

Wrodzone zwłóknienie mięśni zewnątrzgałkowych

Kod Orpha: 45358 Kod OMIM: 609612

Opis choroby *

Definicja

A rare syndromic disorder with strabismus characterized by congenital non-progressive ophthalmoplegia affecting the oculomotor and/or trochlear nucleus/nerve and their innervated muscles. Patients present with abnormal resting position of the eyes (in most cases infraducted and exotropic), limitation of vertical and horizontal gaze, impaired binocular vision, amblyopia, unilateral or bilateral blepharoptosis, and compensatory abnormal head posture. Extraocular manifestations include intellectual disability, peripheral neuropathy, and skeletal abnormalities, among others.

Dane

Klasyfikacja

Choroba

Synonimy

FEOM

FEOM

Kod ORPHA

45358

Kod OMIM

609612

Kod ICD10

H49.8

Kod ICD11

9C82.2

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.