Ichthyosis

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Synonyms of Ichthyosis

- Disorders of Cornification

Subdivisions of Ichthyosis

- Chanarin-Dorfman syndrome (neutral lipid storage disease)
- CHILD syndrome (unilateral hemidysplasia)
- congenital ichthyosiform erythroderma (CIE)
- Conradi-Hunermann syndrome (X-linked dominant chondrodysplasia punctata)
- Darier disease
- epidermal nevi (ichthyosis hystrix, linear epidermal nevus)
- epidermolytic hyperkeratosis (EHK)
- erythrokeratodermia variabilis (EKV)
- Giroux-Barbeau syndrome
- Hailey-Hailey disease (benign familial pemphigus)
- harlequin ichthyosis (harlequin fetus)
- ichthyosis hystrix Curth-Macklin type
- ichthyosis vulgaris (ichthyosis simplex)
- keratosis follicularis spinulosa decalvans
- KID syndrome (keratitis, ichthyosis, deafness)
- lamellar ichthyosis
- multiple sulfatase deficiency
- Netherton syndrome (ichthyosis linearis circumflexa)
- pachyonychia congenita
- palmoplantar keratodermas (PPK)
- peeling skin syndrome
- pityriasis rubra pilaris (PRP)
- Refsum's disease (phytanic acid storage disease)
- Rud's syndrome
- Sjogren-Larsson syndrome
- Tay's syndrome (trichothiodystrophy, IBIDS syndrome)
- X-linked ichthyosis

General Discussion

Ichthyosis is a general term for a family of rare genetic skin diseases characterized by dry, thickened, scaling skin. The various forms are distinguished from one another by: 1) extent of the scaling and how widely and where the scaling is scattered over the body; 2) the presence or

absence and intensity of reddening of the skin (erythroderma); 3) the mode of inheritance; and 4) the character of associated abnormalities.

Signs & Symptoms

Ichthyosis is characterized by scaly and dry skin usually over large areas of the body. The skin may also itch (pruritis) and be red (erythroderma). Babies born with some forms of the disorder may be born covered in a parchment-like membrane called a collodion membrane.

The appearance of the scales may vary; in some forms the scales may be fine and white, while in others the scales may be dark and brown and separated by deep cracks. The more severe forms of ichthyosis can cause other problems. When the skin loses moisture, it becomes dry, tight and inelastic. This rigidity can make moving uncomfortable and can cause the skin to crack and fissure. Thickening of the skin on the soles of the feet can make walking difficult and cracking around the fingers can make even simple tasks painful. In some types of ichthyosis the skin is very fragile and will rub off with the slightest abrasion. Cracks and abrasions then leave the skin open to infection.

Severe scaling on the scalp may interfere with normal hair growth. Thick scales can block pores, making sweating difficult and increasing the risk of overheating. Although the outer skin is thicker in ichthyosis, it is less effective in preventing water and calorie loss by diffusion across the surface of the skin. The rapid turnover of the outer layers of the skin, in some forms of ichthyosis, requires additional energy. Because of greater energy needs, some children with severe ichthyosis may require additional calories to grow normally.

Some people with ichthyosis have trouble closing their eyes completely because the surrounding skin is so tight. This condition, called ectropion, causes the eyelids to flip outward, exposing the red inner lid and causing irritation. If it is left untreated, damage to the cornea may develop leading to impaired vision.

Causes

Most known forms of ichthyosis are hereditary disorders. Some forms are caused by dominant genes; some are caused by recessive genes.

In some forms of ichthyosis, the skin cells are produced at the normal rate, but they do not separate normally at the surface of the outermost layer of skin (stratum corneum) and are not shed as quickly as they should be. In other forms, there is an overproduction of skin cells in the epidermis. The cells reach the stratum corneum in as few as four days, compared to the normal fourteen. New cells are made faster than the old cells are shed and build up in the stratum corneum and underlying layers. The result in both instances is a build up of scale.

Affected Populations

All the ichthyoses are rare disorders. Incidences vary according to disease type. The ichthyoses occur in all populations. Most forms are not bounded by gender, race or ethnicity.

Related Disorders

Ichthyosis congenita (collodion baby; congenital ichthyosiform erythroderma; xeroderma; desquamation of the newborn) is an inherited skin disorder. It is characterized by generalized,

abnormally red, dry, and rough skin with large coarse and fine white scales. Itchiness (pruritus) usually also develops. Skin on the palms of the hands and soles of the feet can be abnormally thick. (For more information, choose “Ichthyosis Congenita” as your search term in the Rare Disease Database.)

X-linked ichthyosis is an inherited skin disorder that affects males. It is caused by a deficiency of the enzyme steroid sulfatase. It is characterized by brownish scales on the back of the neck, back and legs.

Standard Therapies

The dry skin of ichthyosis is treating by applying skin softening emollients. This can be particularly effective after bathing while the skin is still moist. Lotions containing alpha-hydroxy acids, urea, or propylene glycol can also be effective. Skin barrier repair formulas containing ceramides or cholesterol may also improve scaling.

Severe cases of ichthyosis may be treated systemically with oral synthetic retinoids (synthetic derivatives of Vitamin A). Retinoids are used only in severe cases due to their known bone toxicity and other complications.

Investigational Therapies

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving U.S. government funding, and some supported by private industry, are posted on this government website.

For information about clinical trials being conducted at the National Institutes of Health (NIH) Clinical Center in Bethesda, MD, contact the NIH Patient Recruitment Office:

Tollfree: (800) 411-1222

TTY: (866) 411-1010

Email: prpl@cc.nih.gov

For information about clinical trials sponsored by private sources, contact:

www.centerwatch.com