Lamellar Ichthyosis 5

**Alternative Names**
L15

**WHO International Classification of Diseases**
Congenital malformations, deformations and chromosomal abnormalities

**OMIM Number**
606545

**Mode of Inheritance**
Autosomal Recessive

**Gene Map Locus**
17p13.2-p13.1

**Description**
Congenital ichthyosis is a clinically and genetically heterogeneous group of disorders of keratinization characterized by a significant and incapacitating scaling of the skin. Most forms are congenital and display different modes of inheritance. Among them, autosomal recessive lamellar ichthyosis is a severe condition with an estimated prevalence of 1 per 200,000 newborns, presenting a 'collodion-baby' syndrome at birth: the newborn is embedded in a hyperkeratotic skin. Spontaneous shedding of the membrane leaves residual ichthyosis and may lead to grave sequelae, such as joint contractures, ectropion and secondary corneal involvement. Sometimes during adulthood, scarring alopecia, palmoplantar keratoderma and associated erythroderma are observed, and are therefore features of phenotypic variability.

**Molecular Genetics**
By homozygosity mapping, the localization of the gene causing lamellar ichthyosis 5 could be identified to a locus on chromosome 17p. This interval contains about 20 known genes, a number of which have been shown to be expressed in the skin.

**Epidemiology in the Arab World**

**United Arab Emirates**
Krebsova et al. (2001) analyzed two consanguineous families from the United Arab Emirates with congenital ichthyosis. The first family had two affected siblings. The older affected boy was born as a collodion baby. At four weeks of age, he was still erythrodermic and presented with diffuse scaling, which involved also the great folds, and palmoplantar keratoderma. Ichthyosis increased with age and was more marked on the extensor surfaces of the limbs and on the upper back. The younger affected brother was also born as a collodion baby. He showed generalized erythroderma and palmoplantar keratoderma. Ichthyosis also increased with age, involving mostly the extensor surfaces of the limbs and the upper back, but remained mild, with a tendency to erythroderma, and spared the face. In the second affected family, the affected girl presented at three months of age with skin scaling on the limbs, the trunk, and the neck. Lesions increased with age. At the age of 13 years, ichthyosis consisted of thick, dark brown or black adhering scales. Krebsova et al. (2001) performed whole-genome scan in both families. Interestingly, patients from both families were heterozygous at each marker locus analyzed on chromosome 17p. Krebsova et al. (2001) assumed that congenital ichthyosis in these families was not linked to the region on chromosome 17p. In addition, candidate loci on chromosomes 2, 3, 14, and 19 were also excluded indicating that an unknown locus might be involved.

**References**
Krebsova A, Kuster W, Lestringant GG, Schulze B, Hinz B, Frossard PM, Reis A, Hennies HC. Identification, by homozygosity mapping, of a novel locus for autosomal recessive congenital ichthyosis on chromosome 17p, and evidence for further

Contributors
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