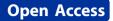


POSTER PRESENTATION



Polymorphisms in the interleukin 4, interleukin 13 and corresponding receptor genes are not associated with Systemic Sclerosis and do not influence gene expression

JCA Broen^{1*}, P Dieude², M C Vonk¹, L Beretta³, B Rueda⁴, A Herrick⁵, J Worthington⁵, N Hunzelmann⁶, G Riemekasten⁷, H Kiener⁸, R Scorza³, C P Simeon⁹, V Fonollosa⁹, P Carreira¹⁰, N Ortego-Centeno¹¹, M A Gonzalez-Gay¹², P Airò¹³, MJH Coenen¹⁴, A Aliprantis¹⁵, J Martin^{4†}, Y Allanore^{16†}, TRDJ Radstake^{1†}

From 5th European Workshop on Immune-Mediated Inflammatory Diseases Sitges-Barcelona, Spain. 1-3 December 2010

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 II-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.

+ Contributed equally

¹Dept. of Rheumatology, Radboud University Nijmegen Medical Center, The Netherlands

BioMed Central © 2010 Broen et al; licensee BioMed Central Ltd.

Full list of author information is available at the end of the article

Conclusions

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

Author details

Dept. of Rheumatology, Radboud University Nijmegen Medical Center, The Netherlands. ²Université Diderot Paris 7, Service de Rhumatologie, Hospital Bichat Claude Bernard, Paris, France. ³Referral Center for Systemic Autoimmune Diseases, University of Milan, Italy. ⁴Instituto de Parasitología y Biomedicina, CSIC, Granada, Spain. ⁵Rheumatic Diseases Centre, University of Manchester, Salford Royal NHS Foundation Trust, UK. ⁶Dept. of Dermatology, University of Cologne, Germany. ⁷Dept. of Rheumatology and Clinical Immunology, Charité University Hospital and German Rheumatism Research Centre, a Leibniz institute. ⁸Dept. of Internal Medicine, Division of Rheumatology, University of Vienna, Austria. ⁹Servicio de Medicina Interna, Hospital Vall d'Hebron, Barcelona, Spain. ¹⁰Servicio de Reumatologia, Hospital 12 de Octubre, Madrid, Spain. ¹¹Servicio de Medicina Interna, Hospital Xeral-Calde, Lugo, Spain. ¹²Servicio de Reumatologia, Hospital Marques de Valdecillas, Santander, Spain. ¹³Servizio di Reumatologia ed Immunologia Clinica, Spedali Civili, Brescia, Italia.¹⁴Dept. of Human Genetics, Radboud University Nijmegen Medical Center, The Netherlands. ¹⁵Dept. of Immunology and Infectious Diseases, Harvard School of Public Health, Boston, MA, USA. ¹⁶Université Paris Descartes, INSERM U781, Hôpital Necker, Paris, France and Université Paris Descartes, Service de Rhumatologie A, Hôpital Cochin, Paris, France.

Published: 25 November 2010

doi:10.1186/1479-5876-8-S1-P47 Cite this article as: Broen *et al.*: Polymorphisms in the interleukin 4, interleukin 13 and corresponding receptor genes are not associated with Systemic Sclerosis and do not influence gene expression. *Journal of Translational Medicine* 2010 8(Suppl 1):P47.