

Opis choroby *

Definicja

A rare X-linked genomic disorder associated with interstitial chromosomal duplications at Xq28 encompassing the <i>MECP2</i> 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 **Synonimy**

Zespół wad wrodzonych 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