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Title: Case-control association analysis of candidate genes in asthma, rhinitis and COPD: A preliminary report

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Body: This study aims to determine the genetic involvement in the susceptibility to asthma, rhinitis and COPD, by candidate gene association analysis, in a large and accurately defined series of Italian subjects, even considering exposure to some environmental contexts and life-styles. The study population included 1075 subjects (aged 20-66 years) from the general population, enrolled in the frame of the Gene Environment Interactions in Respiratory Diseases (GEIRD) study between 2007 and 2010. Cases and controls were diagnosed during a clinical examination that included a detailed interview, pre/post bronchodilator spirometry, methacoline challenge, skin prick tests. A panel of 384 Single Nucleotide Polymorphisms (Tag-SNP), representative of 63 candidate genes with a previous indication of possible association to the studied diseases, was genotyped by a customized GoldenGate Genotyping assay. Presently, genotyping of 725/1075 subjects are completed. A preliminary association study of candidate gene polymorphisms was conducted on these data, for the susceptibility to one or more of the studied phenotypes, by basic association test based on allele frequency comparison. Presence of association (unadjusted p<0.005) was observed between GSTP1 and non-atopic rhinitis, PDE4D and ever asthma with atopy, IL13 and past-asthma, TNS1 and chronic bronchitis. Moreover, a possible association (unadjusted p<0.02) was also found for IL1RL2 with ever asthma, chronic bronchitis, atopic rhinitis and non atopic rhinitis. The analysis is going on to complete the genotyping of all the enrolled subjects and to perform haplotype analysis, to confirm the involvement of these genes in the studied diseases.