

Opis choroby *

Definicja

A rare partial autosomal trisomy/tetrasomy characterized by obesity, global developmental delay and intellectual disability, facial dysmorphism (synophrys, high-arched eyebrows, large posteriorly rotated ears, upturned nose, long smooth philtrum, overbite and high palate), large hands and limb hypotonia. Additional features include seizures and behavioral abnormalities.

Dane

Klasyfikacja

Zespół wad wrodzonych Dup(11)p(15.4)

Dup(11)p(15.4)

Trisomia 11p15.4

Trisomy 11p15.4

Kod ORPHA

300305

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

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*Źródło

orphanet