

European Respiratory Society Annual Congress 2013

Abstract Number: 5224
Publication Number: P1400

Abstract Group: 12.3. Genetics and Genomics

Keyword 1: COPD - mechanism **Keyword 2:** Genetics **Keyword 3:** No keyword

Title: Haplotypes associated with COPD and its severity in Mexican mestizo population

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Body: Background: COPD is a complex disease whose main risk factor is cigarette smoking, there is genetic component that predisposes to develop it; the objective of this study is to evaluate haplotypes associated with COPD and its severity in Mexican mestizo population. Methods: We conducted a study with 299 patients with COPD (P-COPD) secondary to smoking and 531 smokers without COPD (SW-C), P-COPD were classified according to the Global Initiative for Chronic Obstructive Lung Disease (GOLD) 2010, in 2 groups, GOLD I+II and III+IV. It was designed a microarray (Illumina, CA, USA) with 1536 single nucleotide polymorphisms (SNPs); 251 were ancestry informative markers (AIMs), the rest were distributed in 24 candidate genomic regions. To describe the study population used SPSS; population stratification was performed by EIGENSOFT software, the structure of linkage disequilibrium and haplotype generation was performed using the software Haploview 4.2. Results: We find haplotypes associated with COPD in 3 genes: FAM69A, ZNF583 and SLC9A8.

Haplotypes associated

Chr	Gene	Haplotype	HF P-COPD	HF SW-C	p	OR	CI (95%)
1	FAM69A	GAGGC	0.729	0.662	0.0047	1.38	1.10-1.73
1	FAM69A	GAGGG	0.021	0.056	0.0008	0.35	0.18-0.67
19	ZNF583	AAAGGGAACAG	0.523	0.364	3.97E-10	1.91	1.55-2.36
19	ZNF583	AAAGAGAACAG	0.108	0.271	5.75E-15	0.32	0.24-0.44
20	SLC9A8	AAAG	0.151	0.079	5.60E-06	2.06	1.48-2.86
20	SLC9A8	ACAG	0.007	0.061	1.29E-07	0.10	0.03-0.28

Analysis according GOLD, shows an haplotype in IL6R gene (I+II vs III+IV) associated to protection ($p = 8.00E-03$) and other to susceptibility in ADAM19 ($p = 7.40E-03$), both genes encoding proteins involved in the inflammation process. Conclusions: There are haplotypes associated with COPD and GOLD severity in Mexican mestizo population.