

MOLECULAR GENETIC STUDIES IN INDIAN PATIENTS WITH 21 HYDROXYLASE DEFICIENCY

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Congenital adrenal hyperplasia (CAH) comprises a group of autosomal recessive disorders. The most common cause is steroid 21-hydroxylase deficiency due to the mutations in CYP21 gene. This study was conducted to determine the mutations causing this disease and their frequencies in Indian subjects by direct sequencing of the CYP21 gene. A total of 23 patients were recruited for the study from Genetic and Endocrine clinics of All India Institute of Medical Sciences, New Delhi, India. The samples were collected from the patient and both the parents. Following selective amplification of CYP21 against the pseudogene CYP21P sequence analysis of the entire coding region and some of the intronic regions of CYP21 gene was done. We characterized 50 mutant alleles from 21 unrelated patients. It was possible to identify 9 different mutations including 2 novel mutations. These two mutations included a 9 bp insertion in exon 2 and F306V mutation in exon 7 of CYP21 gene. The most common mutations were I2 splice (28.0%), I172N (26.0%) and Q318stop (16.0%). The mutations I172N and Q318stop were found at a higher mutation frequency and gene deletion (8.0%) at a lower mutation frequency than reported from other parts of the world. The genotype-phenotype correlations were made and I2 splice mutation was found to be associated mostly with the salt-wasting disease and I172N with simple-virilizing phenotype. The combination of the mutations also played an important role in determining the phenotype. This study has enabled identification of common mutations for the Asian Indians for the salt-wasting and simple-virilizing patients and the information is useful to devise strategies for prenatal diagnosis.

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