

Trisomia 8p

Kod Orpha: 264450 Kod OMIM:

Opis choroby *

Definicja

Trisomy 8p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 8, with highly variable phenotype ranging from no dysmorphic features and only mild intellectual disability to patients with severe developmental delay, neonatal hypotonia, short stature, profound intellectual disability, mild dysmorphic features (e.g. mild ptosis, hypertelorism, down-slanting palpebral fissures, broad nasal bridge, short, prominent philtrum, abnormal dentition) and structural brain abnormalities. Autism, epilepsy, and spastic paraplegia have also been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Duplication 8p
Duplikacja 8p

Kod ORPHA

264450

Kod OMIM

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Kod ICD10

Q92.2

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl