Abstract Group: 12.3. Genetics and Genomics

Keyword 1: Genetics  Keyword 2: Asthma - mechanism  Keyword 3: Chronic disease

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

Dr. Cristina 20378 Bombieri cristina.bombieri@univr.it 1, Dr. Anna Rita 20385 Lo Presti annarita.lopresti@univr.it 1, Dr. Francesca 20387 Belpinati francesca.belpinati@univr.it 1, Dr. Simone 20388 Accordini simone.accordini@univr.it 2, Dr. Alessandro 20390 Baldan alessandro.baldan@gmail.com 1, Prof. Marcello 20396 Ferrari marcello.ferrari@univr.it MD 3, Dr. Giovanni 20397 Malerba giovanni.malerba@univr.it 1, Prof. Elisabetta 20399 Zanolin elisabetta.zanolin@univr.it 2, Prof. Pier Franco 20400 Pignatti pierfranco.pignatti@univr.it MD 1 and Prof. Roberto 20401 de Marco roberto.demarco@univr.it 2, 1 Dpt. of Life and Reproduction Sciences, Section of Biology and Genetics, University of Verona, Italy ; 2 Dpt. of Medicine and Public Health, Section of Epidemiology and Medical Statistics, University of Verona, Italy and 3 Dpt. of Medicine, Section of Internal Medicine D, University of Verona, Italy.

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.