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Cystic Fibrosis: Understanding the Genetics of this Chronic Condition

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Opinion Article

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DESCRIPTION

Cystic Fibrosis (CF) is a serious, genetic disease that affects the lungs and digestive system. It is caused by mutations in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene, which controls the production of a protein that helps regulate the flow of salt and fluids in and out of cells. When this protein is not produced properly, it results in thick, sticky mucus that clogs the lungs and obstructs the pancreas. CF is a complex disease with many different types and mechanisms. It is important for individuals with CF and their families to work closely with healthcare professionals to manage the disease and improve their quality of life.

The CFTR gene is mutated, which results in CF. The most frequent mutation, designated as "F508," is a deletion of three nucleotides that eliminates the amino acid phenylalanine (F) from the protein's 508th position. 90% of CF cases in the United States and 70% of cases worldwide are caused by this mutation, however CF can also be caused by over 700 other variations. Even though the CFTR gene has two functional copies (alleles) in the majority of people, only one is required for cystic fibrosis prevention. When neither allele is able to create a functioning CFTR protein, CF develops. Consequently, CF is regarded as an autosomal recessive condition.

The CFTR gene, which is 230,000 base pairs long and is located at the q31.2 region of chromosome 7, codes for a 1,480 amino acid long protein. The precise location is on the long arm of chromosome 7, region 3, band 1, sub band 2, designated as 7q31.2, between base pairs 117,120,016 and 117,308,718. The CFTR is structurally a type of gene called an ABC gene. The CFTR protein, which is the gene's end product, is a chloride ion channel crucial for the production of perspiration, digestive secretions, and mucus. This protein has two ATP-hydrolyzing domains, which enable it to utilise ATP as an energy source. Additionally, it has two domains with each having six alpha helices that enable the protein to pass through cell membranes. The protein can be activated by phosphorylation, mostly by cAMP-dependent protein kinase, thanks to a regulatory binding site. A PDZ domain connection holds the protein's carboxyl terminus to the cytoskeleton. Rare ion-transporting cells that control the characteristics of mucus are responsible for producing the majority of CFTR in the lung's airways.

Symptoms of CF can vary from person to person, but typically include chronic cough, frequent lung infections, difficulty breathing, poor growth, and digestive problems. CF is usually diagnosed in infants, but some people may not be diagnosed until later in life. The symptoms of cystic fibrosis often appear in childhood. Because of malabsorption, newborns and infants with cystic fibrosis frequently have big, greasy faeces and are underweight for their age. Meconium blocks the small intestine in 15%–25% of babies, which frequently requires surgery to fix. Occasionally, bile duct obstruction causes neonatal jaundice in newborns. Parents frequently notice salt crystallising on their child's skin or a salty taste when they kiss them because children with cystic fibrosis lose excessive amounts of salt in their sweat.

Progressive lung deterioration, which finally results in respiratory failure, is the main cause of illness and mortality in patients with cystic fibrosis. Typically, this starts as a persistent respiratory illness that persists until medications are administered. People with cystic fibrosis almost always have a chronic infection in the respiratory system, with *Pseudomonas aeruginosa*, fungus, and mycobacteria becoming more prevalent over time. Runny nose and nasal blockage are brought on by upper airway inflammation. Nasal polyps are frequent, especially in children and adolescents. People often experience shortness of breath and a persistent cough that produces phlegm as the condition worsens. Exercise becomes increasingly difficult due to breathing issues, and people who suffer from persistent illness are underweight for their age.