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# HEREDITARY ICHTHYOSIS. PATHOGENESIS AND POSSIBILITIES OF TREATMENT

A. Kansky, B. Podrumac and I. Prelog

## ABSTRACT

Introduction. The term *ichthyosis* includes a number of pathogenetically different conditions, clinically displaying a more or less similar appearance. The underlying metabolic or molecular-biologic mechanisms have been elucidated up to now only in some of these conditions.

Known pathogenetic mechanisms. In recessive x-linked ichthyosis (RXLI) the deficiency of the steroid sulfatase has been demonstrated in cultured fibroblasts, leukocytes and keratinocytes. In certain patients with lamellar ichthyosis (LI) a deficient transglutaminase 1 was detected, the defect was located to a 9.3 cM region of chromosome 14q. In ichthyotic scales of non-bullous ichthyosiform erythroderma (NBIE) an increase of n-alkanes was reported. In bullous ichthyosiform erythroderma (BIE) genetic defects in keratins 1 and 10, and in ichthyosis bullosa Siemens (IBS) in keratin 2e were detected.

Systemic treatment. Unfortunately, there is no specific treatment for various entities of ichthyosis. In severe forms of LI, NBIE and BIE retinoids as *etretinate*, *etretin* and *isotretinoin* have been applied with varied success, in all instances, however, symptoms recurred after the treatment was stopped. The same is true for corticoids. Certain antimetabolites were also used for suppressing the symptoms.

Topical treatment. This is at present still the most important modality. Various ointment bases including salicylates, urea, corticosteroids as well as other ingredients are commonly applied. During the last years an impaired barrier function in ichthyosis, and in the frequently associated atopic dermatitis, is being stressed. For this reason unsaturated fatty acids and cholesterol are incorporated into ointments.

General Management. As it is known that small children with the severe forms of ichthyosis are sensitive to exposure to lower temperatures, to infections and to inappropriate diet, an adequate general regimen has to be observed.

Conclusion. As intense research is going on and new therapeutic modalities can be expected during the coming years.

## KEY WORDS

*hereditary ichthyosis, classification, pathogenesis, systemic, topical treatment*

