

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

*Źródło

orphanet