Researchers at CREAL and UPF discover a common variant in the human genome that protects against asthma and obesity

- The first convincing evidence of the existence of a common genetic variant for asthma and obesity.
- Globally, following a study in 5,800 people, this genetic variant explains 40% of genetic “protection” from having asthma and obesity simultaneously.

Barcelona, 21 of February, 2014. Researchers from the Centre for Research in Environmental Epidemiology (CREAL), a CERCA Institute, and the Department of Experimental and Health Sciences (CEXS) of Pompeu Fabra University (UPF) have found that a variable region of the genome (an inversion, or part of the genome whose sequence is in reverse order to the normal, in 16p11.2) protects against developing two pathologies jointly: asthma and obesity. It is the first time convincing evidence of the existence of a common genetic variant for these two diseases has been reported.

Another novelty of this article, published in The American Journal of Human Genetics, is the use of new bioinformatics tools (inversion) that are capable of analysing the entire genome to identify regions with inversions and to analyse their role in relation to common disease using existing data from previously studied individuals. Inversions are alterations in the genome that may or may not be pathologic.

According to Dr. Juan Ramón González, a researcher at CREAL, an ISGlobal Alliance research centre, and a bioinformatics expert, "Until now, these studies have been very costly because there were no methods for analysing genomic inversions on a massive scale in large populations”. On this occasion, data from a total of 5,800 people in Europe, Asia, Africa and America were analysed.

Differences between continents

The results show that the genomic region analysed varies according to the continent from which the person comes. "This is an example of how changes in the genome can be selected depending on how humans adapt to their environment, in this case, their metabolic needs in relation to climate," says Dr. Luis Pérez-Jurado, a researcher from the Genetics Unit and the Department of Experimental and Health Sciences at Pompeu Fabra University (UPF), and the Network Centre for Biomedical Research on Rare Diseases (CIBERER).

Specifically, only 10% of the population of East Africa has this genomic inversion and 50% of the population of northern Europe (where it is assumed that this alteration has been selected for better adaptation to the cold climate, which requires a higher basal metabolic rate).

Overall, after studying 5,800 people, this genetic variant explains 40% of the genetic "protection or predisposition” to developing asthma and obesity simultaneously.

"We are currently using the methods we have developed to study the role of over a hundred inversions in other diseases. Specifically, we are now studying some inversions that seem to predispose to developing autism or intellectual disabilities, among others”, says the CREAL researcher.

Reference to the article: A Common 16p11.2 Inversion Underlies the Joint Susceptibility to Asthma and Obesity. Juan R. González, Alejandro Cáceres, Tonu Esko, Ivan Cuscó, Marta Puig, Mikel Esnalol, Judith Reina, Valerie Siroux, Emmanuelle Bouzigon, Rachel Nadif, Eva Reinmaa, Lili Milani, Mariona Bustamante, Deborah Jarvi, Josep M. Antó, Jordi Sunyer, Florence
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