Nonbullous Congenital Ichthyosiform Erythroderma

Subjects: Genetics & Heredity

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Nonbullous congenital ichthyosiform erythroderma (NBCIE) is a condition that mainly affects the skin.

genetic conditions

1. Introduction

Many infants with this condition are born with a tight, clear sheath covering their skin called a collodion membrane. Constriction by the membrane may cause the lips and eyelids to be turned out so the inner surface is exposed. The collodion membrane is usually shed during the first few weeks of life. Following shedding of the collodion membrane, the skin is red (erythroderma) and covered with fine, white scales (ichthyosis). Infants with NBCIE may develop infections, an excessive loss of fluids (dehydration), and respiratory problems early in life.

Some people with NBCIE have thickening of the skin on the palms of the hands and soles of the feet (palmoplantar keratoderma), decreased or absent sweating (anhidrosis), and abnormal nails (nail dystrophy). In severe cases, there is an absence of hair growth (alopecia) in certain areas, often affecting the scalp and eyebrows.

In individuals with NBCIE, some of the skin problems may improve by adulthood. Life expectancy is normal in people with NBCIE.

2. Frequency

NBCIE is estimated to affect 1 in 200,000 to 300,000 individuals in the United States. This condition is more common in Norway, where an estimated 1 in 90,000 people are affected.

3. Causes

Mutations in several genes can cause NBCIE. Mutations in the *ABCA12*, *ALOX12B*, or *ALOXE3* gene are responsible for most of cases of NBCIE. Mutations in other genes are each found in only a small percentage of cases. All of the genes associated with NBCIE provide instructions for making proteins that are found in the outermost layer of the skin (the epidermis). The epidermis forms a protective barrier between the body and its surrounding environment. Gene mutations impair the respective protein's function or structure within the epidermis, which prevents this outermost layer of skin from being an effective barrier before and after birth. The abnormal skin

cannot protect against fluid loss (dehydration) or the outside environment, leading to problems controlling body temperature; dry skin; the formation of fine, white scales; and increased risk of infections in people with NBCIE. The skin scales can impair the function of sweat glands under the skin, causing anhidrosis.

In some people with NBCIE, the cause of the disorder is unknown. Researchers are looking for additional genes that are associated with NBCIE.

3.1. The Genes Associated with Nonbullous Congenital Ichthyosiform Erythroderma

- ABCA12
- ALOX12B
- ALOXE3

3.1.1. Additional Information from NCBI Gene

- CASP14
- CERS3
- CYP4F22
- NIPAL4
- PNPLA1

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- congenital ichthyosiform erythroderma
- congenital nonbullous ichthyosiform erythroderma
- NBCIE
- NBIE
- NCIE
- nonbullous ichthyosiform erythroderma

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