Abstract

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Genetic mapping and sequence analysis of the candidate genes causing various forms of genetic skin disorders

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Abstract

Structural organization of skin is maintained by several different cell types expressing a variety of proteins which are responsible for generating complex type of interactions. Several types of human inherited skin disorders including blistering, xeroderma pigmentosum and many others are the result of sequence variants in genes encoding these proteins that disrupt their functions.

In the present study, four families (A-D) of Pakistani origin, segregating various types of skin disorders either in autosomal recessive or autosomal dominant manners, were characterized at clinical and genetic levels. Following characterizing the families at clinical levels, genotyping using microsatellite markers and DNA sequencing were used to search for the disease causing variants in the previously reported candidate genes.

In family A with xeroderma pigmentosum, following establishing linkage on chromosome 3p25.1, a candidate gene XPC was sequenced using Sanger technique. Sequence analysis revealed a novel homozygous pathogenic deletion mutation in affected members of the family. Segregation analysis revealed complete segregation of the variant with disease phenotypes within the family.

Attempt was made to characterize two families (B, C) inheriting Oculocutaneous Albinism Type 2 (OCA2). Family B showed linkage to OCA2 gene on chromosome 15q12-q13.1. The gene was sequenced in one of the affected member but failed to detect causative variant. Family C on the other hand did not show linkage to the tested candidate genes (OCA2, MC1R).

Fourth family D, investigated here, showed a rare nail abnormality Nail Clubbing. Due to limited time available, only one candidate gene HPGD, involved in autosomal dominant nail clubbing, was sequenced in affected members of the family. However, this effort failed to identify causative sequence variant.

The study, presented in the dissertation, identified a novel variant in a candidate gene in one family only. This has enhanced the possibility of identifying novel causative genes in three other families.

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Biography

Warda Ali Khan did master in biomedical engineering from NEDUET. Currently working as TA in the biomedical department of NEDUET. Being a Biomedical Engineer Warda Ali Khan did two internships in government hospitals.

