Poster Presentations

PP-12

What Happens? If Donor and Recipient Carrying PKHD1 and PKD Gene Variants in Renal Transplantation

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Objectives: In this case report, we present a patient who was diagnosed with autosomal dominant polycystic kidney disease (ADPKD) at the age of 1 year and given to our clinic as a 17-year-old renal transplant candidate. In living donor screening, human leukocyte antigens (HLA) 4/6 compatible with the mother was a carrier for polycystic kidney disease-1 (PKD1) and *polycystic kidney disease with/without hepatic disease (PKHD1)* gene variants.

Materials and Methods: HLA tissue typing was performed on the patient and the donor candidate's mother and father. 4/6 HLA compatibility was detected with the mother. Due to the patient's diagnosis of ADPKD, the family analyzed the DNA-next-generation-sequencing "Kapa Hypercap Hereditary" kit. After genetic testing, kidney transplantation was performed from the donor candidate's mother to her daughter. After the transplantation, the patient is under follow-up with stabilized routine follow-up.

Results: The mother was 37 years old and had no history related to ADPKD. However, the maternal genetic results revealed the PKD1 C.2695C>G likely pathogenic and PKHD1 c.5353T>C *VUS* gene variants. The PKHD1 c.525delT pathogenic variant and the maternal PKHD1 c.5353T>C and PKD1 c.2695C>G variants were analyzed. The patient received a kidney from the mother in August 2024. He returns once a month for routine monitoring.

Conclusion: It was determined that the PKHD1 c.525delT pathogenic variant with autosomal dominant inheritance was the underlying cause of the patient's etiopathogenesis. The absence of this variant in the mother of the donor candidate increased the transplant's success. The detection of the PKD1 c.2695C>G probably pathogenic and PKHD1 c.5353T>C VUS variants in heterozygous genotype in autosomal recessive inheritance led to the mother's evaluation as a carrier.

Keywords: PKD1, *PKHD1* gene, renal transplant

PP-13

Association of Severe Spinal Involvement with HLA Alleles in Patients with Radiographic Axial Spondyloarthritis

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There are limited studies showing the association between human leukocyte antigen (HLA) alleles and clinical severity in patients with axial spondyloarthritis (axSpA). In this study, we aimed to determine the HLA alleles that may be associated with disease severity and severe spinal involvement in radiographic axSpA (r-AxSpA) patients. Inclusion criteria for the study were patients with axSpA have a symptom duration of more than 10 years and their Bath Ankylosing Spondylitis Radiology Index, calculated by two rheumatologists. Patients were divided into two groups according to age at diagnosis by receiver operating characteristic analysis: ≤28 years and ≥29 years. HLA genotyping was performed using the sequence-specific oligonudeotide probe method for HLA alleles. Logistic regression analysis and decision tree models were used to assess the association between radiographic parameters and demographic and genetic characteristics. Overall, 100 patients were included. 72 (72%) of (r-AxSpA) patients were HLA-B*27 positive. Sixty-two (62%) of the patients were male, the mean (standard deviation) duration of symptoms was 21.1 (9) years. Syndesmophytes were found in 88.9% of HLA-A*02 homozygous male patients without extra-articular involvement. When the decision tree was evaluated, male sex, any extra-articular involvement, and HLA-A*02 homozygosity were associated with syndesmophyte. Hip involvement appears to be associated with HLA-A*02 homozygosity and early age at diagnosis in the decision tree. As a result, we found out that HLA-A*02 is related to syndesmophyte, but further studies are needed to support the frequency of HLA-A*02 homozygosity and its association with radiographic severity in r-AxSpA patients.