

# COMMON CYP21 GENE MUTATIONS IN CZECH PATIENTS AND STATISTICAL ANALYSIS OF WORLDWIDE MUTATION DISTRIBUTION

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## SUMMARY

*CYP21 gene molecular analysis was performed to determine the mutational analysis of 87 unrelated Czech patients with different forms of steroid 21-hydroxylase deficiency. Eight of the most common point mutations (intron 2 splice, P30L, 8 bp deletion in exon 3, I172N, V281L, Q318X, R356W, P453S) were analyzed using Amplification–created restriction site method. Cluster in exon 6 mutation was analyzed using allele-specific oligonucleotide hybridisation. Deletions and conversions were screened by modification of an sequence specific oligonucleotides hybridisation. The most frequent mutation in Czech patients was intron 2 splice mutation (45.4%). The comparison of mutation frequencies was made among Czech and European population (high frequency of intron 2 splice, 8bp deletion, P30L, P453S and low frequency of deletions/conversions in Czech population) and among Czech and different regions worldwide (low frequency of deletions/large gene conversions, V281L, R356W, high frequency of intron2, 8bp deletion, P30L and P453S in Czech population). We compared common mutation frequencies of different regions worldwide (North and South America, Asia, North Africa, Europe). Significant differences in selected regions were determined by ANOVA statistical analysis (One-sample t-test, confidence interval) at value of  $p < 0.05$ .*

**Key words:** CYP21 gene, frequency of mutations, 21-hydroxylase deficiency, genotype, phenotype, comparison among regions

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