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Common CYP21A2 Gene Mutations in South Indian Congenital Adrenal Hyperplasia Patients

R. Nageshwari[1], M. Dhivakar[2], K. Balakrishnan[2], Sivan Arul Selvan[1] and V. Kumaravel[1*]

[*]Alpha Hospital and Research Centre, Madurai, Tamil Nadu, India
[#]Madurai Kamaraj University, Madurai, Tamil Nadu, India

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ABSTRACT About 90 percent of Congenital Adrenal Hyperplasia (CAH) patients exhibit defects in the CYP21A2 gene that results in steroid 21-hydroxylase deficiency (21- OHD). As more than 100 mutations prevail in CAH, diagnosis and time responsive therapeutic interventions require knowledge of the common mutations in the regional population. Hence, the present study aims to assess the prevalence of nine common CYP21A2 gene mutations in a South Indian, regional population consisting of a group of CAH patients and a group of CAH patients along with their parents. 6 clinically diagnosed CAH patients, 5 clinically diagnosed CAH patients along with their parents were screened for nine common mutations using allele-specific polymerase chain reaction amplification followed by restriction fragment length polymorphism. Out of 11 patients, 8 were identified to be females and 3 were males. 5 patients were simple virilizers and 6 were salt wasters. The age at presentation varied from 1 day to 24 years. Molecular diagnosis of the CYP21A2 gene revealed that the highest number of patients harbored *P30L* and *Q318X*, followed by *P453S*, *I172N*, *In2 splicing*, *R356W* and *Δ8bp deletion* mutations. The results of the study clearly indicates that allele-specific PCR combined with RFLP is reliable for the molecular diagnosis of 21-OHD and can be easily included in small scale, routine laboratory analysis. Cohesively, the study strongly recommends the initiation of Indian national/regional neonatal screening programs for CAH.