Opis choroby *

Definicja

A rare partial duplication of the long arm of chromosome 17 characterized by a combination of features of 17p11.2 microduplication syndrome and Charcot-Marie-Tooth disease type 1A. Patients present with infantile onset of global developmental delay, hypotonia, feeding difficulties, and failure to thrive, as well as childhood onset of peripheral neuropathy with distal extremity weakness or atrophy, gait impairment, sensory loss, reduced or absent deep tendon reflexes of the ankles, and foot deformities. Facial dysmorphism, cardiac and renal anomalies, and syringomyelia may also be observed.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
17p11.2p12 microduplication syndrome
Dup(17)(p11.2p12)
Trisomia 17p11.2p12
Trisomia 17p11.2-p12
Zespół mikroduplikacji 17p11.2p12
Zespół Yuana, Harela i Lupskiego
Dup(17)(p11.2p12)
Trisomy 17p11.2-p12
Trisomy 17p11.2p12
Yuan-Harel-Lupski syndrome

Kod ORPHA
477817

Kod OMIM
616652

Kod ICD10
Q92.3

Kod ICD11
-

---

*Źródło

orphanet