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

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*Źródło

orphanet