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