MOLECULAR-GENETIC ASPECTS OF COPD: CURRENT VIEW ON THE PROBLEM AND PROSPECTS FOR THE FUTURE

O. O. Krakhmalova, A. Y. Tokarieva

Abstract

According to current view chronic obstructive pulmonary disease (COPD) is a progressive heterogenous disease, characterized by persistent respiratory symptoms and air flow limitation due to anatomic abnormalities caused by hazardous exposure, often associated with co-morbidity, which significantly determines prognosis, severity of disease, treatment approach and rehabilitation program. Growing mortality due to COPD is associated with catastrophically worsening ecological conditions, widespread of smoking and harmful professional exposure.

The review presents data from recent studies, dedicated to genetics of multi-factor diseases. The study of the genetic factors of the inheritance of such diseases is complicated by the presence of a large number of genes with small effects involved in the formation of a predisposition to the disease.

The most important risk factor of COPD is smoking. However, only 10–20 % of chronic heavy smokers develop symptomatic COPD, which indicates that there are probably differences in susceptibility to the harm of tobacco smoke, which may be due to genetic factors.

Modern research confirms that COPD is a polygenic disease, which, in turn, leads to an incredible consistency of gene polymorphism in different populations. Currently, the pathogenesis of COPD is considered from the point of view of the theory of oxidative-antioxidant imbalance, the theory of imbalance of proteases-antiproteases and inflammation.

Along with the currently accepted genome-wide analysis of associations, studies of candidate genes, that is, genes whose products are potentially involved in the pathogenesis of the disease, remain important. An important role is played by the choice of genes - the components of a particular gene network, which controls and regulates the vital functions of the body, as well as the genes involved in individual biochemical and pathophysiological pathways and mechanisms.

Despite the fact that at the moment the influence of exogenous factors (tobacco smoke, industrial pollutants, etc.) is a determining risk factor for the development of COPD, literature data confirm that genetic predisposition plays an important role in this pathology. The hypothesis that changes in genes encoding enzymes that alter the redox environment of the lungs may contribute to the risk of COPD developing has been confirmed. The discovery of biomarkers of predisposition to the disease significantly expands the possibilities of its primary prevention and makes it possible to calculate the individual risk of COPD developing, which, in turn, will improve treatment. The search for new and research on already known genes that lead to the development of bronchopulmonary diseases in different populations remains an emerging problem. These areas need to be developed in the future for a deeper understanding of the problem and the search for new methods of treatment and prevention of COPD.

Key words: COPD, molecular-genetic study, gene polymorphism.

Ukr. Pulmonol. J. 2019;4:61-66.

Olena O. Krakhmalova

Institute of Therapy named after L.T. Malaya of National Academy of Medical Science of Ukraine

Head of Cardiopulmonology department

MD, Professor

2-A, L. Malaya Av., Kharkiv, 61039, Ukraine

Tel./fax: 380573739017, womanl@bigmir.net, www.krakhmalova.com