

Opis choroby *

Definicja

CK syndrome is a rare, genetic, X-linked syndromic intellectual disability disorder characterized by mild to severe intellectual disability, infancy-onset seizures, post-natal microcephaly, cerebral cortical malformations, dysmorphic facial features (including long, narrow face, almond-shaped palpebral fissures, epicanthic folds, high nasal bridge, malar flattening, posteriorly rotated ears, high arched palate, crowded teeth, micrognathia) and thin body habitus. Long and slim fingers/toes, strabismus, hypotonia, spasticity, optic disc atrophy, and behavioral problems (aggression, attention deficit hyperactivity disorder and irritability) are additional features.

Dane

Klasyfikacja

Zespół wad wrodzonych X-linked intellectual disability-microcephaly-cortical malformation-thin habitus syndrome
Niepełnosprawność intelektualna sprzężona z chromosomem X - małowłowie - malformacja kory mózgu - szczupły wygląd

Synonimy

Kod ORPHA

251383

Kod OMIM

300831

Kod ICD10

Q87.0

Kod ICD11

-

[*Źródło](#)

orphanet