

Opis choroby *

Definicja

Trisomy 5p is a chromosomal abnormality resulting from the duplication of a segment of variable size of the short arm of chromosome 5, which usually involves the distal band 5p15. The clinical presentation is variable but is always associated with severe intellectual deficit.

Dane

Klasyfikacja

Zespół wad wrodzonych Duplication 5p

Duplikacja 5p

Trisomia krótkiego ramienia chromosomu 5

Duplication of the short arm of chromosome 5

Trisomy of the short arm of chromosome 5

Kod ORPHA

1742

Kod OMIM

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Kod ICD10

Q92.2

Kod ICD11

LD41.41

*Źródło

orphanet