Synopsis of Original Research Paper

Nail deformity and type VII collagen abnormality

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Nail cosmetics have recently made remarkable progress. However, the basic study related to the nail shape has not been revealed yet. The etiology of toenail deformity is not fully understood. Shimizu et al and we have previously reported that certain glycine substitutions in the type VII collagen gene (COL7A1) lead to toenail deformity, in 1999. We explored the possibility that other patients exist with toenail deformity but without blister formation and that this is inherited in an autosomal dominant manner.

The eleven pedigrees, which contain more than one dystrophic epidermolysis bullosa patient, were investigated from the clinical point of view and gene mutation. Additionally, five pedigrees of the patient who have familial toe nail deformity were genetically analyzed. Four out of eleven pedigrees had familial nail deformity. The members who had nail deformity within the four pedigrees had glycine substitution within the collagenous domain of COL7A1. The five familial nail deformity pedigrees without dystrophic epidermolysis bullosa familial nail dystrophy were also investigated for COL7A1 mutation. No mutation was detected within COL7A1 except MspI polymorphism was found within one pedigree. The multiple etiology has been suspected with in familial nail dystrophy.