Poster Presentations

PP-07

Human Leukocyte Antigen Typing Results Among Patients with Probable Celiac Disease

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The objective is to determine the frequency of the presence of risk alleles for celiac disease (CD) among a cohort of patients whose samples were sent for differential analysis of inflammatory bowel disease.

Human leukocyte antigen (HLA)-*DQB1* and *DQA1* genes were typed with LabType SSO Class II DQA1/ DQB1 Typing Test (One Lambda) using LabScan3D. and Celiac Multiplex Real Time PCR Kit (SNP). HLA-DQB1*02/ HLA-DQA1*05 (DQ2.5), HLA-DQB1*03:02 (DQ8) and HLA-DQB1*02/ HLA-DQA1*02 (DQ2.2) were reported as risk alleles for CD.

Seventy-three patients were tested between January 2023 and September 2024. Fifty-two (71.2%) were women and 21 (28.7%) men with an average age of 58. Forty patients (54.8%) were positive and 33 (45.2%) negative for risk alleles. Among 40 patients a total of 45 risk alleles were detected; 22 DQ2.5 (48.9%), 13 DQ8 (28.9%) and 10 DQ2.2 (22.2%). Thirty-five patients had one allele; 19 (47.5%) DQ2.5, 10 (25%) DQ8 and 6 (15%) DQ2.2. Five patients were heterozygous with risk alleles: 2 (5%) DQ2.5/DQ2.2, 2 (5%) DQ2.2/ DQ8 and 1 (2.5%) DQ2.5/ DQ8. Both methods were concordant for risk allele typing except for one patient, where DQ8 was positive with reverse transcription-polymerase chain reaction (RT-PCR) but negative with next-generation-sequencing. RT-PCR could not detect DQ2.2, leading to six (8.21%) DQ2.2 positive patients being negative with RT-PCR.

DQ2.5 homozygosity, the highest risk for severe CD, was not detected. Five patients were heterozygous with risk alleles. Heterozygosity of risk alleles is considered as greater risk, compared with having only one heterodimer. United Kingdom National External Quality Assessment Service and British Society for Histocompatibility and Immunogenetics guideline recommends risk stratification to be included in patient reports.

PP-08

Investigation of Human Leukocyte Antigen and Killer Cell Immunoglobulin-Like Receptor Relationship in Acute Myeloid Leukaemia Patients

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Objectives: Acute myeloid leukemia (AML) is a complex disease and most common type of blood cancer with poor prognosis. AML is a malignancy of the stem cell precursors of the myeloid lineage (red blood cells, platelets and white blood cells other than B- and T-cells). Like other malignancies, it is caused by genetic variations leading to neoplastic changes and clonal proliferation (1). Natural killer (NK) cells, one of the cells of the innate immune system, originate from the bone marrow and constitute 10-20% of the entire lymphocyte population. NK cells, which are involved in the innate immune response, are important cells involved in the immune response against viral infections and tumour cells, especially leukaemia, lymphoma and metastatic tumour cells (2). NK cells are cytotoxic cells that regulate the activity of the immune system through the killer cell immunoglobulin-like receptor (KIR) they express. The activity of NK cells is regulated mainly through KIRs, which are composed of activator and inhibitory receptors and have great genomic diversity. The genes encoding KIRs are located on chromosome 19. Sixteen *KIR* genes have been characterised to date. Fourteen *KIR* genes encode receptors that trigger inhibition (3DL1-3, 2DL1-3, 2DL5) or activation (3DS1, 2DS1-5) or both (2DL4), while two pseudogenes (2DP1 and 3DP1) are not known to encode cell surface receptors. KIRs regulate the function of NK cells by binding with human leukocyte antigens (HLA) class I molecules on the surface of target cells (3-5). In recent years, there has been an increasing number of studies showing an association between AML and KIR. The aim of our study was to determine the frequency of KIR genotypes in patients diagnosed with AML.

Materials and Methods: The study included 22 patients (45.45% female and 54.55% male, with no gender or age difference) who were admitted to Başkent University Adana Dr. Turgut Noyan Research and Medical Centre, Department of Hematology (between 2017 and 2023), diagnosed with AML and died during treatment. HLA typing and KIR typing (Immucor, USA) tests were performed with Luminex method according to the manufacturer's protocol from the patient's DNA sample.

Results: In the study, haplotype A was identified in 6 of 22 patients, haplotype B was identified in 16 of 22 patients, 27.27% and 72.73% respectively. Two of the 6 haplotypes A were C1/C1 and 4 was C2/C1. C1/C1 was determined as 13.64%, C1/C2 was determined as 72.73% and C2/C2 was determined 13.64%. In all patients, KIR3DL3, KIR3DL2, KIR3DP1 and KIR2DL4 (framework genes) were detected. The inhibitory KIR2DL1 which recognize C2 group allele, was most frequently detected in the AML patient group. On the other hand, KIR2DS5 (36.36%) was detected with the lowest frequency. *KIR* gene frequency and profile in AML patients studied are given in Figures 1 and 2, respectively.

Discussion: Previous studies have investigated the relationship between KIR, HLA and AML, but the results have been found to vary according to the