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A Novel Heterozygous Missense Mutation in *COL7A1* Gene in Dystrophic Epidermolysis Bullosa

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ABSTRACT Hereditary Epidermolysis Bullosa is categorized by blister formation on the skin which includes Dystrophic Epidermolysis Bullosa as one of its types. It is characterized by blister formation on hands, feet, knees, and elbows. The severe condition leads to vision loss and blemishes. The objectives included the identification of *COL7A1* gene mutation for DEB susceptibility through sequence analysis of hot spot regions of 73, 74 and 75 exons. The experimental design included the enrolment of DEB-affected two families. The genetic analysis techniques included inorganic DNA extraction, Polymerase Chain Reaction and Sanger sequencing method. The study inferred the identification of novel missense mutation in exon 75 of *COL7A1* gene at (c. 6223 G>T) where the aspartic acid is converted into tyrosine in the heterozygous condition in affected families. The recognized novel missense mutation is silent (heterozygous) which becomes severe when homozygous. However, whole-exome sequencing strategy may identify the causative mutations.