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Association of HLA-DRB1 and DQB1 alleles and haplotypes with rheumatoid arthritis in a Pakistani population

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Introduction & Aim: Rheumatoid arthritis is an autoimmune disease with poorly understood pathophysiology. Genetic components of disease etiology, especially human leukocyte antigen (HLA) associations are well known. Ethnic differences account for a number of variations in disease association with the HLA locus and there seem to be differences in various studies regarding its genetic predisposition. This study was aimed at determining the contribution of DRB1 and DQB1 components of HLA class II in rheumatoid arthritis in a Pakistani cohort.

Method: For this study, 110 patients and 120 healthy controls from the same geographical area and matched ethnicity were enrolled. Blood DNA was isolated from all the subjects and HLA alleles were typed following allele specific amplification. Subsequently, haplotypes were generated and allelic and haplotype distribution frequencies were compared among the patients and controls using χ^2 and Arlequin software. The data obtained by this analysis were also compared with other reported associations found in the Pakistani population by meta-analysis.

Results: HLA allelic status was determined among the patients and controls from the same geographical area to account for differences in ethnicity and environmental factors. Significant associations were found for alleles as well as haplotypes among the patients of rheumatoid arthritis. DRB1*10, DQB1*05 and DQB1*602 were found to be associated with disease susceptibility, whereas DRB1*11 and DQB1*02 had protective effect against the disease. Similarly, haplotype DRB1*10-DQB1*05 was associated disease risk, whereas DRB1*07-DQB1*02 and DRB1*11-DQB1*0301 had a protective effect.

Conclusion: There is a significant DRB1 and DQB1 allele and haplotype association with rheumatoid arthritis susceptibility and protection.

Biography

Ambreen Gul Muazzam has obtained the Certificate in Life Science Enterprise (LSE) from University of Toronto, School of Continuing studies, Mississauga campus, Ontario, Canada. She has joined the Institute of Biomedical and Genetic Engineering (IB&GE) in 2003 as Scientific Officer after receiving her Master's degree in Biochemistry and Molecular Biology from Quaid-i-Azam University, Pakistan. She was then promoted to the post of Senior Scientist in the year 2008. Concurrently, she has completed her MPhil from Quaid-i-Azam University, Islamabad, Pakistan. She has published papers in international journals. Her research interests include population and disease genetics. Her particular research focus is on diagnostics and molecular studies of hepatitis B and C as it is one of the major health problems that Pakistan is facing. She is also involved in cell culturing and cytotoxicity of nanoparticles and organic compounds against human cancerous cell lines. She has also experience working in the Quality Control Department of Pharmaceutical Company as a Microbiologist.

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