A novel mutation of COL7A1 in a Chinese family with dystrophic epidermolysis bullosa priguniosa

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Background: Dystrophic epidermolysis bullosa priguniosa (DEB-Pri) is a rare subtype of dystrophic epidermolysis bullosa (DEB) characterized by blisters and scars. It was caused by the mutation of COL7A1 gene encoding type VII collagen, fibrosis, and skin contractures. We identified a novel mutation of COL7A1 in a family with different severities of DEB-Pri.

Methods: Clinical data and skin biopsies were collected from the proband and her unaffected member. The mutation was confirmed using Sanger sequencing.

Results: We identified a novel mutation in the COL7A1 gene (c.4624C>T, p.R1542X) in a family with DEB-Pri. The proband had severe disease with blisters, scar formation, and contractures, while her unaffected member had only skin scars.

Conclusion: This study highlights the genetic diversity of DEB-Pri and provides a new insight into the pathogenesis of this condition.

Coagulation factor XIII-A subunit missense mutation in the pathobiology of familial dermatofibroma

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Background: Familial dermatofibroma (DDF) is a rare inherited disease characterized by the occurrence of multiple dermatofibromas. Whole exome sequencing revealed a rare shared mutation in the F13A1 gene.

Methods: We performed whole exome sequencing on a family with DDF and identified a novel rare missense variant in the F13A1 gene (c.202A>C, p.T68P).

Results: The variant was found in two unrelated pedigrees with autosomal dominant transmission of multiple dermatofibromas. Whole exome sequencing revealed a rare shared heterozygous missense variant (c.202A>C, p.T68P).

Conclusion: This study highlights the importance of whole exome sequencing in identifying novel rare variants associated with familial dermatofibroma.

Precision medicine: Exome sequencing adds complexity to genotype/phenotype correlation

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Background: Xeroderma pigmentosum (XP) is a rare autosomal recessive DNA repair disorder caused by defects in DNA repair genes.

Methods: We performed whole exome sequencing on a family with XP-C and identified a novel heterozygous mutation in the XPC gene.

Results: The mutation (c.630G>A, p.G192D) was found in two unrelated individuals with different clinical manifestations.

Conclusion: This study highlights the complexity of genotype/phenotype correlation in XP and the importance of precision medicine in identifying novel rare variants.

UV-endonuclease and photolyase DNA repair enzymes increase cytosine gene expression after UVB induced downregulation

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Background: The purpose of this study is to determine gene expression changes induced by UVB and assess the effect of topical UV endonuclease and photolyase in recovery from these changes.

Methods: We performed microarray analysis on skin biopsies from mice before and after UVB exposure.

Results: We identified a novel UVB-induced downregulation of gene expression in the skin, with significant changes in DNA repair genes and cytosine metabolism.

Conclusion: This study highlights the importance of DNA repair enzymes in gene expression regulation and the potential for topical UV endonuclease and photolyase therapy in skin cancer prevention.