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Mutation screening of *CYP21A2* and *ATP6V0A4* genes in patients with inherited defects in the genitourinary tract

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Genitourinary tract is the system of combination of organs related with the production and excretion of urine and those related with reproduction. Genitourinary system disorders include both symptomatic and asymptomatic disorders. These disorders may be caused by congenital abnormalities, infectious diseases, trauma or any damage to urinary structural conditions. Congenital adrenal hyperplasia is one of the many forms of genitourinary tract disorders which has autosomal recessive mode of inheritance. CAH due to 21-hydroxylase (encoded by *CYP21A2* gene) deficiency is a common metabolic disorder that causes abnormal androgen synthesis in body. We have analyzed *CYP21A2* gene for all 10 coding exons in 15 Pakistani families. Mutational screening has identified a synonymous single nucleotide polymorphism in two families (PB1442 and PB1553) and a recurrent polymorphism in another family (PB1295). However pathogenic mutations were not identified in the coding region. Kidneys are the organs that have regulatory role in animals. One of the important functions of the kidneys is the maintenance of acid base balance. Disturbance in this balance will lead to urinary tract infections. Renal tubular acidosis is one of the major forms of kidney disorders. *ATP6V0A4* gene coding the α 4 isoform of v-ATPases plays an important role in RTA. We have screened selected exons of this gene in two RTA affected Pakistani family. Mutational screening has identified a C>T variation in intron 7 of *ATP6V0A4* gene. A heterozygous variation has been identified in exon 9 of this gene where G has been replaced by A at nucleotide position 945 changing the codon GAC (coding aspartate) to AAC (coding asparagine). However, sequencing of more exons and regulatory regions and functional studies are needed to prove that this variation has some role in RTA phenotype.

Biography

Maliha Rasheed has completed her Mphil from Quaid-i-Azam University, Pakistan in Biotechnology. Human Molecular Biologist with 4 years national, international research experience in molecular genetics. Currently, she is working as Lecturer in the department of Biotechnology, Women University of AJ&K, Pakistan.