Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable developmental delay and intellectual disability, movement disorder or gait abnormalities, and dysmorphic craniofacial features (such as facial asymmetry, broad forehead, posteriorly rotated ears, thick lower lip, micrognathia, or cleft palate). A variety of congenital malformations have been reported in addition, including ocular, renal, cardiac, and joint anomalies, among others. Some patients show behavioral alterations (autism, hyperactivity, or anxiety).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
YY1 haploinsufficiency syndrome
Zespół haploinsuficjencji YY1

Kod ORPHA
506358

Kod OMIM
617557

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet