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 hipohydrozy
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

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*Źródło