P269

PREIMPLANTATION GENETIC DIAGNOSIS (PGD) FOR SINGLE GENE DISORDERS/ ACQUIRED DISEASE WITH HLA TYPING: A BRIEF REPORT

Türker Duman[1], Evrim Ünsal[2], Özge Ayvaz[2], Süleyman Aktuna[3], Leyla Özer[3], Fatma Akyigit[3], Hüseyin Tutkak[1], Aysun Baltaci[4], Volkan Baltaci[2]

[1]Ankara University School of Medicine, Ankara, Turkey, [2]Istanbul Bilim University School of Medicine, Istanbul, Turkey, [3]Mikrogen Genetic Diagnostic Laboratory, Ankara, Turkey, [4]Genart Woman Health and Reproductive Biotechnology Center, Ankara, Turkey
Correspondence: dumanturker@yahoo.com

Preimplantation Genetic Diagnosis (PGD) of single gene disorders, combined with HLA matching results of "Mikrogen Genetic Laboratory" summarized between the years of 2012–2014. In this report HLA typing was performed for acquired diseases such as acute myeloid leukemia (AML) and acute lymphoid leukemia (ALL) and also for single gene disorders. This is a retrospective study. Overall, forty-three couples were included in the PGD-HLA cycles. 37 couples undergoing HLA typing were carrying single gene disorders. Thalassemia, Griscelli Syndorme, Wiskott Aldrich Syndrome, Kostmann Syndrome, Krabbe Syndrome, Chronic Granulomatosis, Neutropenia, MHC Class II Immunodeficiency were among the single gene disorders studied. Multiplex Nested PCR followed by RFLP (Restriction Fragment Length Polymorphism) was preferred for mutation analysis. In case of some mutations when disrupted restriction enzyme site was not available Site-Directed Mutagenesis method was performed. Linked informative STR markers were used to exclude risk of Allele Drop Out (ADO) that would introduce false positive/negative results. Informative HLA STR markers were selected and fragment analysis was applied in order to achieve HLA compatible embryos for affected sibling. According to test results of all patients, 69 embryos were normal, 85 were heterozygous and 76 were found to be homozygous mutant. Amplification failure was seen on 28 embryos. After transfer of 40 disease-free HLA matched embryos, 9 HLA matched babies were born. HLA Typing has become a robust technique in the recent years. This

technique enables couples with children that have either single gene disorders or acquired diseases to have healthy babies who will also become a therapy opportunity for their siblings.