

Opis choroby *

Definicja

Ichthyosis-hypotrichosis syndrome is characterised by congenital ichthyosis and hypotrichosis. It has been described in three members of a consanguineous Arab Israeli family. The syndrome is transmitted as an autosomal recessive trait and is caused by a missense mutation in the *ST14* gene, encoding the recently identified protease, matriptase. Analysis of skin samples from the patients suggests that this enzyme plays a role in epidermal desquamation.

Dane

Klasyfikacja

Choroba

Synonimy

Hypotrichosis-congenital ichthyosis syndrome

IHS

Zespół hipotrichozy i rybiej łuski wrodzonej

Zespół IFAH

Zespół rybiej łuski, atrofodermii mieszkowej i hipotrichozy

Zespół rybiej łuski, atrofodermii mieszkowej, hipotrichozy i hipohidrozy

IFAH syndrome

IHS

Ichthyosis-follicular atrophoderma-hypotrichosis syndrome

Ichthyosis-follicular atrophoderma-hypotrichosis-hypohidrosis syndrome

Kod ORPHA

91132

Kod OMIM

602400

Kod ICD10

Q80.8

Kod ICD11

LD27.2

[*Źródło](#)

orphanet