

10. ACEM-Pathsoc meeting Glasgow 5th July 2007

Ultrastructural characterization of ichthyoses and related keratinization disorders

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Normal keratinization or terminal differentiation of the skin consists of the expression of a series of gene-controlled structural proteins and enzymes interacting in a well-defined sequence. Inborn errors of keratinization present pronounced clinical and genetic heterogeneity. By systematic investigation of ultrastructural aberrations it was evident that heterogeneity is even much larger than expected from the clinical features and light microscopy alone. Based on the peculiar and often highly specific ultrastructural features various types of keratinization disorders can now be delineated as distinct nosological entities. Furthermore, the comparison of normal and specifically altered structures and organelles may also serve as a clue to the elucidation of the underlying pathophysiological and/or molecular events. Examples include ichthyosis vulgaris with abnormal keratohyaline; four distinct types of ichthyosis congenita/lamellar ichthyosis characterized by lipid droplets or polygonal clefts within the horny layer, aberrant lamellar bodies and deposition of membrane-like material respectively, epidermolytic hyperkeratosis with keratin clumping; tonotubular keratin in a type of palmoplantar keratosis, and incomplete keratinization in Netherton syndrome.

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