A67 PTPN22 R620W MAY AFFECT AGE AT ONSET AND SURVIVAL OF SSC PATIENTS

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Background and objectives The PTPN22 SNP rs2476601 (1858C→T, R620W) shows association with several autoimmune diseases including SSc. Here we analyse data from SSc

patients concerning the time-dependent development of the disease from early to late stages, dependent on their R620W genotype.

Materials and methods SSc patients (N=235, 81.3% female) were recruited in the University Medical Centers in Freiburg and Cologne. Genomic DNA was purified from peripheral blood. Genotyping was done applying single base extension and MALDI-TOF mass spectrometry (genoSNIP; Bruker Daltonics, Leipzig, Germany) or PCR and RFLP. Statistic analysis was performed using PASW Statistics 18.0.

Results The means of age at onset of Raynaud phenomenon, skin involvement and organ involvement of all patients resembled published data.[1] Of 232 patients genotyped, 19.8% carried the minor T allele and 80.2% did not. T-neg patients were older at onset of RP, skin and organ involvement than T-pos patients (RP: N=169, 47.3±14.7 years vs N=42, 41.3±14.6 years; skin involvement: N=162, 51.4±13.4 years vs N=40, 44.9±13.6 years; organ involvement: N=163, 53.5±12.9 years). These differences were significant for RP (p=0.019) and skin involvement (p=0.006). From 183 patients data on age at onset of RP, skin involvement and organ involvement were available, of whom 157 (85.8%) started with RP, followed by skin involvement and later on organ involvement. Also in this group, T-neg patients were older than T-pos. patients (RP: 48.5± 4.2 vs 40.6±14.3, p=0.004; skin involvement: 51.3±12.9 vs 44.8±13.8, p=0.011; organ involvement: 54.1±12.6 vs 49.1±14.0, p=0.048).

In addition, we analysed the frequencies of T-pos patients dependent on the observation time between the age at last contact and the age at onset of RP (A), skin (B) or organ (C) involvement, again in the group of 157 patients. In all three comparisons (A, B and C) the percentages of T-pos patients increased continuously from short to long observation periods, indicating a possible positive effect of the presence of the minor T-allele on survival.

Conclusions T-pos patients develop disease at an earlier age, but show no difference in progression time. The frequency of T-pos patients was higher in groups of patients observed for a longer period of time, implicating a longer survival period.

REFERENCE

1. **Hunzelmann N**, Genth E, Krieg T, *et al*. The registry of the German Network for Systemic Scleroderma: frequency of disease subsets and patterns of organ involvement. *Rheumatology* 2008;**47**:1185–92.