Opis choroby *

Definicja

A partial autosomal trisomy characterized by developmental delay and intellectual disability, generalized hypotonia, postnatal growth retardation, variable brain and heart anomalies and dysmorphic features, including frontal bossing, round face, full cheeks, low-set ears, broad nasal bridge, short nose with anteverted nares, long philtrum, thin upper lip vermilion, and everted, thick lower lip. Unspecific associated congenital anomalies have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Duplication 12p
Duplikacja 12p

Kod ORPHA
1699

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.B1

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*Źródło

orphanet