Opis choroby *

Definicja

A rare, genetic, chromosomal anomaly syndrome resulting from partial duplication of the long arm of chromosome 2 characterized by congenital pendular nystagmus associated with bilateral cutaneous syndactyly between the third and fourth fingers.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Syndactyly-nystagmus syndrome due to dup(2)(q31.1)
Dup(2)(q31.1)
Trisomia 2q31.1
Syndactyly-nystagmus syndrome due to trisomy 2q31.1

Kod ORPHA
294026

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
-

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*Źródło

orphanet