

Letters to the Editor

Absence of HLA-DQ2 and HLA-DQ8 does not exclude celiac disease in Brazilian patients

Key words: Celiac disease. HLA. Brazilian population. HLA-DQ2 antigen. HLA DQ-8 antigen.

Dear Editor,

The genetic predisposition is well known in celiac disease (CD). In a recent review, Vives et al. (1) related that about 90 % of CD patients expressed HLA-DQ2, and 5 % of them have the HLA-DQ8. The remaining 5 % of CD patients have one of the two genes that form the HLA-DQ2 heterodimer but do not present the full heterodimer. HLA-related genes are necessary but not sufficient to cause the disease. On the other hand, HLA class II DQ2 and DQ8 are responsible for approximately 40 % of genetic risk to CD. Also, their absence has a high negative predictive value and, in the clinical practice, could exclude a diagnosis of CD (1,2). However, is it really true for a population with a high admixture of races as Brazilian population?

The present study investigated CD patients from Southern Brazilian population, which was researched HLA-DQ2 and HLA-DQ8 alleles. A total of 101 consecutive patients, composed of 83 female and 18 male subjects, with median age of 30 years (rank: 3 to 87 years), all auto-declared with European ascendancy, were evaluated and were diagnosed using positive serological tests (IgA antiendomysium) and confirmed using histological findings in duodenal biopsies (Marsh classification). HLA-DQ2 and HLA-DQ8 presence was typed using DNA amplified by polymerase chain reaction. The presence of

HLA-DQ2 was observed in 80 (79.2 %) of the 101 celiac patients and HLA-DQ8 in 26 (25.7 %). In 101 cases, HLA-DQ2 and HLA-DQ8 were concomitantly detected in 14 (13.9 %), HLA-DQ2 positive was found in 66 (65.3 %), and HLA-DQ8 positive was observed in 12 (11.9 %). Nine patients (8.9 %) did not present HLA-DQ2 and/or HLA-DQ8 positive. All these cases, in spite of HLA-DQ2 HLA-DQ8 being both negative, had clinical evidence, positive serology, and confirmed CD diagnosis after duodenal biopsies. Besides, recovery was documented after following a gluten-free diet-based treatment. Martins et al. (3) investigating 90 Brazilian celiac patients in Brasilia (Mild West Region) showed that with one exception (1.1 %), all the patients carried DQ2 and/or DQ8 alleles. Castro-Nunes et al. (4), studying 73 patients with CD from Northeastern Brazil, described 50 (68.5 %) with genotype DQ2, 13 (17.8 %) with DQ8, five (6.8 %) with DQ2 and DQ8 concomitantly, and five (6.8 %) without any of these genotypes. Similar observations have been reported from European (5) and American populations (6). Latin American celiac patients (Argentina, Chile and Uruguay) also showed 7 % of negative results for DQ2 and/or DQ8 in a multicenter report (7).

Concluding, in the present study, HLA-DQ2 and HLA-DQ8 were detected in most CD patients, corroborating the literature reports, although in minor percentage. However, the finding of 8.9 % of diagnosed CD patients negative for HLA-DQ2 and/or HLA-DQ8 alleles emphasizes that in a high admixture population, such as the Brazilian population, even in individuals with European ascendancy, the absence of these markers does not exclude CD.

Lorete Maria da Silva Kotze¹, Renato Nisihara²,
Shirley Ramos da Rosa Utiyama³ and Luiz Roberto Kotze¹

¹Service of Gastroenterology. Cajuru Hospital. Pontifical Catholic University of Paraná. ²Department of Medicine. Positivo University. ³Laboratory of Immunopathology. Clinical Hospital. Federal University of Paraná. Curitiba-Paraná, Brazil

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