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Title: Case control study of chronic obstructive pulmonary disease and its progression

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Body: Chronic obstructive pulmonary disease is a multifactorial disorder caused by environmental determinants and genetic risk factors. PCR-RFLP analysis was performed for 391 cases and 514 control individuals from Ufa (Russia) to analyze the contribution of 55 polymorphisms of the genes involved in extracellular matrix remodeling, xenobiotic metabolism, antioxidant and inflammation pathways to COPD manifestation and progression. We investigated the gender-dependent differences for susceptibility to COPD. Our results showed that the CYP1A1 rs4646903 (P=0.0069), IL6 rs1800795 (P=0.021), SERPINA3 rs4934 (P=0.016) were associated with COPD only in female patients. In man cases, the NQO1 rs1131341 (P=0.0002), MMP3 rs35068180 (P=0.001), CYP2A6 rs71790353 (P=0.000001), CYP2S1 rs338583 (P=0.0008) CAT rs1001179 (P=0.0069), GSTP1 rs1695 (P=0.0007) were associated with COPD. The CYP2A6 rs71790353, NQO1 rs1131341, EPHX1 rs1051740, SOD3 rs1799895, TIMP2 rs8179090, ILRN rs71941886, VDBP rs4588 were associated with early-onset COPD. We found a significant interaction of the smoking status and CYP1A1 rs4646903 (P=0.047), CYP2F1 rs11399890 (P=0.029), CAT rs1001179 (P=0.0082), VDBP rs7041 (P=0.027), ADAM33 rs2280091 (P=0.0068) in patients with early-onset COPD. The severity of COPD was modified by MMP9 rs3918242 (P=0.023), LTA rs909253 (P=0.0073), IL6 rs1800795 (P=0.015), CYP2E1 rs2031920 (P=0.015). The relationship between the ADAM33 rs2280091 (P=0.02), UGT2B7 rs7439366 (P=0.0007), IL1RN rs71941886 (P=0.034), VDBP rs4588 (P=0.0064), IL6 rs1800795 (P=0.028) and emphysema risk was found. These results further contribute to the understanding of hereditary predisposition to COPD.