Opis choroby *

Definicja

A constitutional genomic disorder due to the presence of a supernumerary derivative 22 chromosome and characterized by severe intellectual disability, characteristic facial dysmorphism (micrognathia, hooded eyelids, upslanting downslanting parebral fissures, deep set eyes, low hanging columnella and long philtrum), congenital heart defects and kidney abnormalities.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Der(22)t(11;22) syndrome
Zespół nadliczbowego der(22)
Zespół Der(22)t(11;22)
Supernumerary der(22) syndrome

Kod ORPHA
96170

Kod OMIM
609029

Kod ICD10
Q92.6

Kod ICD11
LD41.Q

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*Źródło

orphanet