Rodzinne wrodzone porażenie nerwu bloczkowego Kod Orpha: 91498 Kod OMIM: 136480

Opis choroby *

Definicja
Familial congenital palsy of trochlear nerve is a rare, genetic, neuro-ophthalmological disease characterized by congenital fourth cranial nerve palsy, manifesting with hypertropia in side gaze, unexplained head tilt, acquired vertical diplopia, and progressive increase in vertical fusional vergence amplitudes with prolonged occlusion. Facial asymmetry (i.e. hemifacial retrusion, upward slanting of mouth on the side of the head tilt, mild enophthalmos of paretic eye) and superior oblique tendon abnormalities (such as absence, redundance, misdirection) are frequently associated. Some asymptomatic cases have been reported.
Dane

Klasyfikacja
Choroba

Kod ORPHA
91498

Kod OMIM
136480

Kod ICD10
H49.1

Kod ICD11
9C81.1

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.