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A novel *COL7A1* mutation in a Korean patient with Hallopeau-Siemens recessive dystrophic epidermolysis bullosa

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ABSTRACT. Dystrophic epidermolysis bullosa (DEB) is an inherited skin fragility disorder that presents various clinical manifestations. DEB is characterized by separation of sublamina densa tissue and abnormalities in the anchoring fibrils that result from mutations in *COL7A1* and subsequent defects in type VII collagen. A 16-month-old boy was diagnosed with Hallopeau-Siemens recessive DEB on the basis of typical skin lesions composed of multiple blisters with moderately healed erosions, scarring on trauma-exposed body sites, including hands and feet, pseudosyndactyly and flexion contractures of the toes, and severely dystrophic nails on the right hand. Genomic DNA from the patient and parents were subjected to direct sequencing for the *COL7A1* gene. Two heterozygous mutations were detected in the affected child; one novel mutation designated c.4232delC in exon 38 and a single-base

substitution (c.6573+1G>C) in intron 81. Deletion of a single cytosine at codon 1411 within exon 38 had produced a frameshift mutation that created a stop codon at codon 1427 (p.Pro1411Leufs*17). This intronic base substitution had led to aberrant splicing and a premature termination codon. This is a novel mutation of *COL7A1* associated with DEB in a Korean patient, adding to the range of *COL7A1* mutations related to DEB.

Key words: Hallopeau-Siemens recessive dystrophic epidermolysis bullosa; *COL7A1*; Mutation analysis

