

Genotype and phenotype correlations in diabetic patients in Uruguay

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ABSTRACT. To differentiate among different types of diabetes is becoming an increasingly challenging task. We investigated whether the patient's genetic profile is useful to identify the particular type of diabetes, to determine the corresponding hyperglycemia pathogenesis and treat accordingly. Three hundred and thirty-eight diabetic patients, diagnosed according to American Diabetes Association criteria, were recruited from 2004 to 2008 in diabetes health reference centers. We analyzed the major gene for type 1 diabetes susceptibility (HLA DQ/DR). In order to improve our understanding of the pathogenesis of the resulting hyperglycemia and to implement a more adequate treatment for the patients, we reclassified our sample ac-

ording to the presence or absence of the genetic markers. We found that a higher percentage of people than expected have immunological disease, independent of their phenotype, with a relative risk of 4.62 (95% confidence interval). This methodology allowed us to establish an association between the genotype and its resulting phenotype. We found significant differences; the phenotypic classification did not reflect immunological disease based on genotype. Moreover, when we examined markers, body mass index and age of onset, we found that many people have an intermediate phenotype between type 1 and type 2. This genetic data can help provide an accurate definition of the disease and would therefore provide the physician a better possibility of providing adequate treatment.

Key words: Antibodies; Diabetes mellitus; HLA