Phenylalanine as a possible Treatment to Cystic Fibrosis

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Abstract

Cystic fibrosis (CF) is a genetic disorder that affects mostly the lungs, but also the pancreas, liver, kidneys, and intestine. Long-term issues include difficulty breathing and coughing up mucus as a result of frequent lung infections.

CF is inherited in an autosomal recessive manner. It is caused by the presence of mutations in both copies of the gene for the cystic fibrosis transmembrane conductance regulator (CFTR) protein. The most common mutation, ΔF508, is a deletion (Δ signifying deletion) of three nucleotides that results in a loss of the amino acid phenylalanine.

Phenylalanine (Phe or F) is an α-amino acid, it is found naturally in the breast milk of mammals. It is used in the manufacture of food and drink products and sold as a nutritional supplement for its reputed analgesic and antidepressant effects.

Not enough research has been made regarding the potential of CF treatment by supplementation of Phenylalanine, the amino acid absent in the encoded gene of the malfunctioning protein that is symptomatic to the disease

Key Words: Cystic Fibrosis; CF; Phenylalanine; CFTR protein; CFTR Gene

Cystic fibrosis (CF) is a genetic disorder that affects mostly the lungs, but also the pancreas, liver, kidneys, and intestine.

CF is inherited in an autosomal recessive manner. It is caused by the presence of mutations in both copies of the gene for the cystic fibrosis transmembrane conductance regulator (CFTR) protein. Those with a single working copy are carriers and otherwise mostly normal. CFTR is involved in production of sweat, digestive fluids, and mucus. When CFTR is not functional, secretions which are usually thin instead become thick. The condition is diagnosed by a sweat test and genetic testing. Screening of infants at birth takes place in some areas of the world.

The most common mutation in the CFTR protein, ΔF508, is a deletion (Δ signifying deletion) of three nucleotides that results in a loss of the amino acid phenylalanine (F) at the 508th position on the protein. This mutation accounts for two-thirds (66–70%) of CF cases worldwide.

Not enough research has been made regarding the potential of CF treatment by supplementation of Phenylalanine, the aminu acid absent in the encoded gene of the malfunctioning protein that is symptomatic to the disease.

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