Ichthyosis Vulgaris

Alternative Names
Ichthyosis Simplex

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities> Other congenital malformations.

Incidence per 100,000 Live Births
0-1

OMIM Number
146700

Mode of Inheritance
Autosomal dominant

Gene Map Locus
1q21.3

Description

Hereditary ichthyosis vulgaris is an autosomal dominant genetic disorder, member of a group of cutaneous disorders of keratinization that appear similar, both clinically and histologically. It is the most common form of ichthyosis, accounting for more than 95% of cases. The skin in hereditary ichthyosis vulgaris looks and feels normal at birth, but gradually becomes rough and dry in early childhood. Symptoms may include a dry and scaly skin, presence of small tile-like scales, flaky scalp, and deep painful cracks in the skin. Most cases of ichthyosis vulgaris are mild, but some cases are severe and can cover large areas of the body, including the abdomen, back, arms, and legs.

There is no cure for ichthyosis, but moisturizing and exfoliating the skin on a daily basis keeps the symptoms mild and manageable.

Molecular Genetics

Mutations in the filaggrin (FLG) gene have been identified as the cause of ichthyosis vulgaris and was shown to be the major predisposing factorfor atopic dermatitis. The encoded protein is important in functionally maintaining effective skin barrier. Two common FLG null mutations (p.R501X and c.2282del4) were identified in patients with ichthyosis vulgaris and they predispose to eczema and secondary allergic diseases. Patients carrying homozygous and compound heterozygous mutations may be severely affected, whereas heterozygotes showed mild disease or were asymptomatic, suggesting semidominant inheritance with incomplete penetrance in heterozygotes.

Epidemiology in the Arab World

Saudi Arabia

Al-Zayir and Al-Amro (2006) conducted a study between January 1990 and December 1995 to document the clinical and epidemiological features of patients with primary hereditary ichthyosis (PHI) in Saudi Arabia. Out of 10455 dermatology patients; a total of 71 patients (44 males and 27 females) were diagnosed with PHI. Consanguinity among the parents of the patients was significantly high (85%); parents of 60 of the 71 patients were married to either first-degree relatives or to second-degree relatives. Fifty-three of the 71 patients had positive family history of PHI. Ichthyosis Vulgaris (IV) was the most common type among the Saudi patients accounting for 44.7%. of the cases. All patients with IV presented after the third month of life.

References

Al-Zayir AA, Al-Amro Al-Alakloby OM. Clinico-epidemiological features of primary hereditary ichthyoses in the Eastern province of Saudi Arabia. Int J Dermatol. 2006; 45(3):257-64. PMID: 16533225

Related CTGA Records
Epidermolytic Hyperkeratosis

Ichthyosiform Erythroderma, Congenital, Nonbullous, 1
Ichthyosis, Lamellar, Autosomal Dominant
Ichthyosis, X-Linked

External Links

http://emedicine.medscape.com/article/1112753-overview
http://www.mayoclinic.org/diseases-conditions/ichthyosis-vulgaris/basics/definition/con-20024401
https://www.nlm.nih.gov/medlineplus/ency/article/001451.htm

Contributors

Nada Assaf: 26.6.2015