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and F19Y mutant galectin-8 from peripheral blood mononuclear cell lysates of healthy individuals with different genotypes as well as with recombinant wild-type and F19Y mutant galectin-8 proteins. Furthermore, we evaluated the association of regulatory region polymorphisms of the LGALS1 (Rs4820293, Rs4820294) and IL2Rβ (Rs743777, Rs228941) genes in 146 Caucasian myasthenia gravis patients compared to 291 ethnically matched controls.

Results We found a strong association of the F19Y galectin 8 gene polymorphism with rheumatoid arthritis, and a mild one with myasthenia gravis. Moreover, the polymorphism also correlated with age at disease onset in the case of rheumatoid arthritis. The F19Y substitution did not appear to affect carbohydrate binding in solid-phase assays markedly. Also, a significant difference was found in the distribution of the Rs4820293/Rs743777 polymorphism haplotypes ($p < 0.01$) in patients with myasthenia gravis and controls but not in rheumatoid arthritis. The Rs4820293 polymorphism of LGALS1, previously not described to be associated with any disease, did not affect LGALS1 expression in peripheral mononuclear cells and skeletal muscle.

Conclusions This is the first study of an association between a galectin-based polymorphisms leading to a mutant protein and autoimmune diseases, with evidence for antagonistic pleiotropy.

A7.4 ASSOCIATION OF GALECTIN SINGLE NUCLEOTIDE POLYMORPHISMS WITH AUTOIMMUNE DISEASES

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Background and Objectives Galectins are potent immune regulators. Surprisingly, genetic association of galectin genes with autoimmune diseases have not yet been studied. A polymorphism in the coding region of the galectin-8 gene (Rs2737713; F19Y) and a novel galectin-1 and interleukin 2 receptor β haplotype were investigated for association with rheumatoid arthritis and myasthenia gravis.

Materials and Methods A case-control analysis and a related quantitative trait-association study were performed to investigate the association of the galectin 8 gene polymorphism in patients (myasthenia gravis 149, rheumatoid arthritis 214 and 134 as primary and repetitive cohorts, respectively) and 365 ethnically matched (Caucasian) healthy controls. Distribution was also investigated in patients grouped according to their antibody status and age at disease onset. Comparative testing for lectin activity was carried out in ELISA/ELLA-based binding tests with both wild-type

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A7.4 Association of Galectin Single Nucleotide Polymorphisms with Autoimmune Diseases

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