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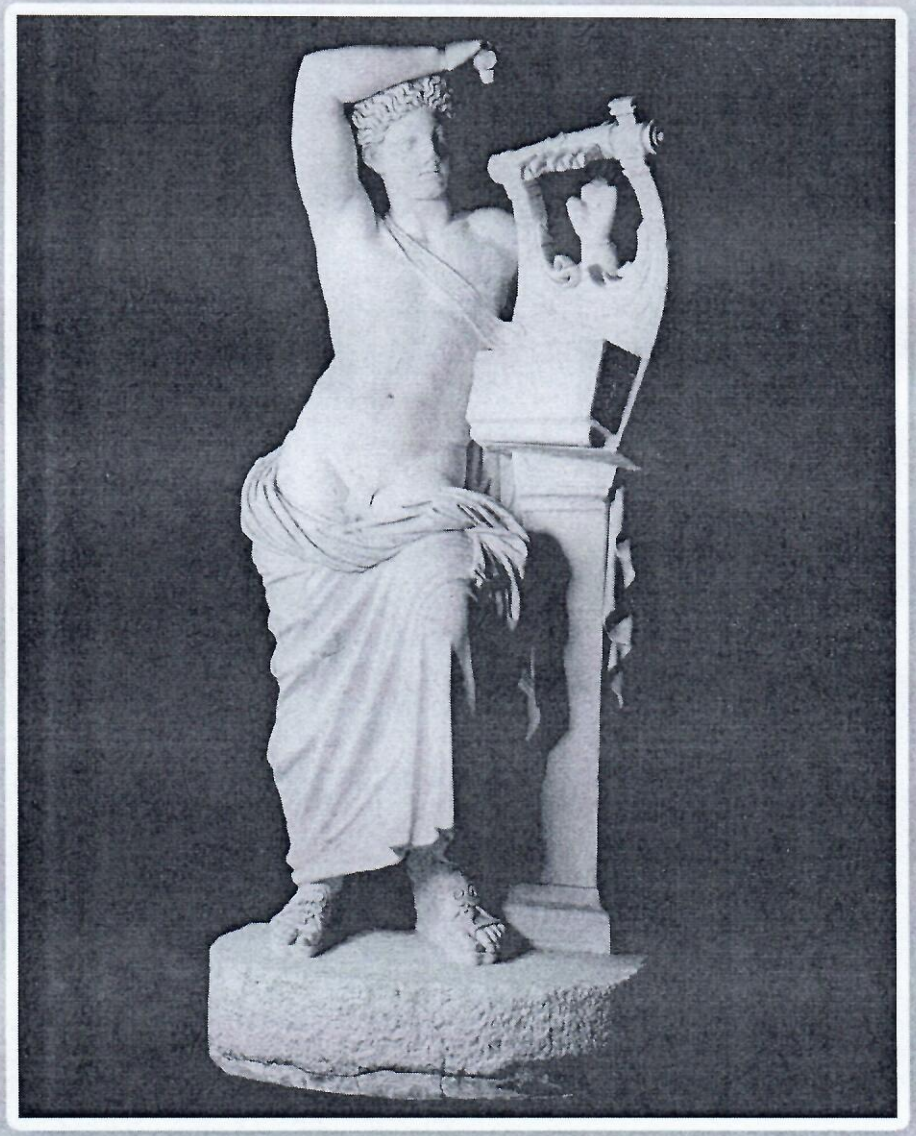

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Supplement

P-058 - DETERMINATION OF MONOSOMY 7 IN ANEMIA APLASTICA USING FLUORESCENCE IN SITU HYBRIDIZATION

Gordana Ilieva[1], Elena Sukarova-Angelovska [1], Violeta Anastasovska[1], Svetlana Koceva[2],

[1]Genetic Laboratory, Department of Endocrinology and Genetics, University Pediatric Clinic, Skopje, Republic of Macedonia , [2]Department of Hematology, University Pediatric Clinic, Skopje, Republic of Macedonia,

Background: Aplastic anemia (AA) is a hematologic disorder characterized with peripheral blood pancytopenia and hypocellular bone marrow. Bone marrow transplantation and immunosuppressive therapy are standard treatment strategies for severe AA. Monosomy 7 (del 7q) is the most common chromosomal aberration in aplastic anemia. Presence of this aberration is crucial for the therapy. Materials and Methods: A boy with clinical signs of pancytopenia was diagnosed at the Department of Hematology, University Pediatric Clinic, Skopje, Macedonia, according to standard criteria. We performed standard G-banded karyotype from bone marrow and fluorescence in situ hybridization (FISH) on interphase nuclei using del 7q deletion probe LPH 025 (Cytocell). Standard protocols for karyotyping and FISH were used. Results: The karyotype in this case was unsuccessful since the bone marrow was poor in cells. The FISH analysis, using del 7q deletion probe, showed presence of two signals in the interphase nuclei on 10% of cells, which indicates monosomy 7. This finding was key factor for further treatment strategy for the patient. It was decided antithymocyte globulin not to be given, and hematopoietic stem cell transplantation was performed. After transplantation the patient started with normal cell production. Conclusion: Fluorescence in situ hybridization analysis is rapid, sensitive and reliable tool for detection of chromosomal aberrations especially in cases when standard karyotype is unsuccessful .

KEYWORDS: Aplastic anemia, karyotype, FISH, monosomy 7

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**.*

Prof. Dr. Dijana Plaseska-Karanfilska
President of Macedonian Society of Human Genetics

Prof. Dr. Mehmet Ali Ergun
President of Medical Genetics Association of Turkey