Ichthyosis, Congenital, Autosomal Recessive 4A

Alternative Name
ARCI4A
Ichthyosis Congenita IIB
ICR2B
Ichthyosis, Lamellar, 2
LI2

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
601277

Mode of Inheritance
Autosomal recessive

Gene Map Locus
2q35

Description
Congenital ichthyosis is an autosomal recessive disorder of variable severity. Affected patients are born with a collodion membrane which is shed one or two weeks after birth, making apparent the presence of scales all over the body. Patients may have palmoplantar keratoderma. Affected babies are often born prematurely. Prevalence is estimated approximately at 1/100,000-1/1,000,000 individuals. Congenital Ichthyosis is a genetically heterogeneous disease, and several subtypes of the condition have been identified based on defects in different genes. ARCI4A is a rare subtype that is caused due to defects in lipid transport in skin cells.

Diagnosis of the condition is through clinical examination and molecular genetic testing. Management is based on daily use of emollients or keratolytics. Oral retinoids are useful in severe forms of the disease. Affected neonates rarely survive infancy, and those who do develop severe non-bullous congenital ichthyosiform erythroderma.

Molecular Genetics
Mutations in the ABCA12 gene are responsible for causing ARCI4A. This gene encodes an ATP-binding cassette (ABC) transporter, which is known to be involved in active lipid transport, especially in skin cells. Studies on animal models have suggested that the protein plays a critical role in the formation of the skin's permeability barrier via an effect on the generation of a highly specialized class of ceramide esters.

Epidemiology in the Arab World

Saudi Arabia

Wakil et al., (2016) described two unrelated Saudi patients with ARCI born to consanguineous parents. Both patients had thick adherent polygonal large scales all over the body and erythema. The authors identified one novel mutation (p.Phe2300Leu) and one reported mutation (p.Ser1157Leu) in the ABCA12 gene in the two families. Combined approach of homozygosity mapping and direct sequencing analysis was helpful in confirming the diagnosis of these patients.

References
Wakil SM, Binamer Y, Al-Dossari H, Al-Humaidy R, Thuraya RA, Khalifa O, Finsterer J, Meyer BF, Al Owain M. Novel mutations in TGM1 and ABCA12 cause autosomal recessive congenital ichthyosis in five Saudi families. Int J Dermatol. 2016; 55(6):673-9. PMID: 27061915.

Related CTGA Records
ATP-Binding Cassette, Subfamily A, Member 12

External Links
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=313

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