

## Discovery of genes that predispose a severe form of COPD

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A study by Ramcés Falfán-Valencia, researcher at the National Institute of Respiratory Diseases (INER), found that the mestizo Mexican population has a number of variations in certain genes that predispose them to develop the most severe form of chronic obstructive pulmonary disease (COPD).



This respiratory disease occurs mainly in people who have been exposed to cigarette smoke or fires in rural populations, where they use biomass for cooking.

"We found variations in the genome or mutations calls SNPs associated with <u>disease severity</u>, located in the genes of the receptor Interleukin (IL6R) involved in the inflammatory process of COPD and in the ADAM19 gene that is responsible for maintaining the structure of the lung firm," said the also member of the National System of Researchers.

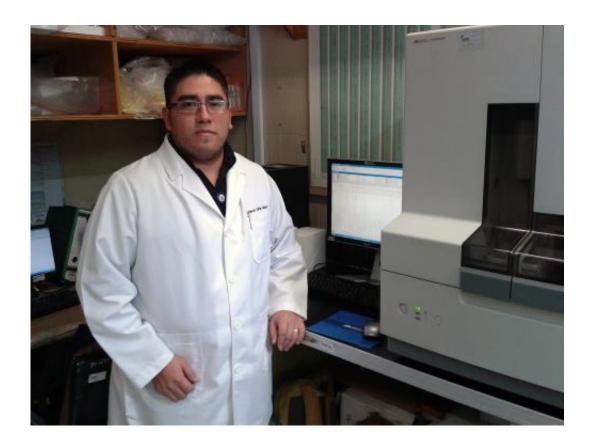
When suffering from COPD, the lung undergoes such deterioration that its architecture fragments and prevents gas exchange of carbon dioxide for oxygen, leaving no air available for the body, therefore no oxygen for the brain.

To understand why some patients reached a severe level and others remain in moderate one, a survey was conducted, were the results indicated that there are variations in the IL6R gene that lead to a medium to moderate disease, while the ADAM19 genetic factor was associated with individuals at severe levels.

"In the we lung have cells that are responsible for eliminating pathogens in every breath, but in patients who develop COPD it causes a more severe inflammation and a relentless disease develops, with which was initially a defense mechanism," explained the researcher at INER .

COPD is progressive and only partially reversible, while the disease worsens, the patient expresses increased difficulty breathing, coughing up phlegm and in chronic phases, the use of medical oxygen is vital in addition to drugs that decrease pain, as bronchodilators and anti-inflammatory pharmaceuticals, to survive.





Ramcés Falfán-Valencia

The research analyzed 299 patients with COPD, which were classified based on the severity of the disease, the international medical guide published in 2011 by the Global Initiative for Chronic Obstructive Lung Disease (GOLD for its acronym in English) was taken as reference.

According to the National Survey of Addictions (ENA) 2011 conducted by the National Institute of Public Health, if people start smoking at age 15 effects are seen after 50 years.

The goal of personalized genomics is to find more efficient therapeutic alternatives based on individual genetics. (Agencia ID)



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