

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
Duplikacja 8q

Synonimy

Duplication 8q
Duplikacja 8q

Kod ORPHA

1752

Kod OMIM

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Kod ICD10

Q92.2

Kod ICD11

LD41.70

*Źródło

orphanet