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〈講　　演〉

第49回日本香粧品学会（2024）・特別講演 I

皮膚に現れる様々なパターンから機能と病態を考える

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Functional and Pathological Analyses of the Diverse Morphological Patterns Observed on the Skin

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Abstract

Genetic mosaicism refers to the phenomenon of a genetic change (postzygotic mutation) occurring in one cell during development, with its daughter cells distributed on a particular part of the body and manifesting some pathological conditions.

In cutaneous mosaicism, if the causative genetic change occurred prenatally, the resulting skin lesions manifest during childhood. One such example is an epidermal nevus. Conversely, if the identical genetic change occurs in adults, the consequence is seborrheic keratosis or acrochordon. Consequently, the characteristics, distribution of the resulting skin lesions and the disease names will differ depending on whether the genetic change occurred during development or aging.

The term "mosaicism" is typically used to describe diseases in which cells with genetic changes that occurred during development give rise to lesions. However, symptoms caused by the clonal expansion of cells with genetic changes that occurred after birth are also considered to be mosaicism. Therefore, it is essential to comprehensively re-examine the pathogenesis of the mosaic disorders and to redefine and classify the pathogenetic mechanisms not only in genetic manifestations but also in chronological disease development.

Key words:　genetic mosaicism, somatic pathogenic variant, seborrheic keratosis, acrochordon, epidermal nevus.