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Mini Review

Cystic Fibrosis

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ABSTRACT

Cystic fibrosis also known as CF is a life threatening, hereditary disease which is non- curable autosomal recessive disorder and is chronic in nature. Almost, 100,000 of the world population is suffering from this disease at the current time and it is most commonly found in Caucasians. CF is caused due to the mutations caused in CETR genes that is cystic fibrosis trans membrane regulator gene.

In this disease the mucus becomes thicky and accumulates in organs associated with the respiratory system and GIT such as lungs, sinuses, pancreas, intestine, hepatobiliary tree, vas derens etc. causing dehydration and abnormality of chloride channels in mucus and sweat secreting cells, therefore causing problems in gastrointestinal tract and respiratory tract. It increases the risk of malignant tumors and cancer in gastrointestinal and pancreato-biliary tract and recurrent or acute pancreatitis, thus affecting the gut. Respiratory system is highly affected in this disease and the risks of bronchitis, asthma, pneumonia and several other air borne allergies is very high.

Keywords: Cystic; Fibrosis; Pancreato-biliary tract; Hepatobiliary tree

INTRODUCTION

Cystic fibrosis commonly known as CF is an incurable, chronic, hereditary and life-shortening disorder. It is an autosomal linked recessive disorder caused by the mutations in the CFTR genes that is cystic fibrosis trans membrane regulation genes and they mainly target on our respiratory tract and gastro-intestinal tract, thus making our immune system weak. This disease includes abnormality of the chloride channels in mucus and sweat producing cells, making the mucus thicker leading to its accumulation in the various ducts leading to several problems such as pneumonia, bronchitis, allergies, pancreatitis etc [1].

Research results show that over 1 lakh of the current world's population is suffering from this disease out of which 95% of them are Caucasians. 50% of the patients suffering from CF have osteopenia in which the frequency of production of new bone reduces as compared to the reabsorption of the bones and 10%-34% of them faces osteoporosis in which their bones become brittle and weak making them physically inactive. Symptoms vary from patient to patient and can include cough, repeated lung infections, inabaility to gain and loose weight, fatty stools, acute bronchitis, cough can be chronic with blood, pain in abdomen, diarrhoea, heatburn, bulky stools, constipation, wheezing, sinusitis, fatigue, pulmonary hypertension, shortness of breath, male infertility, pneumonia and salty sweat [2].

Majorly affected systems due to this disease are GIT and respiratory system. It increases the risk of malignancy in gastrointestinal pancrato-biliary tracts. Malnutrition and malabsorption leads to pancraetitis and presence of indigested fat and protein fibers could be found during stool analysis. This disease also increases the risk of cardiovascular disease and altered sex hormone production leading to delayed puberty, physical inactivity potentiates poor bone health [3].

Testing of the disease includes measuring the sweat chloride value, in patients suffering from CF the sweat chloride value falls below 60 m mol/l. CF is among first disease to have general population genetic screening test and one of the most common indications of prenatal and preimplantation genetic diagnosis for single gene disorder. Medical imaging is used to monitor pathologic changes caused by disease [4].

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Treatment depends on the severity. Treatment may lower down the symptoms and reduce complications but cannot cure the disease. New born screening helps in the early diagnosis. Supportive care, medications such as dietary supplements, antibiotics, penicillin and cough medicines, self-care, drug therapy, extensive physiotherapy, medical procedure such as chest wall oscillation and lung transplantation can be of great use in curing the problems to a huge extent [5].

As, it is clear from the research studies that this disease cannot be cured therefore we should lay more emphasis on the techniques that would increase the life span of the individual suffering from CF. As, it is well said that prevention is better than cure, therefore all the preventive measures should be taken by the patient, his family and medical staff so that the patient could suffer less and researchers should continue their work in the field of making this disease curable from non-curable [6].

LITERATURE REVIEW

Objective

To find new techniques to treat this mutation in the genes and to increase the survival rate of the victims suffering from this disease using several prenatal and preimplantation genetic diagnosis methods [7]. Using previous data from several researches and literature review of available sources such as PubMed and Google Scholar to explore the area of cystic fibrosis and to see it from specific perspective [8]. In 1955, new centers were established with programs of aggressive and comprehensive care leading to the increased longevity of the person suffering from CF. CF centers provided opportunity for the conduction of clinical trials and also were meant to invent and discover new therapies for the patient suffering from CF. The opening of these centers provided a way for the conduction of new diagnostic test, opportunities for research and prospects for using gene as therapy. In 1959, the discovery of the sweat chloride tests made it easier to test this disease in the milder cases [9]. Earlier, this disease was considered as Celiac syndrome and was placed in the category of separate disease which included symptoms such as malabsorption of fat and protein, steatorrhoea, growth failure, pulmonary infection and nutritional failure which were studied earlier during the autopsy studies of the malnourished infants [10].

The increased sodium reabsorption in airways defect in chloride channels leading to chlorine secretion in response to cAMP were the most common symptoms that were seen for this disease. Earlier, this disease was also known as mucoviscidosis that was generalized exocrionopathy as it caused inflammation and widening in the ducts of the exocrine ducts such as bile ducts, mucus glands etc., thus making the sweat salty and mucus thicker [11].

Cystic fibrosis signs and symptoms vary, depending on the severity of the disease. Even in the same person, symptoms may worsen or improve as time passes. Some people may not experience symptoms until their teenage years or adulthood. People who are not diagnosed until adulthood usually have milder disease and are more likely to have atypical symptoms, such as recurring bouts of an inflamed pancreas (pancreatitis), infertility and recurring pneumonia. People with cystic fibrosis have a higher than normal level of salt in their sweat. Parents often can taste the salt when they kiss their children. Most of the other signs and symptoms of CF affect the respiratory system and digestive system.

DISCUSSION

The diagnosis of this disease is not available, in CF we can only increase the life span of a person by pre-implanting the lungs of a person with high immune resistance, but this procedure also increases the life span by 5 years only. No permanent solutions are yet available and therefore new techniques are yet to be discovered for CF thus, it is life limiting and will definitely lead to death (Figure 1) [12].

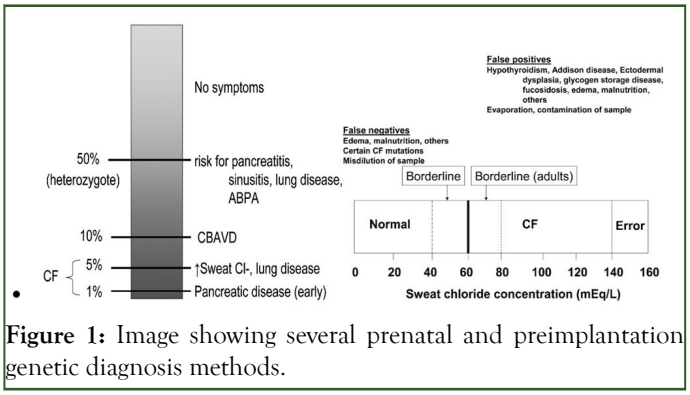

Figure 1: Image showing several prenatal and preimplantation genetic diagnosis methods.

Research results have shown that over 1 lakh of the world population is suffering from this disease but only preventive care methods have been found out for CF. We need to work more to find new and cheaper way to save patients life and increase their life span. The most common way to test this disease includes commercial testing of the 86 most common alleles after which in 93% of the patients the presence of the CF can be identified. After, 1989 when the CF gene was introduced the testing of the disease was done by the direct identification of 2 mutant CF alleles [13].

Recent research results shows that the lesions in cAMP-regulated chloride channel, CFTR, that is expressed in many epithelial cells, including sweat duct, airway, pancreatic duct, intestine, biliary tree and vas deferens giving rise to increased sweat chloride concentration, lung disease along with bacterial infection and bronchiectasis, pancreatic insufficiency, intestinal obstruction, biliary cirrhosis and congenital bilateral absence of vas deferens, often in combination. Initially, it was considered a pathologic diagnosis. But, now testing is done by measuring the sweat chloride value, as in patients with CF the value of sweat-hloride becomes 60 m mol/l or elevates above it. In order to get reconfirmation of the disease the second test is conducted even then if the results are unclear then the sperm of the semen is checked because ones with CF are azoospermic sperms and checking of gall bladder, pancreas, liver functions are checked and NPD that is nasal potential difference is measured.

CONCLUSION

Its management therapies include only drug therapies, extensive physiotherapy, nutritional support and several gene and mutation therapies. Yet, we need to work on life enhancing techniques that could increase the life of the patient. The most common techniques for the diagnosis of CF included sweat chloride tests, Pilocarpine ionotophoresis and testing of the immune reactive trypsin levels in newborns blood spot are the latest techniques for the identification of CF. These techniques have increased the life expectancy of the patient from 6 months to 30 years. But it is necessary that the centers that are conducting these tests and researches should meet national standards and should lay more emphasis on increasing of life expectancy of and individual suffering from CF.

REFERENCES

1. di Sant'Agnese PA. Fibrocystic disease of the pancreas, a generalized disease of exocrine glands. J Am Med Assoc. 1956;160(10):846-853.
2. Andersen DH, Hodges RG. Celiac syndrome: V. genetics of cystic fibrosis of the pancreas with a consideration of etiology. Am J Dis Child. 1946;72(1):62-80.
3. Farber S. Pancreatic function and disease in early life v. pathologic changes associated with pancreatic insufficiency in early life. Arch Pathol. 1944;37:238.
4. di Sant'Agnese PA, Darling RC, Perera GA, Shea E. Abnormal electrolyte composition of sweat in cystic fibrosis of the pancreas: Clinical significance and relationship to the disease. Pediatrics. 1953;12(5):549-563.
5. Gibson LE, Cooke RE. A test for concentration of electrolytes in sweat in cystic fibrosis of the pancreas utilizing pilocarpine by iontophoresis. Pediatrics. 1959;23:545-549.
6. Rieder F, Biancani P, Harnett K, Yerian L, Falk GW. Inflammatory mediators in gastroesophageal reflux disease: Impact on esophageal motility, fibrosis, and carcinogenesis. Am J Physiol Gastro Liver Physiol. 2010;298(5):G571-G581.
7. Chiang DJ, Pritchard MT, Nagy LE. Obesity, diabetes mellitus, and liver fibrosis. Am J Physiol Gastro Liver Physiol. 2011;300(5):G697-G702.
8. de Carvalho AA. Function tests in the celiac syndrome and in cystic fibrosis of the pancreas. Pediatr Pratica. 1961;32:261-264.
9. Letondal P. Pediatrics; celiac disease, cystic fibrosis of pancreas and congenital megacolon. Union Med Can. 1957;86(11):1271-1277.
10. Chaptal J, Jean R, Bonnet H, de Paulet CR. Celiac disease and celiac syndrome; 11 case reports. Sem Hop. 1957;33(25/4): 1622-30P.
11. Chung AW, Morales S, Snyderman Se, Lewis JM, Holt Jr LE. Studies in steatorrhea: Effect of the level of dietary fat upon the absorption of fat and other foodstuffs in idiopathic celiac disease and cystic fibrosis of the pancreas. Pediatrics. 1951;7(4):491-502.
12. Venuta A, Bertolani P, Casarini R, Ferrari F, Guaraldi N, Garetti E, et al. Coexistence of cystic fibrosis and celiac disease. Description of a clinical case and review of the literature. Pediatr Med Chirurgica Med Surg Pediat. 1999;21(5 Suppl):223-226.
13. di Sant'agnese PE, Andersen DH. Celiac syndrome: IV. Chemotherapy in infections of the respiratory tract associated with cystic fibrosis of the pancreas; observations with penicillin and drugs of the sulfonamide group, with special reference to penicillin aerosol. Am J Dis Child. 1946;72(1):17-61.