GENETIC PROFILING IN TAKAYASU ARTERITIS: EXPLORING HLA LOCI AS SUSCEPTIBILITY AND PROGNOSTIC MARKERS

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Takayasu arteritis (TA) is a large-vessel vasculitis with an as-yet undefined etiopathogenesis, though immunogenetic factors, particularly HLA polymorphisms, are considered to play an important role. This study investigates the distribution of selected HLA class I and II alleles in individuals diagnosed with TA and assesses their correlation with clinical phenotypes, age at onset, and treatment response.

HLA genotyping was performed in a cohort of 25 unrelated TA patients, focusing on HLA-A, -B, -C, -DRB1, and -DQB1 loci. Allele and haplotype frequencies were compared to two control populations: one comprising 1992 individuals (for HLA-A, -B, -DRB1), and the other 159 unrelated healthy donors (for HLA-C, -DQB1).

The HLA-B*52 allele was significantly overrepresented in the TA group (10% vs 1.2% in controls; p=0.0004), and its presence was linked to more aggressive disease course, earlier onset, and suboptimal treatment response. Although increased frequencies of HLA-A*32, B*15, B*57, and the DRB1*15-DQB1*05 haplotype were also observed among patients, these associations did not retain significance after correction for multiple testing. Notably, HLA-C*03 was found in nearly one-third of patients, exclusively among those with milder clinical profiles, suggesting a potential protective immunogenetic signature.

Our data reinforce the relevance of HLA-B*52 in disease susceptibility and prognosis in TA. While additional alleles and haplotypes showed suggestive trends, their role warrants further exploration in larger cohorts. The possible protective influence of HLA-C03 represents an intriguing avenue for future investigation.

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