

## Opis choroby \*

### Definicja

A congenital genodermatosis with skin/mucosae involvement, characterized by very tight and thin skin with erosions and scaling, associated to a typical facial dysmorphism, arthrogryposis multiplex, fetal akinesia or hypokinesia deformation sequence (FADS) and pulmonary hypoplasia without neurological abnormalities.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Lethal hyperkeratosis-contracture syndrome  
Tight skin contracture syndrome  
Letalna dermatopatia restrykcyjna  
Zespół hiperkeratozy i przykurczy  
Lethal restrictive dermatopathy  
Lethal tight skin-contracture syndrome

#### Kod ORPHA

1662

#### Kod OMIM

619793

#### Kod ICD10

Q82.8

#### Kod ICD11

EE6Y

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#### \*Źródło

orphanet