Opis choroby *

Definicja

A rare chromosomal anomaly syndrome, resulting from the partial deletion of the short arm of chromosome X, principally characterized by classical Norrie disease (bilateral, severe retinal malformations and opacity of the lens leading to congenital blindness, on occasion associated with progressive sensorineural deafness and intellectual disability), microcephaly, hypotonia, psychomotor and growth delay, moderate to severe mental handicap and disruptive behaviour. Clinical phenotype is highly variable and immunodeficiency, epilepsy and hypogonadism have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Atypical Norrie disease due to del(X)(p11.3)
Atypowa choroba Norriego z powodu del(X)(p11.3)
Atypowa choroba Norriego z powodu mikroduplikacji Xp11.3
Atypical Norrie disease due to nullisomy Xp11.3

Kod ORPHA
261501

Kod OMIM
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Kod ICD10
H35.5

Kod ICD11
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*Źródło

orphanet