

## 〈シンポジウム I〉

### 『皮膚の機能を知る～皮膚科学研究の最新情報～』

## 皮膚バリア障害の最新情報

秋山 真志

### Update on Skin Barrier Defects

Masashi AKIYAMA

#### Abstract

Three major components of the stratum corneum barrier are intercellular lipid layers, cornified cell envelope and keratin-filaggrin degradation products. Research into the pathomechanisms of genetic keratinization disorders have dramatically advanced and led to the identification of several causative genes and molecules underlying the genetic defects. In most types of the genetic keratinization disorders, pathogenic mechanisms are associated with defects in skin barrier function. The causative molecules underlying ichthyosis subtypes include ABCA12 and filaggrin. Loss of ABCA12 function leads to a defective lipid barrier in the stratum corneum, resulting in the most severe type of keratinization disorders, harlequin ichthyosis and ABCA12 is a known keratinocyte lipid transporter associated with lipid transport in lamellar granules. Filaggrin is a key protein involved in skin barrier function. Mutations in the gene encoding filaggrin (*FLG*) have been identified as the cause of ichthyosis vulgaris and shown to be major predisposing factors for atopic eczema.

**Key words:** ABCA12, atopic dermatitis, filaggrin, ichthyosis, skin barrier.