Opis choroby *

Definicja

A rare X-linked genomic disorder associated with interstitial chromosomal duplications at Xq28 encompassing the MECP2 gene. It is characterized in males by infantile onset hypotonia, severe global developmental delay, intellectual disability, progressive spasticity, seizures, gastrointestinal symptoms and recurrent respiratory infections. In females, the phenotype is more variable.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MECP2 duplication syndrome
Dystalna duplikacja Xq
Telomerowa duplikacja Xq
X-linked intellectual disability syndrome, Lubs type

Kod ORPHA
1762

Kod OMIM
300260

Kod ICD10
Q99.8

Kod ICD11
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*Źródło

orphanet