PERSPECTIVE

Cystic fibrosis: Gene deficiency

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INRODUCTION

While initially thought of as a group of related syndromes, Cystic Fibrosis (CF) is now understood to be a single disease with a wide range of symptoms caused by the widespread tissue distribution of the ion channel and regulator, cystic fibrosis transmembrane conductance regulator, which is defective in CF (CFTR). The pancreas ability to operate and the kind of mucosal secretions are both affected by defective CFTR protein. The second of these consequences most likely contributes significantly to the CF patients reduced resistance to many infections.

With the notable exception of the chronic lung infection caused by the gram-negative bacterium *Pseudomonas aeruginosa*, many CF related illnesses have been successfully managed as a result of these factors. The ability of *P. aeruginosa* to acquire a mucoid phenotype, which renders this microbe resistant to both the innate and acquired immunologic defenses of the host, is one of the factors that contribute to its virulence. Other factors include antibiotic resistance, the capacity to use quorum-sensing signals to form biofilms, the destructive potential of a variety of its microbial toxins, and the ability to utilize quorum sensing.

DESCRIPTION

The clinical syndrome known as Cystic Fibrosis (CF) includes gastrointestinal, nutritional, and other problems as well as a chronic lung infection. The Cystic Fibrosis (CF) genetic basis is a well-known, severe monogenic recessive condition that is most frequently encountered in Caucasian people of European ancestry and is caused by mutations in the CFTR gene. Even while the gene deficiency causes a wide range of medical issues for the patient, the most troublesome clinical trait, chronic *Pseudomonas aeruginosa* lung infection, enables the fundamental pathologic process in CF to be classified as an infectious disease. 80% to 95% of CF patients eventually pass away from respiratory failure brought on by a persistent bacterial infection and related airway inflammation.

Research efforts exploded after the genetic flaw that causes CF was identified in 1989, and this resulted in a better knowledge of the molecular mechanisms behind the disease's numerous phenotypic presentations. The extent of the connection between CFTR mutant forms and persistent bacterial respiratory infections, particularly those caused by *P. aeruginosa*, is yet unclear. Understanding this connection is crucial because the majority of the disease's morbidity and mortality are caused by this infection and the resulting inflammation.

During infancy and early childhood, the lungs of CF patients are frequently colonised or infected with germs like *Staphylococcus aureus* and *Haemophilus influenzae* that may harm the epithelial surfaces, resulting in increased attachment of and eventual replacement by *P. aeruginosa*. But there have never been published suitable clinical trials to ascertain the part played by these organisms in the etiology of lung illness in CF patients. These microorganisms would be regarded as a true infection requiring treatment if they were found in Bronchoalveolar Lavage (BAL) fluid taken from the lung.

The progression of CF patients to respiratory failure due to *S. aureus*, non-type able *H. influenzae*, and comparable organisms isolated from oropharyngeal cultures is yet unknown. Instead, the pathogenic function of *S. aureus* and non-type able *H. influenzae* in the onset of lung illness in CF patients is primarily inferred from clinical anecdotes, but is otherwise devoid of any strong evidence from research in the peer reviewed literature.

There hasn't always been a persistent pseudomonal sinopulmonary infection associated with CF. The documented prevalence of CF pseudomonal infections was minimal before 1946. But according to a number of publications, *P. aeruginosa* rose to the top of the food chain in the airways of CF patients by the 1960's.

Although universal neonatal screening for immunoreactive trypsin is used in some countries, the diagnosis of cystic fibrosis is typically made clinically, allowing for extremely early diagnosis immediately after birth. Chronic respiratory infections, gastrointestinal problems that cause malabsorption, and nutritional deficits are common CF presentation symptoms. A sweat test is used to make the final diagnosis. The "gold standard" for diagnosis continues to be a sweat chloride concentration of more than 60 mmol/liter as assessed on two or more occasions by quantitative pilocarpine ionophoresis.

Neonates, whose sweat chloride levels may be momentarily high and older persons, whose sweat chloride levels typically raise, both of which might result in false positive tests, can make interpretation difficult. Similar to severe protein malnutrition, hypoproteinemic edema (leakage of fluid from serum due to low serum protein level) and hypochloremia (loss of chloride electrolytes due to dehydration) in malnourished patients can result in false negative tests. False negative sweat tests may also result from the specific CFTR mutations that each patient's unique combination of CFTR mutations carries.

Channels, transporters, and proteins related to the apical cytoskeleton scaffolding of epithelial cells are among the proteins with which CFTR interacts in its function as a conductance regulator. These proteins are prepared to take part in the secretory processes that were formerly attributed to the ORCC but are now recognized as being controlled by CFTR. Investigations into these interactions have clarified confusing results, such as the increased Na^+ absorption observed in CF airways, an aberration that led to a clinical study of aerosolized amiloride, which blocks salt absorption, for the treatment of CF. Both directly and indirectly through CFTR facilitated ATP release, the ORCC and CFTR appear to interact. Studies showing a failure to activate the ORCC when PKA and ATP are given to either side of the channel in the absence of CFTR imply a direct connection between the CFTR and the ORCC.

CONCLUSION

When applied to the extracellular surface of cultured human airway epithelia, ATP and other nucleoside triphosphates are known to increase Cl secretion. Additionally, it was discovered that ATP and UTP are equivalent *in vivo* Cl secretagogues. In fact, both normal and CF airway epithelial cells responded to nanomolar quantities of extracellular ATP or UTP by stimulating ORCC, supporting the idea that ATP controlled ORCC *via* a purinergic receptor.

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