Opis choroby *

Definicja

A partial deletion of the long arm of chromosome 4 characterized by complex behavioral difficulties, developmental and delay/ intellectual disability, and minor dysmorphic features, including subtle facial asymmetry (most prominent in the mandible), mild hypotelorism, long nasal bridge, small low-set ears, narrow mouth, and mild hand deformities, such as bilateral short 5th metacarpals, and short hands.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Proximal del(4)(q25)
del(4)(q25) proksymalna
Monosomia proksymalna 4q25
Proximal monosomy 4q25

Kod ORPHA
502437

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet