

Opis choroby *

Definicja

Ichthyosis-alopecia-eclabion-ectropion-intellectual disability syndrome is an ectodermal dysplasia syndrome characterized by severe generalized lamellar ichthyosis at birth with alopecia, eclabium, ectropion and intellectual disability. Although similar to Sjögren-Larsson syndrome, this syndrome lacks the presence of neurologic or macular changes. There have been no further descriptions in the literature since 1987.

Dane

Klasyfikacja	Synonimy
Choroba	Jagell-Holmgren-Hofer syndrome Zespół Jagella, Holmgrena i Hofera

Kod ORPHA	Kod OMIM	Kod ICD10
2269	242510	Q82.4

Kod ICD11
LD27.0Y

*Źródło

orphanet