

HUMAN LEUKOCYTE ANTIGENS AND GAUCHER DISEASE: IS THERE AN ASSOCIATION?

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Background: Gaucher disease (GD) is caused by the reduced activity of lysosomal enzyme glucocerebrosidase, which leads to the accumulation of glucocerebroside in cells and a chronic stimulation of the immune system. GD is divided into 3 main types according to the presence or absence of neurological involvement and to its presentation (acute or chronic). Gaucher cells show an increase in their expression of HLA-DR antigens on their surface, and there is an increase in levels of antigen-presenting molecules. Over 100 diseases have already been associated to HLA antigens; however, this association has never been studied in GD. Objectives: To analyze the variability of HLA genes in a Southern Brazilian sample of GD patients, to compare it with controls, and to look for associations with clinical manifestations. Methodology: Thirty-one GD patients (24 mild, 4 moderate, and 3 severe) were included in the study. They were typed for HLA A, B, and DR and compared to 250 healthy controls. The clinical data were obtained from the review of medical records. Results/Discussion: There was a significant difference in the frequency of B37 allele among patients when compared to controls ($p=0.011$, OR 13.28). An association was found between DR11 ($p=0.008$) and DR13 ($p=0.011$) alleles and the severity of the disease. DR11 allele seems to be associated to neurologic compromise, while DR13 seems to be associated to osteonecrosis. Conclusion: Our data suggest a possible association of HLA variants and GD. The HLA variants must be further studied, for they seem to be a phenotype-modifier factor for GD.