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Relationship of a Novel c.429delC Deletion in Hairless Gene HR with Alopecia in Two Families from Southern Punjab, Pakistan

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Atrichia with papular lesions (APL) is a rare autosomal recessive form of total alopecia, characterized by hair loss soon after birth and the development of papular lesions of keratin-filled cysts over extensive areas of the body. Two consanguineous families were enrolled from Basti Mochi Wala, Mouza Gulab Shah in Muzaffargarh District of Punjab (Pakistan) having multiple siblings suffering from alopecia. The aim of this study was to find out the genetic mutation(s) in hairless (HR) gene, if any, in the enrolled subjects. A questionnaire was filled for each subject on the sampling site in order to collect epidemiological data associated with the disease. Patients from both families exhibited congenital atrichia with papular lesions (APL) including hair loss in the scalp, pubic and other body parts. Polymerase chain reaction (PCR) was used to amplify all the over lapping intron exon regions of HR gene followed by DNA sequencing. Analysis of the DNA sequence revealed a novel deletion c.429delC in exon 2 of HR gene. Due to this deletion Proline at 144 changed into Lysine causing frame shift leading to premature termiation of the protein after 23 amino acid residues (p.P144LfsX23), resulting in a truncated HR protein with 166 amino acid residues. The mutation followed Mendalian pattern of inheritance as all the patients are homozygous for the mutation while parents were heterozygous and unaffected siblings from both families were either heterozygous for the reported mutations or they lacked this mutation.

Key Words: APL, HR gene, PCR, c.429delC, DNA sequencing.

Biography

Maryam Ijaz is a third year PhD student at Institute of Zoology, Bahauddin Zakariya University Multan, Pakistan.. She is working the genetic basis of various rare disorders in Pakistani population. She has published 5 papers in reputed journals and few more are in progress.

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