Opis choroby *

Definicja

A rare partial deletion of the long arm of chromosome 6 characterized by a variable clinical phenotype that includes a characteristic craniofacial dysmorphism (including microcephaly, broad nose with prominent nasal root and bulbous nasal tip, large ears that may be malformed and low-set, characteristic downturned mouth, and short neck), global development delay, intellectual disability, and variable, non-specific, congenital malformations. Muscular hypotonia, seizures, retinal anomalies, and variable brain abnormalities have been reported in association.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
75857

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
LD44.60

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*Źródło

orphanet