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MUCOVISCIDOSIS A GENETIC DISEASE OF THE EXOCRINE GLANDS AND MANIFESTED AS A PATHOLOGY OF THE RESPIRATORY ORGANS

Boynazarov Abbosjon Anvarovich
Assistant of the Department Pediatrics
Central Asian Medical University

Abstract
Cystic fibrosis (CF), also called mucoviscidosis, is an autosomal recessive genetic disease that affects the exocrine function of the lungs, liver, pancreas, small bowel, sweat glands, and urogenital system. The condition causes thick mucus and other fluids to build up and clog different parts of the body, including the lungs and digestive tract. When mucus accumulates in the lungs of people with mucoviscidosis, it can cause frequent lung infections, which over time can cause severe damage to the lungs and inhibit breathing.

Keywords: Mucoviscidosis, viscous mucus, dry cough, pneumonia, chromosome 7, cystic fibrosis transmembrane regulator (CFTR) gene.

Introduction

Mucoviscidosis or cystic fibrosis is a disease based on genetic mutations. A dysfunction in certain glands causes the increased production of viscous mucus in the lungs, among other things. As a result of this mucus production, the organs are unable to function properly. Productive coughing is hindered as a result, patients suffer from shortness of breath and dry coughing, the susceptibility to respiratory tract infections such as bronchitis or pneumonia is increased. The life expectancy of affected people is still significantly lower than it is for healthy persons. However, with proper and timely treatment, patients can now live significantly longer lives than they could 20 years ago.

The cause of cystic fibrosis is a defect in chromosome 7. The affected gene is a protein that acts in the cell membrane as a chloride channel. Because of the gene defect, the protein is altered and the channel function is impaired. To date, more than 1000 different mutations of the CFTR gene are known. Cystic fibrosis only arises when both parents inherit a mutated gene. If both parents are carriers of one mutated and one healthy gene, the probability of a child receiving two intact gene copies is 25%. The probability that a child with one intact and one mutated copy is healthy, but can pass on the mutation is 50%, while the probability that the child is ill is 25%.

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If both parents are diseased, all children will inherit the illness. To date, science is unable to provide a conclusive explanation as to why a defect that engenders such a severe illness, is so prevalent and does not disappear over time. Therefore, a conclusion has been drawn that the cystic fibrosis illness must carry with it an evolutionary advantage. First, it was suspected that the disease progression of cholera is positively affected thanks to the gene defect. The dissemination of CF does not support this theory, however, as cystic fibrosis often appears in areas in which cholera is seldom diagnosed, and vice versa. A connection to typhoid was also suspected, but could not be proven. The bacterium depends on the CFTR channels in order to arrive in the intestinal cells. The most possible explanation is a protection against tuberculosis, which is accompanied by a predisposition to cystic fibrosis. This correlation has been relatively well demonstrated in clinical tests. The main symptoms of cystic fibrosis are a chronic dry cough, advancing shortness of breath, mucus in the lungs and pancreas, digestive disorders, all the way to delayed growth, intestinal obstructions, infertility, and jaundice. The symptoms are more or less pronounced depending on the severity of illness. Women can learn while pregnant whether their child is affected by CF, by undergoing a genetic analysis of the amniotic fluid. Later on, the illness can be diagnosed through sweat tests that point to elevated levels of salt in the sweat, and radiographic images that indicate blocked airways.

Cystic fibrosis is an autosomal recessive inherited genetic disorder that results from a homozygous defect of the cystic fibrosis transmembrane regulator (*CFTR*) gene on chromosome 7q31.2. The gene encodes for the corresponding CFTR protein, which regulates chloride ion transport across cell membranes. There are over 2000 mutations of the CTFR gene identified to date, with the most common being delF508 (deletion of the codon for phenylalanine at the 508 position) affecting 66% of cases. CFTR gene mutations are classified into 6 categories based on the synthesized protein structure and function as follows:

class I: mutations resulting in premature stop codons; CFTR expression is severely reduced or absent

class II: mutations resulting in CFTR misfolding and increased degradation; functional CFTR reacting the cell surface is reduced

class III: mutations that impair regulation of the CFTR channel; abnormal gating with reduced opening

class IV: mutations that impair CFTR ion conductance

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class V: mutations of CFTR promotor or splicing; CFTR protein is normal in structure and function, but reduced in number

class VI: mutations that reduce stability of CFTR; increased turnover of CFTR and reduced duration at the cell surface

The precise way in which CFTR mutations cause disease is complex and still under investigation. However, the most widely accepted explanation is that these mutations reduce extracellular chloride ion transport, leading to production of abnormally thickened mucus and eventual organ dysfunction. This is now known to be an oversimplification, as it has been shown that the CFTR protein also regulates the transport of sodium, bicarbonate, and glutathione. Unlike in other tissues, CFTR in sweat glands function in the opposite way, responsible for intracellular chloride ion transport. In patients with cystic fibrosis, the CFTR mutation results in excess chloride, sodium and fluid loss onto the skin surface, hence the use of the sweat test for diagnosis.

Early treatment is essential and responsible for the dramatic increase in life expectancy, now reaching 40 years or more; treatment options include dietary changes (pancreatic enzyme and vitamin supplementation), physiotherapy and airway clearance techniques, cystic fibrosis transmembrane regulator (CFTR) modulators such as ivacaftor-lumacaftor, ivacaftor-tezacaftor, and elexacaftor-tezacaftor-ivacaftor combination therapies for the delF508 mutation, ivacaftor monotherapy for the G551D mutation, anti-inflammatory therapy (e.g. azithromycin), prolonged courses of antibiotics with multiple agents, oral and inhaled corticosteroids, lung transplantation, and specific management of complications such as diabetes mellitus, hemoptysis, and distal intestinal obstruction syndrome (DIOS).

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