



The Rare Ichthyoses

I. Sjögren-Larsson syndrome

This condition is characterised by three features: a) ichthyosis; b) neurological problems which are often severe and c) mild to moderate mental retardation. The neurological problems can cause weakness and partial paralysis in the legs, but often these children learn to walk unaided or with the help of crutches. It is inherited as autosomal recessive affecting one in four children, assuming both parents are carriers. It is caused by an abnormality in a chemical (fatty alcohols) in the skin and nerves. The gene has been found allowing pre-natal diagnosis early in pregnancy. Treatment includes regular skin care, moisturising creams, bath oil, physiotherapy, as well as support from a number of specialists - dermatologist, neurologist and orthopaedic surgeon. Acitretin or neotigason is a retinoid drug taken by mouth which is related to vitamin A and can be helpful for this condition. It needs careful monitoring under hospital supervision. Orthopaedic surgery, such as the release of tightened tendons, is beneficial.

II. Conradi-Hünemann syndrome

In this condition ichthyosis is associated with bone abnormalities and cataracts. X rays of the skeleton in babies shows tiny spots at the growing end of the long bones called stippling, which is characteristic for this condition. The ichthyosis or scaling of the skin is often in lines or swirled patterns. It is inherited as a disorder of the X-chromosome and seen in girls. Treatment is along the same lines as the other more severe ichthyoses.

III. Keratitis-ichthyosis-deafness (or KID) syndrome

This combination is rare and most cases are sporadic with no family history. The skin changes develop in infancy with spiny thickening of



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