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Title: Association study of ANXA11-R230C with sarcoidosis

Fotini 18196 Bardaka bardakafotini@yahoo.gr MD [1,2], Zoe 18223 Daniil zdaniil@med.uth.gr MD [1], Katerina M. 18224 Antoniou katerinaantoniou@yahoo.gr MD [3], Antigoni 18225 Mamara antigoneta@gmail.com MD [2], Foteini 18226 Malli mallifoteini@yahoo.gr MD [1], Konstantinos I. 20805 Gourgoulianis kgourg@med.uth.gr MD [1], Anastasios E. 20841 Germenis agermen@med.uth.gr MD [2] and Matthaios 20844 Speletas maspel@med.uth.gr MD [2]. [1] Respiratory Medicine Department, University of Thessaly, Larissa, Greece, 41110 ; [2] Laboratory of Immunology and Histocompatibility, University of Thessaly, Larissa, Greece, 41110 and [3] Respiratory Medicine Department, University of Crete, Heraklion, Greece, 71003 .

Body: Introduction: Recently, a genome wide association study demonstrates that the polymorphism ANXA11-R230C (rs 1049550) is strongly associated with sarcoidosis, a chronic granulomatous disease (GD) of unknown etiology. Aim: Considering that also other diseases are accompanied by granulomas formation, as observed in 10% of patients with common variable immunodeficiency (CVID), the purpose of this study was to confirm the association of ANXA11-R230C with sarcoidosis and to examine its possible association with GD in CVID. Methods: DNA was extracted from peripheral blood of 71 patients with sarcoidosis (M/F:24/47, mean age: 52.5 years, range:20-75), 101 normal individuals (M/F:35/66, mean age: 54.1 years, range:19-76), and 19 CVID patients (M/F:7/12, mean age: 28.1 years, range: 2-60), two of which had granulomatous disease (one with familial CVID history including a sister without GD). A PCR-RFLP protocol was designed to detect ANXA11-R230C and statistical analysis was performed using the SPSS software (ver.-10.0).Results: The difference in the allele frequency of ANXA11-R230C between patients with sarcoidosis and healthy individuals was not marginally reached to be significant (p=0.073). The allele frequency of ANXA11-R230C in CVID patients was 50.0% and no significant difference between patients with and without GD was observed. However, in the family with CVID, the member with GD carried only the R alleles, associated with granulomas formation, while her sister was heterozygous. Conclusions: The association of ANXA11-R230C with sarcoidosis was not confirmed, while the emerged contribution of this polymorphism in the granoulomas formation in CVID needs to be further clarified.