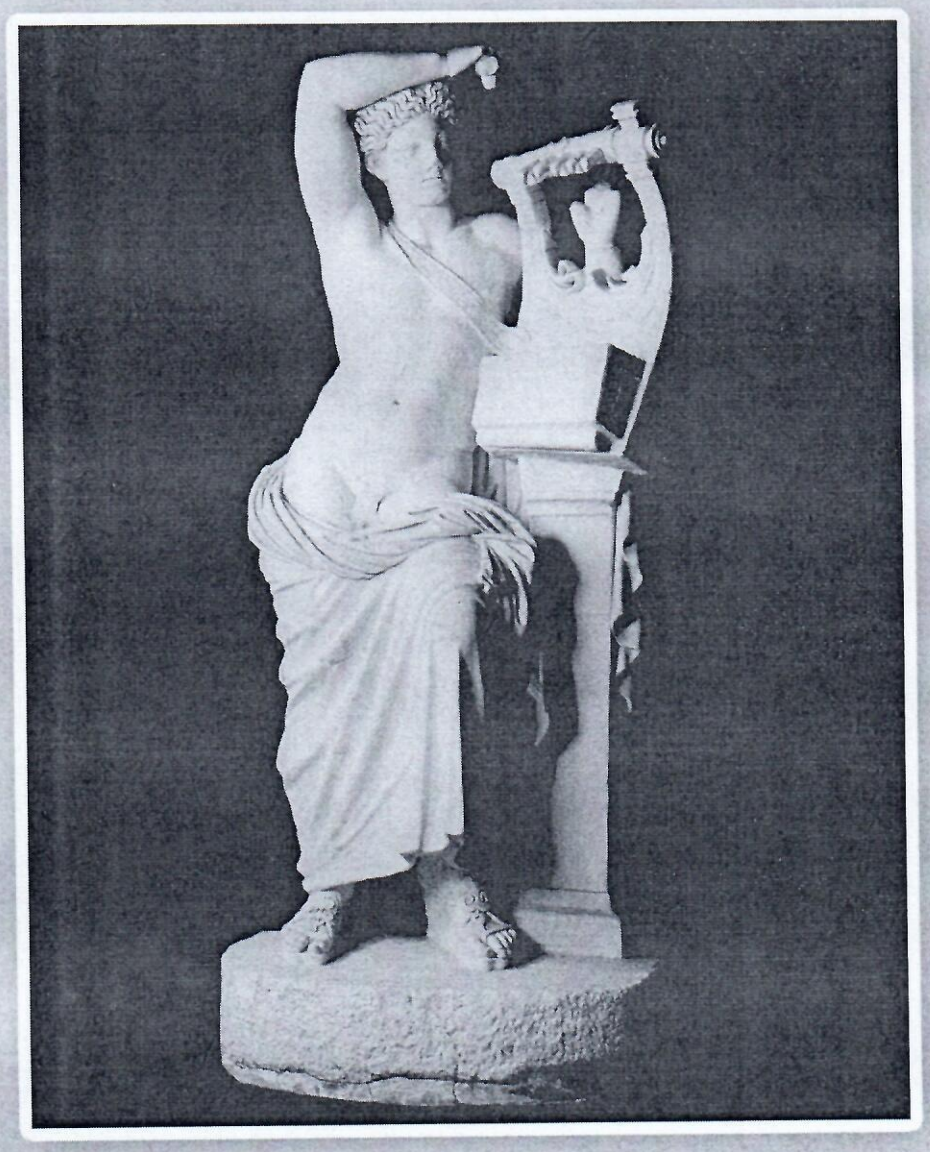
BJMG

VOL 22, 2019
SUPPLEMENT 1

ISSN 1311-0160

Balkan Journal of Medical Genetics



International Journal of Medical Genetics
Supplement

P-071 - DETECTED GENOTYPES IN MACEDONIAN PATIENTS WITH SIMPLE VIRILIZING FORM OF CONGENITAL ADRENAL HYPERPLASIA

Violeta Anastasovska[1], Mirjana Kocova[1], Elena Sukarova-Angelovska[1], Nikolina Zdraveska[2], Gordana Ilieva[1],

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

Introduction: Congenital adrenal hyperplasia (CAH) ranks among one of the most frequent recessive inborn errors of metabolism. Classical simple virilizing (SV) form of CAH, mainly associated with I172N mutation in the CYP21A2 gene, leads to virilization of external genitalia in newborn females and pseudoprecocious puberty in both sexes, due to reactive androgen overproduction. Materials and Methods: Molecular analysis of the nine most common mutations in CYP21A2 gene was performed in 32 Macedonian patients with clinical diagnosis of SV form of CAH, using PCR/ACRS method. Results: Seven different mutations were detected on 87.5% (56/64) of the alleles. Complete genotype was detected in 27/32 SV patients (84.4%). The most prevalent was IVS2/IVS2 genotype found in 7/27 (25.9%) of the patients followed by P30L/I172N found in 5 (18.5%), P30L/IVS2 in 4 (14.8%), P30L/P30L in 3 (11.1%) and I172N/I172N genotype found in 2 (7.4%) of the patients. The genotypes P30L/Q318X, P30L/R356W, P30L+V281L/V281L, IVS2/I172N, I172N/Q318X and Del 8ntG110/V281L were detected in only one patient each. Two of the SV patients had genotype with IVS2 on the one of the alleles with no detected mutation on the second allele. In 3/32 (9.4%) SV patients no mutation was detected. Conclusions: The most prevalent genotype among the Macedonian patients with SV form of CAH was IVS2/IVS2. We observed wide genotypic variability in SV CAH, and low prevalence of I172N mutation in exon 4 which is considered as a typical SV mutation.

KEYWORDS: 21-hydroxylase deficiency, CYP21A2 gene, simple virilizing form

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