

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, Philippe Dieude, M Vonk, L Beretta, B Rueda, A Herrick, J Worthington, N Hunzelmann, G Riemekasten, H Kiener, et al.

► **To cite this version:**

Jca Broen, Philippe Dieude, M Vonk, L Beretta, B Rueda, 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. 5th European Workshop on Immune-Mediated Inflammatory Diseases, Dec 2010, Sitges-Barcelona, Spain. BioMed Central, 8 (Suppl 1), pp.P47, 2010, Journal of Translational Medicine. <10.1186/1479-5876-8-S1-P47>. <inserm-00663872>

HAL Id: inserm-00663872

<http://www.hal.inserm.fr/inserm-00663872>

Submitted on 27 Jan 2012

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



POSTER PRESENTATION

Open Access

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 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.

Author details

¹Dept. of Rheumatology, Radboud University Nijmegen Medical Center, The Netherlands. ²Université Diderot Paris 7, Service de Rhumatologie, Hôpital 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 Reumatología, Hospital 12 de Octubre, Madrid, Spain. ¹¹Servicio de Medicina Interna, Hospital Xeral-Calde, Lugo, Spain. ¹²Servicio de Reumatología, 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.

† Contributed equally

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

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