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

Synonimy
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