Lifestyle & Culture

Exploring the genetic basis of cystic fibrosis


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Cystic fibrosis (CF) is a genetic disease that affects numerous children and adults worldwide. This disease has been known to cause various problems related to the respiratory, digestive and reproductive systems.

CF is a rare disease that primarily affects individuals of northern European ancestry. In this population, approximately 1 in 2,500 new-borns is diagnosed with CF. The disease is less common in other ethnic groups, such as African Americans and Asians, presenting in approximately 1 in 17,000 and 1 in 90,000, respectively. However, CF can occur in any ethnic group and it is important to note that carrier testing is available for all populations. According to the EU data, approximately 1 person in 20,000 in Malta has CF. Worldwide, it is estimated that CF affects approximately 70,000 people.

CF affects both males and females equally and the disease is usually diagnosed in infancy or early childhood. Symptoms of CF vary depending on the severity of the disease and can include chronic cough, wheezing, shortness of breath, recurrent lung infections, poor growth and weight gain and frequent bowel movements that are greasy and foul-smelling.

Like with many other genetic disorders, CF results from a change or mutation in a particular gene, known as the cystic fibrosis transmembrane conductance regulator (CFTR) gene. A gene is a set of instructions passed down from parent to child which relays information to our body on the way we grow and develop.

The resultant genetic change highlighted above causes a problem in the protein produced by the CFTR gene, known as the CFTR protein. This protein helps move salt and water in and out of the cells in our bodies, however, in people with CF, this protein does not function properly resulting in a build-up of thick mucus in different parts of the body, primarily in the respiratory and digestive systems. This build results in recurrent infections since increased mucous provides optimal conditions for bacteria to grow.

In spite of his health challenges, Renson Mizzi was able to pursue his passion for hip-hop music and became a semi-finalist on Malta's Got Talent 2022.

CF is an autosomal recessive disorder, which means that a person must inherit two copies of the defective CFTR gene, one from each parent, to develop the disease. People who carry one copy of the gene are called carriers and do not have the disease, but they can pass it on to their children. If both parents are carriers, there is a 25% chance with each pregnancy that their child will have CF.

Due to its severity, CF may cause a range of symptoms, including persistent coughing with phlegm or blood, wheezing, shortness of breath and frequent lung infections. Patients suffering with CF may also experience poor growth or weight gain despite eating a healthy diet, difficulty gaining weight and irregular bowel movements. CF is usually diagnosed in infancy or early childhood through new-born screening, which involves testing a small blood sample for elevated levels of a protein. If levels are high, further testing is done to confirm the diagnosis.

Although there is currently no cure for CF, there are several treatments available that can help manage the symptoms and improve quality of life for people with the disease. The goals of CF treatment include clearing mucus, preventing or treating lung infections, improving nutrition, managing complications, such as diabetes and liver disease, improving lung function and enhancing quality of life.

Some of the latest treatments available for CF include CFTR modulator therapy. These medications target the underlying cause of CF by improving the function of the defective CFTR protein. These drugs have been shown to improve lung function while reducing the number of lung infections.

In addition to CFTR modulator therapy, there are several other treatments available for CF, including airway clearance techniques, such as chest physiotherapy and the use of a vibrating vest, which helps to loosen and clear mucus from the lungs. Antibiotics are also commonly used to treat and prevent lung infections in people with CF.

Managing nutrition is also an important aspect of CF treatment, as people with CF may have difficulty absorbing nutrients from food. A high-calorie, high-fat diet is often recommended, along with pancreatic enzyme replacement therapy, which helps the body digest fat and protein.

Despite its symptoms, individuals with CF are still able to lead a normal lifestyle. Attest to this is Renson Mizzi, a young man thriving despite being diagnosed with cystic fibrosis. In spite of his health challenges, Renson was able to pursue his passion for hip-hop music and became a semi-finalist on Malta's Got Talent 2022. His talent and determination inspires many and gives hope to other individuals living with rare and chronic diseases.

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