Opis choroby *

Definicja

A rare partial autosomal trisomy/tetrasomy characterized by global developmental delay, intellectual disability, autistic behavior, muscular hypotonia, macrocephaly and facial dysmorphism (frontal bossing, short palpebral fissures, low set, dysplastic ears, short or shallow philtrum, high arched or narrow palate, micrognathia). Other associated clinical features include sleep disturbances, seizures, aplasia/hypoplasia of the corpus callosum, skeletal abnormalities (large hands and feet, long fingers and toes, talipes).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(5)(p13)
Dup(5)(p13)
Trisomia 5p13
Trisomy 5p13

Kod ORPHA
329802

Kod OMIM
613174

Kod ICD10
Q92.3

Kod ICD11
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*Źródło

orphanet