## Opis choroby *

Definicja

4p16.3 microduplication syndrome is a rare genetic syndrome that results from the partial duplication of the short arm of chromosome 4. It has a highly variable phenotype, principally characterized by psychomotor and language delay, seizures and dysmorphic features such as high forehead with frontal bossing, hypertelorism, prominent glabella, long narrow palpebral fissures, low set ears and short neck. Eye abnormalities (glaucoma, irregular iris pigmentation, hyperopia) have also been reported.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Distal duplication 4p
Duplikacja dystalna 4p
Duplikacja telomerowa 4p
Trisomia 4pter
Trisomia dystalna 4p
Distal trisomy 4p
Telomeric duplication 4p
Trisomy 4pter

**Kod ORPHA**
96072

**Kod OMIM**
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**Kod ICD10**
Q92.3

**Kod ICD11**
LD41.31

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*Źródło

orphanet