannot be found but there are some public health measurements that are provided by them to follow for the prevention from this disease.