DISTRIBUTION OF HLA-DQ RISK ALLELES IN A PATIENT COHORT REFERRED FOR SUSPECTED CELIAC DISEASE

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Diagnosis of coeliac disease (CD) is based on serological and histological criteria, but HLA-DQ typing is valuable, particularly for excluding the diagnosis in patients who lack the relevant heterodimers: HLA-DQB1*02/HLA-DQA1*05 (DQ2.5), HLA-DQB1*03:02 (DQ8), and HLA-DQB1*02/HLA-DQA1*02 (DQ2.2). This study aimed to assess the distribution of these risk alleles in a cohort of patients whose samples were submitted for differential diagnosis of inflammatory bowel disease.

HLA-DQB1 and DQA1 genes were typed using the LabType SSO Class II DQA1/DQB1 Typing Test (One Lambda) with LabScan3D. DQ2.5, DQ8, and DQ2.2 were considered risk alleles for CD.

Between January 2023 and August 2025, 188 patients were analyzed: 133 women (70.7%) and 55 men (29.3%), with a mean age of 40.5 years. Overall, 107 patients (56.9%) carried at least one risk allele, while 81 (43.1%) were negative. Among positive patients, 126 risk alleles were identified: 59 (46.8%) DQ2.5, 39 (31.0%) DQ8, and 28 (22.2%) DQ2.2. Most (88; 82.2%) carried a single allele, while 19 (17.8%) were heterozygous for two. The distribution was: 43 (40.2%) DQ2.5, 32 (29.9%) DQ8, 13 (12.1%) DQ2.2, 12 (11.2%) DQ2.5/DQ2.2, 4 (3.7%) DQ2.5/DQ8, and 3 (2.8%) DQ2.2/DQ8.

DQ2.5 was the most frequent heterodimer, followed by DQ8 and DQ2.2. The likelihood of developing CD varies among heterodimers, and individuals with two CD risk alleles are more susceptible than those carrying only one. According to UK NEQAS and BSHI guidelines, patient reports should indicate the presence or absence of risk alleles and, where possible, include risk stratification.

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