A44 POLYMORPHISMS IN THE INTERLEUKIN 4, INTERLEUKIN 13 AND CORRESPONDING RECEPTOR GENES ARE NOT ASSOCIATED WITH SYSTEMIC SCLEROSIS AND DO NOT INFLUENCE GENE EXPRESSION

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Aim Polymorphisms in the interleukin 4 (IL-4), IL-13 and their corresponding receptors have previously been found associated with systemic sclerosis (SSc). In this study the authors aim to validate these previous observations and scrutinise their effects on gene expression.

Patients and methods The authors genotyped a cohort consisting of 2488 SSc patients and 2246 healthy controls, derived from The Netherlands, Spain, UK, Italy, Germany

and France. Taqman assays were used for genotyping two single-nucleotide polymorphisms (SNPs) within *IL-4* (*Q576R*/rs1801275) and the *IL-4* α *receptor* (*–590C/T*/rs2243250). In the *IL-13* gene two SNPs were genotyped *R130Q* (rs20541) and *–1112C/T* (rs1800925.) In the *IL-13* α *receptor* gene, the *43163:G/A* (rs6646259) variant was genotyped. In addition, the authors investigated the effect of these polymorphisms on corresponding gene expression with RT-PCR in B cells, T cells, plasmacytoid dendritic cells, monocytes and myeloid dendritic cells.

Results None of these polymorphisms was found to be enriched in the SSc population or in any SSc clinical subtype and there was no influence of these polymorphisms on development of either pulmonary arterial hypertension and declineer of forced vital capacity in 15 years of follow-up. In addition, the authors did not observe an effect on expression levels in the cell subtypes.

Conclusions This data show that these polymorphisms do not play a role in SSc and do not influence gene expression levels.