Screening For Cystic Fibrosis

Cystic fibrosis (CF) is a hereditary life-threatening disease, which causes damage to the digestive system and the lungs, eventually, limiting an individual's ability to breathe over time. It is caused by a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which is responsible for the coding of the CFTR protein. The CFTR protein controls the movement of salt and water in and out of body cells. The mutation in the CFTR gene causes the CFTR protein to be ineffective, causing fatal symptoms of cystic fibrosis to occur. Population diagnosed with respiratory (lung) cystic fibrosis often face breathlessness, coughing, lung infections, damaged airways, nasal polyps, etc. People detected with digestive system cystic fibrosis experience nutritional deficiencies, diabetes, intestinal obstructions, distal intestinal obstruction syndrome, etc. These symptoms significantly affect an individual's well-being and quality of life.

Several Scientists are trying to find ways to cure this disorder, although currently there is no cure to eradicate cystic fibrosis. However, there are various treatments available that may help relieve symptoms of this condition, improve a person's quality of life and life span (1). The outlook for people with cystic fibrosis has improved dramatically in recent years, largely due to screenings (2). To detect CF in an individual showing symptoms, they must complete screenings. This scientific report focuses on the role of screenings in the treatment and potential cure of cystic fibrosis. Therefore, this report poses the following question:

'Are screenings for Cystic Fibrosis helping to reduce its prevalence in the population?'

Background

Cystic fibrosis occurs due to a mutation in a gene on chromosome number 7, one of 23 pairs of chromosomes that children inherit from their parents (3). The parts of the body significantly affected by cystic fibrosis include sweat glands, digestive system, respiratory system, and reproductive system (4). The sweat glands are affected by the mutation as the sodium and chloride present in the sweat are elevated. Whereas the digestive system is affected in a way that the enzymes are prevented from reaching the small intestine to digest food, due to the thick and sticky mucus blocking the pancreatic ducts. Pancreatic insufficiency results in incomplete digestion and poor absorption of nutrients. The cilia move dust, germs, mucus, and bacteria from the smaller passages to the larger ones where they can be coughed out. However, in a diseased individual, the thick and sticky mucus cannot be moved out of the smaller passages easily, which eventually clogs the air passages, causing long-term symptoms of cough and lung infections. Several lung infections could cause lung failure and breathing problems. Individuals analyzed with Cystic fibrosis are inherited with normal external reproductive organs however, about 95% of men and 25% of women with cystic fibrosis are infertile (5). This is due to thick and sticky mucus blocking the vas deferens in men and the cervix in women, making them infertile.

Cystic fibrosis is an autosomal recessive condition, hence for an individual to inherit the disorder the parents must each carry one copy of the mutated gene, signifying that the parents are carriers (heterozygous). This process can be explained through the punnet square (figure 1),

where parent 1 (paternal) and parent 2 (maternal) are both carriers of the gene. Even though both the parents might be carriers there is only a 25% chance that the gene is not expressed, meaning that the offspring is not either a carrier nor are they being affected by the mutation. Rather there is a 50% probability of the offspring having the allele not being expressed, meaning that they are a carrier (usually does not display the symptoms of the disease). Which leaves only a 25% chance for the offspring to be inherited with cystic fibrosis.

There are several treatments available to reduce the symptoms caused by the condition. These include; medications, antibiotics, chest physical therapy, home care, gene therapy, lung transplant, anti-inflammatory treatment, nutrition, etc. However, the two main increasing applications of treatment related to CF are newborn carrier screening and carrier screening. Newborn carrier screening for CF has now been implemented in the majority of countries where CF is common, including, North America, Australia, and several parts of Europe (6). Newborn screening allows the patients diagnosed with CF to have early access to treatments, therefore, reducing the symptoms resulting in a better quality of life. However, carrier screening aims to detect if people have or carry changes to the gene that carries cystic fibrosis (7). Carrier screening is the only way to find out if an individual is a CF carrier prior to pregnancy. This enables informed reproductive choices before the birth of a child with CF. Carrier screening is a significant process and must be performed by all couples as this is crucial for genetic counseling and if a positive CF result is found then assist in making the decision of abortion or termination.

Evidence

Carrier screening first began in 1989 in the Brittany region of France (8). In 1989 a study in New South Wales, Australia on CF diagnosis by neonatal (newborn) carrier screening and by clinical presentation, which depicted that children diagnosed with cystic fibrosis by Neonatal carrier screening had a better survival rate than children diagnosed with CF by clinical presentation. This can be seen in the graph below:

Individuals diagnosed with CF by neonatal screening have over a 75% survival rate after the age of 25. Whereas, patients diagnosed with CF by clinical presentation have a survival rate of 50% by the age of 25. This clearly shows that newborn carrier screening increases the life span of an individual.

Another research done in 2005 by the department of pediatrics in Europe, displays whether newborn screening for cystic fibrosis leads to better-quality diagnosis. This study also compares individuals who received neonatal screening and individuals who had no screening:

“We used long-term survival, early mortality, nutritional and pulmonary status, and the number of hospital admissions as outcome measures. Effects on the reproductive behavior of the parents and relatives were also assessed. In 2 studies, a similar trend for the improved long-term survival rate of the screened cohort was observed, whereas in 2 other studies CF NBS (newborn screening) appeared to prevent CF-related deaths in infancy and early childhood…. In most studies, patients who were screened were found to have less lung damage than their non-screened peers. CF NBS significantly reduced the number of affected children who required hospitalization. In Brittany, France, a reduction of 15.7% in CF prevalence at birth was attributed to the introduction of an NBS program for CF. We conclude that there is accumulating evidence that CF NBS prevents early CF-related deaths and leads to a substantial and prolonged health

gain for patients with CF” (9).

Carrier screenings provide several reproductive options in future pregnancies among parents of CF patients and CF patients with an intention of having children. Having reproductive choices for pregnancies reduces the prevalence and symptoms of CF in the population. The preferred reproductive choices selected by parents of CF patients for future pregnancies include spontaneous pregnancy with prenatal diagnosis 53.1%), spontaneous pregnancy without prenatal diagnosis (6.2%), preimplantation genetic diagnosis (56.2%), and adoption (6.25%). Whereas, CF patients preferred preimplantation genetic diagnosis (100%) and adoption (12.5%). This is shown in the table below:

Evaluation

Even though several different sources were used to obtain the data for this report, there have been several limitations. One of which includes the relevancy of the evidence found. 2 of the 3 sources used to discover the evidence were extremely outdated, therefore, affecting the research’s relevancy. Source one was created in 2013 however, it contains information that is extremely outdated as it was from the 1980s. Source 2 was written in 2005 which is once again outdated. However, source 3 was fairly relevant as it was written in 2015, providing accurate information.

Another limitation includes the sample size of source 3, as it only surveys 32 parents of CF and 16 CF patients. The limited sample size could hinder the accuracy and validity of the results. Even though sources 1 and 2 provided significant information regarding proving the research question, there was no sample size given. Therefore, future research needs to be done relating to the quality and accuracy of these sources.

The most significant limitation was trying to find relevant information that is ideal to answer the research question. Due to the difficulties faced in relation to finding relevant information the research question was changed 2 times before relevant research was obtained. As well as that within-study 2, not all the information was accessible meaning that it was just an abstract of an article. Therefore, the accuracy of the information used may not be correct.

Lastly, there was a lack of evidence to answer the research question. This could be due to the fact that there haven’t been major studies done on cystic fibrosis and screenings. However, it was found that there is a lot of awareness for cystic fibrosis, but there are not that many studies or researches being done to prevent it. Therefore, it is recommended that further studies are prepared so that a cure could be sustained for this disease.

Conclusion

To conclude, the evidence found in this report supported the research question, ‘Are screenings for Cystic Fibrosis helping to reduce its prevalence in the population?’ it was found that newborn carrier screening and carrier screening are helping to reduce the prevalence and symptoms of cystic fibrosis as early treatment can occur, improving an individual’s quality of life. Carrier screenings allow reproductive decisions regrinding CF future pregnancies to be made to reduce the prevalence of the disease in society. however, future research needs to be conducted regarding the reliability of the sources, to accurately answer the research question