

European Respiratory Society Annual Congress 2012

Abstract Number: 1422

Publication Number: P777

Abstract Group: 3.1. Molecular Pathology and Functional Genomics

Keyword 1: Sarcoidosis **Keyword 2:** Genetics **Keyword 3:** Immunology

Title: Is a functional variant of ANXA11 R230C associated with impaired apoptosis? Pilot data

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Body: Sarcoidosis is a granulomatous disorder of an unknown etiology, where the granuloma formation has been associated with impaired apoptosis of activated inflammatory cells. There is no information concerning the influence of ANXA11 R230C, a functional annexin A11 variant associated with protection/disease modification of sarcoidosis, on the sensitivity of peripheral blood mononuclear cells (PBMC) to apoptosis yet. We therefore compared the sensitivity to apoptosis of PBMC obtained from 30 sarcoid patients and 9 healthy controls. Tributyltin was used as apoptosis stimulus; annexin V positive cells were detected by flow cytometry. Pilot analysis was performed in subgroups according to ANXA11 R230C (rs1049550) genotype (TT, n=7; CC, n=7). When compared to healthy controls, lower number of annexin V positive cells was detected in tributyltin-stimulated PBMC from sarcoid patients (mean 55.9%; range 23.3–77.2%) than in cells from controls (64.8%; 54.7–74.1%; p=0.01). After subdivision according to genotypes, the number of annexin V positive cells did not differ between patients carrying TT genotype (50.8%; 23.3–58.8%) and those with CC genotype (54.5%; 41.7–71.7%; p>0.05). Genotyping and investigations on association between apoptosis and genotypes in larger patient/control groups are under progress. In conclusion, PBMC obtained from sarcoidosis patients showed more apoptosis resistant phenotype than cells from control subjects. In our pilot cohort, wild type ANXA11 genotype and its R230C variant did not differ in terms of sensitivity to apoptosis. Further studies in a larger patient cohort are ongoing. Grant support: IGA MZ CR NS/11117, IGA_PU_LF_2012_07, CZ.1.05/2.1.00/01.0030.