

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 Der(22)t(11;22) syndrome

Synonimy

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

*Źródło

orphanet