Assessing Trends in HLA-DQ Genotype Prevalence for Celiac Disease

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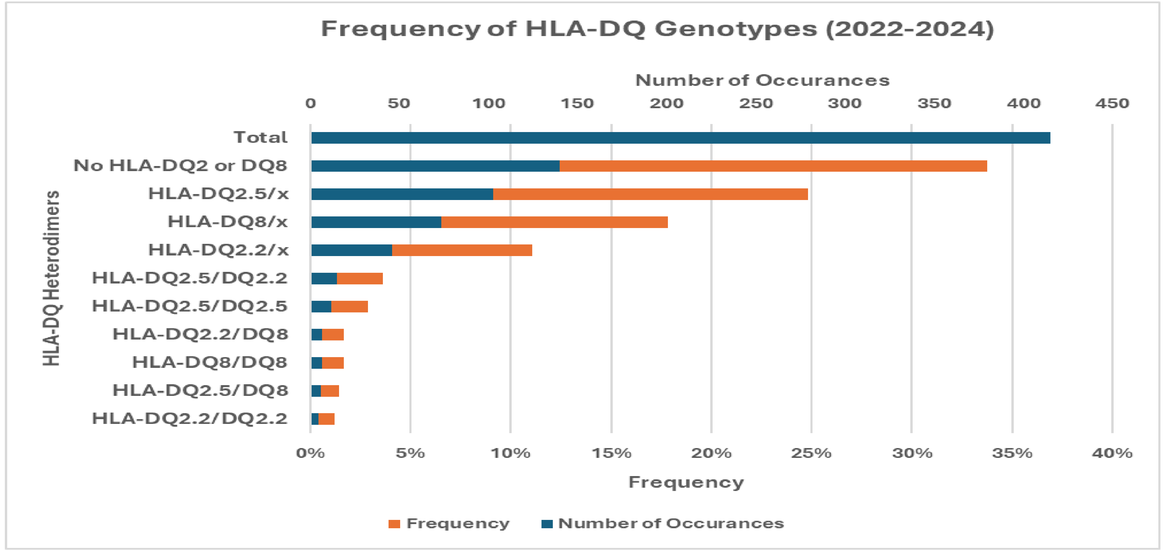
PURPOSE

Human Leukocyte Antigen (HLA) typing is crucial in understanding genetic susceptibilities to various autoimmune diseases, including Celiac disease. The association between Celiac disease and certain HLA-DQA1~DQB1 heterodimers, primarily HLA-DQ2.5 ad HLA-DQ8, has been extensively documented. This study aims to analyze the prevalence and trends of HLA-DQ genotypes among individuals tested for Celiac disease over a span of three years. The analysis focuses on identifying the most prevalent alleles and their association with Celiac disease, providing critical insights for the HLA community.

METHODS

We analyzed data from 415 individuals tested (2022-2024) for HLA-DQ genotypes associated with Celiac disease. HLA typing was performed by RSSO methodology (ThermoFisher-One Lambda) and using HLA Fusion v.4.6 for data analysis. The dataset was aggregated by HLA-DQ genotypes to observe trends, using a comparative model to evaluate genotype frequencies as proportion of the total dataset. We designated DQB1*02:01/DQA1*05:01, DQB1*02:02/DQA1*02:01, and DQ8 as HLA-DQ2.2/DQx, HLA-DQ2.2/DQx and HLA-DQ8/DQx, respectively where x represents any other DQ allele present. A detailed examination involved statistical and visualization analysis to compare the frequency of each HLA-DQ genotype.

RESULTS



The bar plot visualizes the frequency distribution and number of occurrences of HLA-DQ genotypes observed from 2022-2024. It clearly shows the distribution and prevalence of different HLA-DQ genotypes within the dataset, with"NoHLA-DQ2 or DQ8" being the most numerous categories, followed by specific genotypes such as "HLA-DQ2.5/X, "HLA-DQ8/x and "HLA-DQ2.2/X"

The analysis revealed notable trends in the occurrence of HLA-DQ genotypes. Individuals without HLA-DQ2 or DQ8 genotypes represent the largest group with 140 (34%) occurrences, indicating that a large portion of the tested individuals do not carry the genotypes strongl associated with Celiac disease (Figure 1). The HLA DQ2.5/x heterodimer is the next most prevalent, with 103 (25%) occurrences, followed by HLA-DQ8/x with 74 (18%) occurrences, aligning with their established association with Celiac disease. The HLA-DQ2.2/x genotype was also notable with 46 (11%) occurrences contributing to the diversity in genotype observed in thi cohort. There is consistency in genotype distribution, highlighting the prevalence of HLA-DQ2.5/x and HLA DQ8/x genotypes in HLA association with Celia disease.

CONCLUSION

This analysis of HLA-DQ genotype prevalence offers crucial insights into the genetic landscape of Celiac disease, highlighting the significance of HLA-DQ genotyping in disease assessment. The study revealed notable trends in genotyping distribution, emphasizing the prevalence of genotypes associated with low and high risk of the disease. The observed trends and genotype distributions provide valuable insights for clinicians and researchers, aiding in the refinement of diagnostic and therapeutic approaches.

CITATIONS

1. Rok Seon Choung, John R. Mills, Melissa R. Snyder, Joseph A, Murray, Manish J. Ghandi (2020) Celiac disease risk stratification based on HLA-DQ heterodimer (HLA-DQA1-DQB1) typing in a large cohort of adults with suspected celiac disease. Human Immunology 81 (2-3), 59-64.