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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 (IL4), interleukin 13 (IL13) and their corresponding receptors have previously been found associated with systemic sclerosis (SSc). In this study we aim to validate these previous observations and scrutinize their effects on gene expression.

Patients and methods

We genotyped a cohort consisting of 1902 systemic sclerosis patients and 1503 healthy controls, derived from France, The Netherlands, Spain, United Kingdom, Italy and Germany. Taqman assays were used for genotyping three SNPs correlating with IL-4 and receptor; interleukin 4 alpha receptor Q576R (rs1801275), interleukin 4 RI75V (rs1805010), and −590C/T (rs2243250). In the Il-13 gene the following SNPs were genotyped; R130Q (rs20541), (-1112C/T), rs1800925 and rs6646259 (base 43163:G/A). In addition, we 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. In addition, we did not observe an effect on expression levels in the cell subtypes.

Conclusions

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

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