Amerindian mtDNA haplogroups and celiac disease risk HLA haplotypes in mixed-blood Latin American patients

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Background and Objective: Risk haplotypes have been described in celiac disease (CD), but the influence of native genes on CD in Hispanic Americans is unknown. The aim of the study was to measure the frequency of Amerindian mitochondrial DNA (mtDNA) haplogroups (inherited by the maternal line) in mixed-blood patients with CD from Chile, Argentina, and Uruguay, and to assess the relation between these and human leukocyte antigen (HLA) alleles and haplotypes and clinical presentations. Patients and Methods: Clinical history, histological data, and genetic studies were conducted following 2 protocols: a case-control study of 72 Chilean patients with CD and controls, and an assessment of 43 (additional) samples of celiac patients from Chile, 96 from Argentina, and 57 from Uruguay, compared with the mtDNA frequency in the corresponding country. HLA typing was performed by a commercial kit, and mtDNA was determined by means of polymerase chain reaction and restriction fragment length polymor