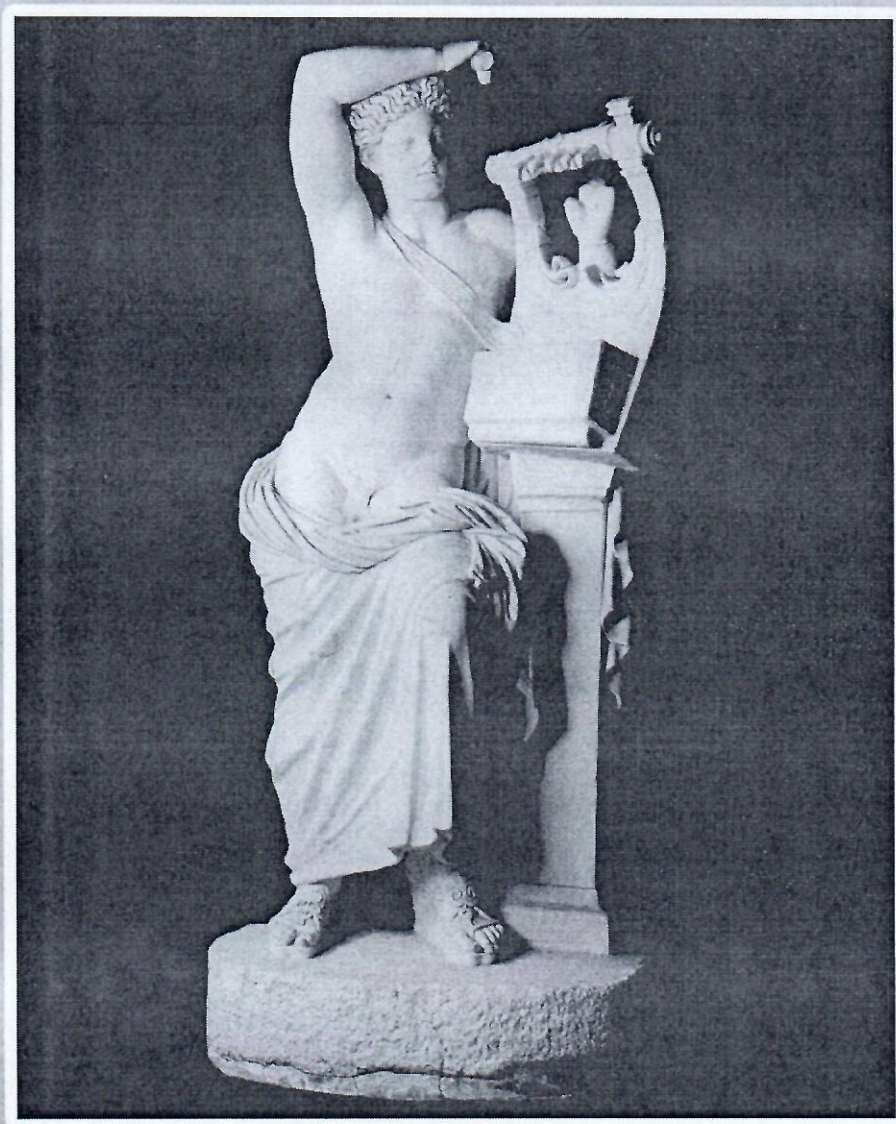


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P-032 - FREQUENCY OF DETECTED GENOTYPES IN PATIENTS WITH SALT-WASTING FORM OF 21-HYDROXYLASE DEFICIENCY

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Introduction: Steroid 21-hydroxylase deficiency is a most frequent cause of congenital adrenal hyperplasia (CAH), due to mutations in the CYP21A2 gene. Nine pseudogene-derived CYP21A2 point mutations account for about 80% of all CYP21A2 defects. Classical CAH can present as severe salt-wasting (SW) and simple virilizing (SV) form. **Materials and Methods:** We have performed molecular diagnosis of the nine common CYP21A2 point mutations in 24 Macedonian patients with clinical and laboratory signs of severe SW form of CAH, evaluated at Department of Endocrinology and Genetics, University Pediatric Clinic, Skopje, Republic of Macedonia, using differential PCR/ACRS. **Results:** In 91.7% (22/24) of the SW patients complete genotype was detected. Seventeen (77.3%) patients were homozygous, and five (22.7%) were compound heterozygotes. Two patients harboured none of the tested mutations. The most common genotype was IVS2/IVS2 found in 14/22 (63.6%) patients, followed by Q318X/Q318X and IVS2/Q318X observed in 2/22 (9.1%) patients, each. Other genotypes Del8ntG110/Del8ntG110, R356W/Del8ntG110, IVS2/V281L+Q318X+R356W and Q318X/Del9nt(c.1271_1279) were observed in only one patient, each. No phenotypic differences in SW patients were detected based on genotype. **Conclusion:** The most prevalent genotype among the Macedonian patients with SW form of CAH was IVS2/IVS2. Our results support the role of the IVS2 splice mutation in the SW phenotype of the disease.

KEYWORDS: congenital adrenal hyperplasia, salt-wasting form, CYP21A2 gene, point mutations, steroid 21-hydroxylase



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Certificate of Participation

This certificate is presented to

VIOLETA ANASTASOVSKA

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