

ATOPIC ECZEMA/DERMATITIS

FILAGGRIN-GENE MUTATION HAVE NO EFFECTS ON THE SEVERITY OF ATOPIC DERMATITIS.

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Background: Filaggrin is known to be degraded into natural moisturizing factor. Mutations of filaggrin-gene, which are culprit factors for ichtyosis vulgaris, have been considered to play important roles in the pathogenesis of atopic dermatitis (AD).

Objective: This study aimed to determine whether filaggrin-gene mutations have any role on the regional severity of AD.

Patients and Methods: Fifty four patients with AD were enrolled in the study. In the 54 patients with AD, 7 kinds of filaggrin loss of function mutation found in Japanese were detected by genotyping with Taqman probes or direct sequencing with gDNA obtained from peripheral blood. Amount of filaggrin protein was evaluated using ELISA from scales in the lesions. Severity of the lesion was evaluated on cubital and neck by objective Scoring of AD (o-SCORAD). Barrier function was determined by measuring trans-epidermal water loss (TEWL) and skin water content (SWC).

Results and Conclusion: Filaggrin-gene mutations were detected 8 patients out of the 54 patients. Amount of filaggrin protein in non-lesion skin was lower in the 8 patients with mutations than in the 46 patients without mutations. However, in the lesions there were no significant differences in amount of filaggrin protein, oSCORAD, TEWL and SWC between the patients with mutations and those without mutations. These results indicate minimum role of filaggrin-gene mutations in the skin inflammation of AD.





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