## 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