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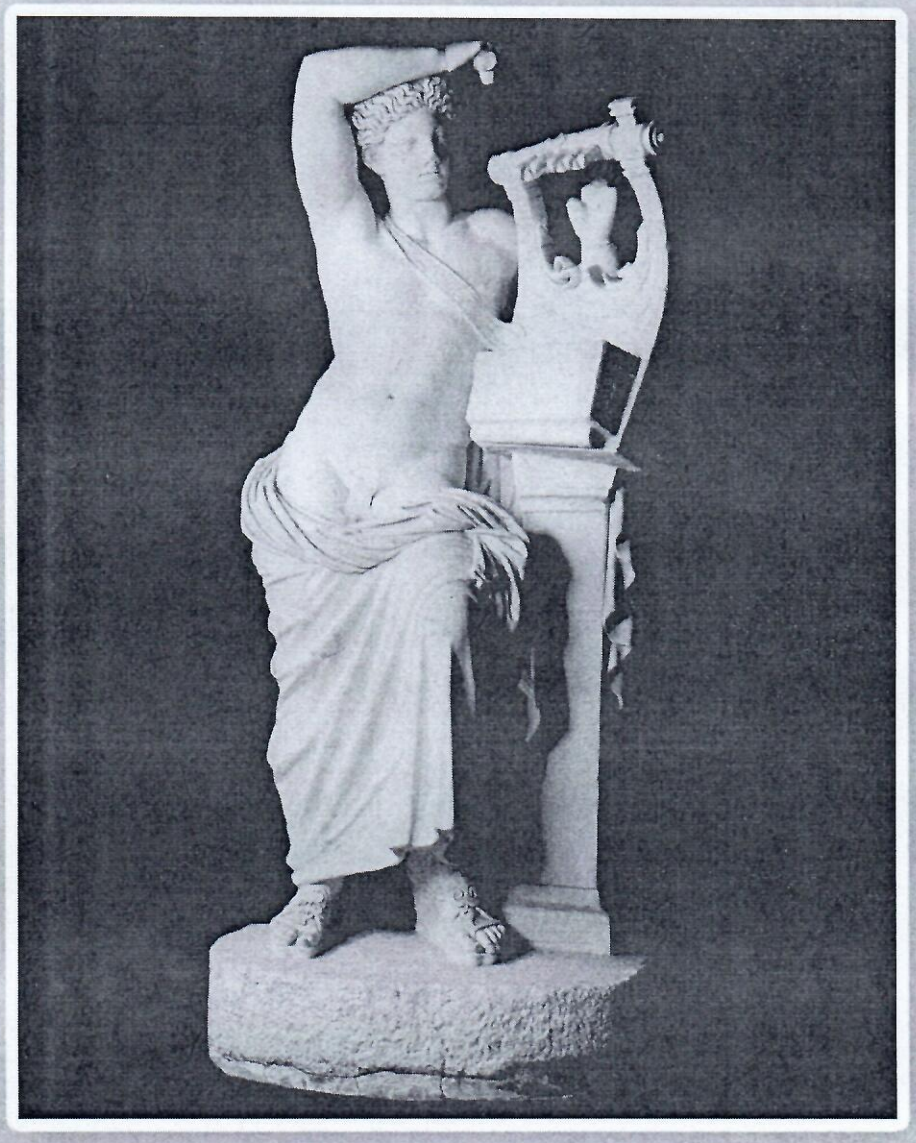

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P-060 - Chromosomal abnormalities in couples that undergo assisted reproduction technologies

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Introduction: Recent progress in reproduction technologies helps many couples in conceiving pregnancy. Considerable portion of pregnancies obtained in this way have an unfavorable outcome, ending up with unsuccessful conception, recurrent miscarriage, stillborn, or birth of a child with certain genetic condition. Many pathogenic mechanisms such as chromosomal instability, single gene mutations, imprinting, and, most likely, idiopathic reproductive loses are underlying factors for poor pregnancy outcome. Among all, chromosomal rearrangement in one of the partners could be found in 2-4% of high-risk couples. Materials and methods: We present several cases where chromosomal changes and polymorphic chromosomal variants were found in couples that had performed some of the assisted reproduction technologies (ART) repeatedly. Results: Balanced reciprocal translocations between variable chromosomes, as well as Robertsonian translocations were detected. Inversion of chromosome 9 was found in infertile couples more frequently that in general population. Despite performing ART, unsuccessful implantation and miscarriage were the most frequent outcomes in these cases. Genetic counseling and the best treatment options were offered in all. Discussion and conclusion: Reciprocal chromosomal breakage could occur between variable chromosomes due to several mechanisms: existence of polypurine/polypirimidine repeats along DNA, microdeletions/duplications, regions where prolonged replication occurs, etc. The size of the chromosomal segment involved in the translocation has a crucial role in unfavorable pregnancy outcome. Karyotype is still the only method that provides diagnosis of balanced chromosomal rearrangements. Although disputed by some authors due to the cost of the analysis, selective kariotyping should be mandatory in high-risk couples.

KEYWORDS: infertility, selective karyotyping

13th Balkan Congress of Human Genetics

17-20 April 2019
Trakya University, Edirne - TURKEY

Certificate of Participation

This certificate is presented to

VİOLETA ANASTASOVSKA

for acknowledgement as a participant in the ***13th Balkan Congress of Human Genetics*** *which was held in Edirne, Turkey on* ***17-20 April 2019****.*

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