Opis choroby *

Definicja

A partial autosomal trisomy characterized by developmental delay, intellectual disability, prenatal and postnatal growth retardation, congenital heart, genitourinary and skeletal anomalies, and dysmorphic facial features, including high and broad forehead, hypertelorism, upslanting palpebral fissures, broad nose, dysplastic and low set ears, micrognathia. Phenotypic features vary in relation to the duplication size.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Duplication 8q
Duplikacja 8q

Kod ORPHA
1752

Kod OMIM
-

Kod ICD10
Q92.2

Kod ICD11
LD41.70

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*Źródło

orphanet