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

Synonimy
X-linked intellectual disability-microcephaly-cortical malformation-thin habitus syndrome
Niepełnosprawność intelektualna sprzężona z chromosomem X - małogłowie - malformacja kory mózgu - szczupły wygląd

Kod ORPHA
251383

Kod OMIM
300831

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet