

Opis choroby *

Definicja

A rare, syndromic ichthyosis characterized by a collodion membrane at birth, generalized congenital ichthyosis, microspherophakia, myopia, ectopia lentis, short stature with brachydactyly and joint stiffness, and occasionally mitral valve dysplasia.

Dane

Klasyfikacja

Choroba

Synonimy

15q26.3 microdeletion syndrome

Zespół mikrodelecji 15q26.3

Kod ORPHA

363992

Kod OMIM

613195

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet