Ichthyosis Hystrix (Lambert Type)

Synonyms: ichthyosis hystrix gravior, epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosiform erythroderma, Lambert type ichthyosis

The name ichthyosis hystrix comes from the Greek words ichthyosis meaning scales like a fish and hystrix meaning like a porcupine. The condition was first described in the Lambert family in England in the early 18th century.

The designation of epidermolytic hyperkeratosis is used for the condition that is also called bullous congenital ichthyosiform erythroderma when generalised and ichthyosis hystrix when localised. They are presumably separate entities. Most types of ichthyosis are hereditary rather than acquired.

Epidemiology

This is a rare condition of autosomal dominant inheritance, although sporadic cases do occur. It was once thought to be a rare example of a Y-linked condition but this has been disproved and there is an equal sex incidence. The degree of penetrance of the condition is variable within families.

Presentation

It is apparent at birth or in the neonatal period as erythema with blistering affecting either a localised area of skin, or it can be more generalised. Blisters then heal with apparently normal skin but can recur. However, skin gradually becomes hyperkeratotic and scaly, particularly in flexures. Scales are hard and verruciform with parallel ridges running over the flexures, neck and hips. There is often a characteristic smell of rancid butter.

The whole body is affected, with the exception of the face, genitals, palms and soles.

Differential diagnosis

There are about 25 different forms of ichthyosis and obviously the generalist will not be expected to recognise each. Differing features include age at onset, the appearance of the skin, areas that may be affected or spared including flexures, palms, soles and face. The family history may also be indicative. Skin biopsy may be helpful. There may be features that are not dermatological that point to a specific syndrome.

The most common form of ichthyosis is ichthyosis vulgaris.

Investigations

Biopsy and electron microscopy may be useful in the early diagnosis of genetic disorders. As a general rule, dominant conditions tend to have defects of structural proteins whilst in recessive disorders there are quantitative impairments of enzymes and this may help differentiate similar clinical pictures of different origin.[2]

Management
This is a chronic disorder that requires continuous therapy. The main approach to treatment of ichthyosis includes hydration of the skin and application of an ointment to prevent evaporation. Hydration promotes desquamation by increasing hydrolytic enzyme activity and the susceptibility to mechanical forces. Pliability of the stratum corneum is also improved.

- Lactic, glycolic and pyruvic acids are effective for hydrating the skin. Lactic acid is available as a 5% proprietary preparation. Twice-daily applications have been shown to be superior to petroleum-based creams for control of ichthyosis.
- Removal of scales can be aided by keratolytics such as salicylic acid.
- Proprietary products often contain urea or propylene glycol. Moisturisers containing urea in lower strengths such as 10 or 20%, produce a more pliable stratum corneum by their hydratant action.
- Propylene glycol draws water through the stratum corneum by establishing a water gradient. Thick skin is then shed following hydration.
- Topical retinoids, usually tretinoin, may be beneficial. A good result with etretinate is reported. [3]
- Ichthyosis does not respond to steroids, but a mild topical steroid may be useful for pruritus.

Complications

This is a dermatological condition that affects all ages from birth. Therefore it is a visible and unsightly condition that affects children and adolescents too. Hence they are likely to be cruelly teased and will lack confidence in amorous encounters. Avid attention to management of the skin will minimise the problem.

It is a rare disease and the literature is very limited. There is remarkably little about prognosis. There are a number of case reports of complications such as squamous cell carcinoma. There may also be associated seizures, mental deficiency, eye problems, bone malformations and atrophy of the brain. This is not mentioned by all authors and is presumably a variable manifestation. Furthermore, the condition tends to run in families and such disastrous complications would not seem conducive to reproduction.

Prevention

There does not appear to be any prenatal test that can be employed. As it is a dominant condition, presumably one parent is affected. The literature seems to make no reference to limited penetrance. There will be a 1 in 2 chance of any children being affected.

Further reading & references

- Epidermolytic Hyperkeratosis (EHK), Foundation for Ichthyosis and Related Skin Types (FIRST) - Website; including images
- Foundation for ichthyosis and related skin types; Learning resource

1. Bullous Congenital Ichthyosiform Erythroderma (Epidermolytic Hyperkeratosis), Online Mendelian Inheritance in Man (OMIM)

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