PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
International Journal of
HUMAN GENETICS

Int J Hum Genet, 19(4): 170-178 (2019)
PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
DOI: 10.31901/24566330.2019/19.04.742

Mutational Analysis of TYR Gene Causing Oculocutaneous Albinism in Families from District Peshawar

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KEYWORDS Mutation. Oculocutaneous Albinism. Peshawar. Protein Stability. Tyrosinase

ABSTRACT Oculocutaneous Albinism is a cluster of genetic diseases specified by full or partial loss of biosynthesis of melanin in melanocytes cells. The aim of current research was to determine the mutational status of this gene in the enrolled OCA diagnosed consanguineous families. DNA was isolated from all collected samples using inorganic method of extraction. The samples were screened for identification of *TYR* mutations in the enrolled families using sequencing of the exons of this gene. A homozygous missense mutation (*c.132T>A*) in the *TYR* gene was recognized in the OCA affected individuals in the consanguineous family-II whereas sequence analysis of normal individuals have shown no sequence change in respective exon. A single missense substitution (*p.Ser44Arg*) was identified in the *TYR* using PolyPhen 2, MutationTaster and I-Mutant software. The current study is important in terms that it confirms important mutation causing Oculocutaneous albinism in this region.